



# CHARGE SYNDROME

## *A MANAGEMENT MANUAL FOR PARENTS*

Version 2.1  
2002

*Edited by Meg Hefner, M.S. and  
Sandra L.H. Davenport, M.D.*

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- ♦ The Minnesota DeafBlind Project
- ♦ The Maryland Jaycees
- ♦ CHARGE Syndrome Foundation, Inc.

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**- Meg Hefner [meg@chargesyndrome.org](mailto:meg@chargesyndrome.org)**

### **Missing Medical Issues**

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# Welcome to Version 2.1 of the

## CHARGE Syndrome Management Manual for Parents

Meg Hefner, M.S.  
April, 2002

Welcome to version 2.1 of the CHARGE Syndrome Management Manual for Parents. Version 1.0 of the Manual (1999) included the *Medical* issues of CHARGE syndrome. These sections (see the Introduction to the Medical Section) should help you and the people working with your child to better understand the medical implications of a diagnosis of CHARGE.

Version 2.0 (2001) added information on the *Developmental* and *Educational* implications of CHARGE syndrome. It was not possible to include even all of the good information we have come across in this version.

Version 2.1 (2002) compiles versions 1.0 and 2.0 into a single format and updates the contact information for the contributors. Additions and updates to the Manual will be done periodically. Updates and additions may be distributed through the CHARGE Syndrome Foundation newsletter, *CHARGE Accounts*, (hint, hint – join the Foundation and get the newsletter). To be sure you get word of all of the updates:

- 1) **Resister your Manual** by mailing or faxing the completed registration page to:  
CHARGE Syndrome Foundation, Inc.  
409 Vandiver Dr Ste 5-104  
Columbia, MO 65202-1563  
  
Fax (573)499-4694
- 2) **Join** the Foundation (membership application can be found behind the registration page or at [www.chargesyndrome.org](http://www.chargesyndrome.org)) and receive the newsletter, *CHARGE Accounts*.

### How to compile your Manual:

Get a large (2" or greater), sturdy **3-ring binder** to house the Manual. The pages are hole-punched to fit a binder.

Get several sets of 3-ring **dividers with tabs** to divide the sections (Intro, Stories, Medical, Developmental, Glossary, Resources) and sub-sections. Dividers with a pocket can be helpful to keep track of loose papers. All pages are dated.

Many families find it helpful to keep their own medical records in the appropriate section of the Manual



Meg Hefner, Marion Norbury, and Sandra Davenport at Portland Conference, 1995



Marion Norbury, Jim Thelin and Sandra Davenport at Portland Conference, 1995

## CHARGE SYNDROME MANAGEMENT MANUAL REGISTRATION FORM

\*\*\*\* This is very important\*\*\*\*

There will be updates to this Manual from time to time. In order to receive notification of future updates, you must complete the bottom portion of this form and return it to the CHARGE Syndrome Foundation.

I purchased/received this Manual on \_\_\_\_ / \_\_\_\_ / \_\_\_\_

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## **HISTORY OF CHARGE SYNDROME FOUNDATION, INC. and THE MANAGEMENT MANUAL FOR PARENTS (2.1)**

The CHARGE Syndrome Foundation is incorporated in Columbia, Missouri where it has its roots. It began in 1982 with the Deaf-Blind Project in the Division of Genetics, Department of Child Health, University of Missouri, Columbia. The primary participants in the Deaf-Blind Project were Dr. Joyce Mitchell (Medical Informatics), Dr. Sandra Davenport (Genetics and Developmental Pediatrics), Dr. James Thelin (Audiology), and Meg Hefner (Genetics).

### *CHARGE Syndrome: a booklet for parents*

Although the stated aim of the Deaf-Blind Project had to do with diagnosis of CHARGE and description of the features for the benefit of geneticists and other physicians, we quickly recognized that families need information, too. Quota Club of Columbia, a local service organization for business and professional women, had a pot of money and was looking for a project to fund which had something to do with hearing loss. As a result of Jim Thelin's connection with Quota Club, Jim and Meg became the primary authors of *CHARGE Syndrome: A Booklet for Families*, with lots of input from Sandy and Joyce. Quota Club member Marion Norbury volunteered to help send out the booklets. More than 15 years later, Marion has become the Executive Director of the CHARGE Syndrome Foundation and Foundation materials have taken over her house.

In 1988 we printed the first 1,000 Booklets, figuring they would last five years or so (we thought CHARGE was rare!). The typical scenario was as follows: A family would write or call and request one booklet. A week or so later, the same family would request 5-10 booklets so they could be passed out to relatives, doctors, teachers, and others who worked with their child. The booklets were gone in a year, and went through several more printings in the following years. Until the Manual became available, the *Booklet for Families* was the best initial source of information on CHARGE available to families.

After publication and distribution of the booklet, we began to get calls and letters from families asking things like, "Are there any other children with CHARGE in New Jersey?" "Does anyone else have a baby with feeding problems?" "Is my child's heart defect a typical one for CHARGE?" and so on. It was clear we needed a way for families to get more information and to share their information and stories with one another as well as with us.

### *CHARGE Accounts* newsletter

In 1989, Marion, Sandra and Meg published the first edition of the newsletter - *CHARGE Accounts*. The first few years were a bit shaky with issues not always coming out on time, but it was a success with families. Marion has taken primary charge of the newsletter, with editorial assistance from Meg. It is now published four times a year and distributed to all members of the CHARGE Syndrome Foundation.

### International CHARGE Syndrome Conferences

In 1993, we incorporated as the CHARGE Syndrome Foundation, Inc. and held our first International CHARGE Syndrome Conference in St. Louis, Missouri. It was attended by over 250 people, including about 50 children with CHARGE. We continue holding CHARGE conferences every two years with rotating geographic locations.

As of 2001, conferences have been held in St. Louis, Portland (Oregon), Boston, Houston, and Indianapolis. Foundation membership is over 400 and we receive anywhere from 10-20 requests for information every week - by snail mail, e-mail and phone. Our quarterly newsletter *CHARGE Accounts* goes to more than a dozen countries besides the United States. A toll-free number (800-442-7604) and website ([www.chargesyndrome.org](http://www.chargesyndrome.org)) make it easy for families to contact the Foundation for information and support.

#### CHARGE Syndrome Foundation, Inc

The CHARGE Syndrome Foundation has a 12-member elected Board of Directors (primarily parents) who are responsible for publicizing CHARGE, developing additional materials for families and professionals, raising money for conferences and research, planning the conferences and determining the direction of the Foundation. The CHARGE Foundation Professional Advisory Board is made up of more than 30 professionals including a variety of medical and educational specialists, all with a specific interest in people with CHARGE and their families.

#### The Original Team

Although the original team has dispersed geographically, (Marion is still in Columbia, Missouri; Meg is in St. Louis; Jim is in Knoxville, Tennessee; and Sandra is in Bloomington, Minnesota), we remain interested and active in CHARGE (thanks in no small part to e-mail).

#### Listserv on the Internet

In 1997, Casey Fisher established a listserv for CHARGE. Since then, a very active group of families, friends and professionals have been sharing their problems, support and knowledge over the Internet through the listserv (see the CHARGE website for a link). Although not an official part of the Foundation, the list is recognized as one of the most important aspects of our being able to communicate with one another and support each other.

#### The Management Manual

Although the Booklet was really good information in 1988, it quickly became obsolete. Through the conferences, we learned more and more about CHARGE - the features involved, the complexity of the kids, the need of the families to learn as much as possible about CHARGE syndrome. At the Portland conference in 1995, we gathered information on about 50 children and young adults with CHARGE. This information was assimilated with the collective knowledge of the Professional Advisory Board and published in the *Journal of Pediatrics* in 1998. This information and lots more was then assembled and edited into version 1.0 (Medical Aspects of CHARGE) of the Manual, published in 1999. Families said that was great, but what about other aspects of CHARGE such as development, education and behavior? By 2001, we had assembled version 2.0, incorporating a tremendous amount of information about those aspects of CHARGE. We know we are not finished - this is a never-ending project. That's part of why it is in a binder format. Future updates to the Manual will appear in *CHARGE Accounts*. We welcome any feedback you have, as well as suggestions for future updates/additions to the Manual.



# CHARGE STORIES

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## COLOBOMA

by Tim Hartshorne

Jacob's retinal colobomas were diagnosed during his first year. The coloboma in his right eye involves the macula and accounts for significant visual loss. His right eye is also smaller than his left. The coloboma in his left eye involves only part of the macula, and he seems to receive useful vision in that eye. His acuity in both eyes is poor, especially his right eye. In addition, he has astigmatism in both eyes.

A recent, functional vision evaluation found that Jacob could detect objects in his lower, right and left visual fields, but not in his upper field. His responses to visual stimuli were consistent with poor depth perception.

Jacob wears corrective lenses. He received his first pair at under a year of age, and wore them very consistently for a few years before developing a habit of throwing them. After several years of consistent effort, his school staff managed to teach him to wear his glasses while at school. However, at home he prefers to not wear them. When he is wearing his glasses he appears to be more attentive to the world around him.

Jacob's eyes are checked twice a year. A major concern with retinal colobomas is retinal detachment. So far Jacob shows no detachment.



# HEART

by Jackie Kenley

Our daughter, Laura, was born in 1985 at the University of California San Francisco hospital. On her second day of life, a heart murmur was detected by the pediatric staff. The cardiology staff was then brought in for a consult and after an echocardiogram it was determined that Laura had a major heart defect common to children with CHARGE, tetralogy of Fallot. Laura had a good birth weight and we were advised that it would be optimal for Laura to grow and be older before the heart repair. As our family was dealing with the other problems our new little girl had (such as hearing and vision loss), we were grateful that the surgery could be put off. We took Laura home and cared for her while she gained weight. She also became a loving part of our family.

At thirteen months, the cardiology team felt Laura was ready for her repair. Her brother, then 6, and sister 2, were taken to the hospital. A member of the staff explained the procedure to them and showed them where Laura would be. It was a long procedure with Laura on a heart lung machine. After the first surgery, Laura had bleeding complications which only occur in a small percent of the patients. We knew at that time that Laura often fell into the "small percent" population. She had to go back on the heart lung machine while they found the area that needed suturing. Finally Laura was taken to ICU. After surgery, it was eight days before Laura could be extubated. She was kept "knocked out" with morphine during this time. It seemed an eternity for us, but finally Laura was able to come home for a quiet recovery.

Laura has had another surgery for mitral valve repair...thirteen years later. This time, Laura quickly extubated herself (much to the staff's surprise) in the ICU. Her nurses noted she was breathing fine on her own and did not seem to be in distress! She had gone into the hospital on Thursday and came home the following Tuesday! We have all witnessed an increase in learning and overall activity since this repair.

Laura's brother and sister seemed much more emotional and concerned by the second surgery. I know that they were very relieved when it was over. We do not foresee any further heart surgeries. However, with Laura, we know there is always the possibility of needing care for her heart. Our family is thankful daily for her "well-repaired heart"!

# ATRESIA OF THE CHOANAE

by Debbie Matasker

My son Michael was born in 1997. He had a very eventful birth. We knew there were problems at 33 weeks, because I had developed polyhydramnios. I had an emergency C-section because of fetal distress. The polyhydramnios was probably caused by Michael's choanal atresia.

My son was delivered, cried once and then turned blue. The neonatologist tried to pass catheters down both nares but they were completely blocked, so he was immediately intubated. A CAT scan showed bilateral bony and membranous choanal atresia. The doctors at the hospital where he was born had only seen this six times in 20 years.

At two weeks of age, Michael had his first choanal atresia repair. Stents were placed for two weeks. After one week of having the stents out, his passages closed. The ENT operated again, same result. A third operation was done, again unsuccessfully. I finally got Michael discharged from the hospital where he was born and got him to Columbia Presbyterian-Babies Hospital in NYC. The ENT there operated on him for the fourth time, placing stents that had no resemblance to the other stents. These were huge stents that even stuck out of his nose and were inserted through his upper lip. This ENT left the stents in for a long eight months. Our new ENT performs about eight choanal atresia surgeries a year and sees approximately two new kids with CHARGE a year.

The stents were removed under anesthesia by our new ENT. The other ENT removed them by clipping the sutures inside the nose, which hurt Michael. Now at 26 months, his passages are still wide open. Another ENT, Dr. Cotten in Cincinnati Children's Hospital, who has the most experience in this area, said that our ENT did an exceptional job, just as he would have done. Dr. Cotten is known worldwide for tracheal reconstruction and decannulating children in cases that were particularly difficult.

Hopefully Michael's passages will remain open; our ENT feels confident that they will. This was a major medical milestone to have surpassed.

## AIRWAY MANAGEMENT

by Susan Appell

Abby, our five-year-old CHARGER, has swallowing dysfunction resulting in copious secretions requiring frequent suctioning, tracheomalacia, bilateral choanal atresia (repaired), sleep apnea, obstructive airway as well as other medical conditions related to CHARGE syndrome. Abby's respiratory complications resulted in repeated pneumonia, pseudomonas, and frequent respiratory infections during the first year of life. During this time, Abby's cardiologist indicated that pulmonary hypertension was evident in her echocardiogram.

At 10 months old, Abby was hospitalized at Johns Hopkins Children's Center in Baltimore for pneumonia or pseudomonas. Her pulmonary physician ordered a sleep study which determined Abby had sleep apnea/obstructive airway. She explained that Abby could not grow or develop properly if she was not going into the appropriate levels of sleep which allowed her body to properly oxygenate itself. We had two options: placing a trach or using the Bi-Pap Airway Management System when Abby was sleeping. Abby has used the Bi-Pap machine from 10 months of age until today. It has been a true blessing and has allowed Abby to develop mentally and physically. Since there is no evidence that Abby suffers from pulmonary hypertension. She is able to fight off colds and has not used an antibiotic for over three years. Her secretions remain copious and her swallowing has not improved, however, if it were not for this machine, Abby would be trached or worse she may not be alive today.

The Bi-Pap Airway Management System is exactly that - it is a machine that helps manage a person's airway when he/she has difficulty breathing due to sleep apnea/obstructive airway. It does so by blowing a predetermined amount of pressure into a person's airway when they inhale with a predetermined amount of constant air pressure when they exhale. The machine can be adapted to a person's specific needs. Our Bi-Pap is made by: Respironics Inc., Murrysville, PA, 15668-8550, 412-733-0200, FAX: 412-733-0299, 800-345-6443.

The machine itself is smaller than a computer monitor and is portable. The unit filters the air and has adjustments to control the air pressure. Air pressure adjustments are made as a result of a sleep study and then routinely checked by our medical equipment company which services our machine every three months in our home. One short piece of special tubing comes out of the main machine and attaches to the humidification chamber. The humidification chamber is a small plastic bowl that sits on a hot plate. The hot plate has a temperature control which monitors the water temperature. Another tube leaves the chamber and is attached to the mask which has little portholes to allow air to escape so that carbon dioxide levels do not build up inside the mask. The child wears a little silk bonnet around her/his head which Velcro's to the mask. There are various masks available depending on your child's needs. We have learned to prepare her machine first (turned on and distilled water poured in chamber). We put Abby's mask on and then attach tubing so that air doesn't blow in her face while attaching the bonnet. We also learned to put a piece of duoderm on the bridge of her nose to reduce the pressure from the mask to prevent a pressure sore and to place Lacrilube in her eyes to prevent any drying in case the seal around the mask loosens and air blows toward the eyes. We requested an alarm system be attached to the machine so that in case of a power outage or Abby's pressure dropped an alarm would alert us.

Abby has gone from a very sickly infant who woke up every half an hour due to coughing, to a happy and fairly healthy five year old who now looks forward to going to bed and eagerly wants her mask on. She knows that this machine helps her and it surely shows when she is able to stretch out across her double bed and relax to enjoy a sound nights sleep without interruption. When Abby is on her Bi-Pap her secretions stop and unfortunately this is the only time she gets relief.

## GROWTH

by Marilyn Ogan

For two years, Kristin followed a pattern of growth that ran below but parallel to the standard growth curve at the pediatrician's office. Kristin's growth then began to level off, falling further away from the normal curve as the curve began to climb. Dr. Sandra Davenport suggested we consider an endocrinology evaluation for Kristin. We consulted a pediatric endocrinologist, who reviewed her growth pattern, evaluated Kristin's nutritional intake and energy expenditure and ran some tests. The testing involved L-DOPA stimulation and blood drawing every hour for 4-5 hours. Kristin "failed" the test and was diagnosed with severe growth hormone deficiency.



*Kristin - Fall 2001*

Kristin began growth hormone replacement therapy in October 1995. We have had more than three years of treatment and it seems to have helped her very much. She has responded well to the therapy. One of the first things we noticed was increased muscle tone. Kristin suddenly had more strength and endurance than before. Her development skyrocketed and she was GROWING! In the 18 months prior to testing, Kristin had grown only 1/4 inch! Kristin has now reached the stage where the faster-than-normal growth rate is declining, but she is still progressing and responding to therapy. Our endocrinologist stated that Kristin was not now experiencing tremendous growth, but he felt that without growth hormone replacement therapy, Kristin would not be growing at an acceptable rate. We have to agree.

Kristin will receive growth hormone therapy for several years, until she achieves her maximum growth potential. The only drawback to the therapy from our point of view is that it is done by injection. Kristin gets a shot five nights per week. We rotate the injections between four sites. At age six, Kristin had already reached a stage where she asked to take a "rest" from the "help-me-grow shots." She had several years of treatment remaining. I anticipate it being a big issue for the family in the near future

If you suspect a problem or are concerned about your child's growth, seek appropriate services for diagnosis. A pediatric endocrinologist would be your best choice. Don't wait for puberty to see if the child "catches up" -- that is too late for growth hormone therapy to help. As a rule of thumb, any child who is only as tall as children two or more years younger or who falls away from his or her previous growth pattern should be considered for an evaluation. And remember,

children with CHARGE have lots of reasons for being small, but a few really do have growth hormone deficiency.

The Human Growth Foundation (HGF) is an excellent source for information if you have questions about growth patterns, deficiency, hormone treatment, and even ways for an individual to cope with short stature. They have multiple publications (for small fees), publish several newsletters, and offer a parent-to-parent support program.

Human Growth Foundation, 7777 Leesburg Pike, Suite 202-S, Falls Church, VA 22043  
(800) 451-6434 phone (703) 883-1776 fax  
<http://www.medhelp.org/web/hgf.htm> email: [hqfound@erols.com](mailto:hqfound@erols.com)

Another source of information is the MAGIC Foundation for Children's Growth. Their website has some marvelous information about growth hormone therapy. the Frequently Asked Questions page is very informative.

MAGIC  
1327 N. Harlem Avenue  
Oak Park, IL 60302  
(708) 328-0808 phone



## BEHAVIOR

by Ana Saruski

When my son Joel's behavior became an obstacle in school and social situations, I consulted his neurologist and tried several medications to address mainly his aggressive behavior towards his peers and sibling. This aggressive behavior included pushing, pulling hair, and kicking. Several trials of different medications failed to help improve his behavior. After seeing the negative effects of some of these medications, and reading about the potential side effects, I decided to consult a natural nutritionist, who put Joel on a sugar free diet for his behavior, and dairy free diet to improve his asthma. I immediately saw changes in his behavior, which I attributed to this diet and the combination of vitamin supplements that the nutritionist recommended. His aggressive outbursts decreased, and his teachers and therapists commented on the changes in his behavior, and felt that he was much easier to work with since he started on the diet.



Joel's diet consists of pureed foods. On a daily basis, I mix the following supplements in his food: acidophilus to prevent yeast overgrowth; amino acids for protein supplementation; B complex to improve his behavior; calcium supplement to replace the calcium he used to get in dairy products; and a natural multi-vitamin. Joel has been on this diet for nearly one year. In the past month I also added a DHA supplement called "DHA Junior", which is especially designed for children 3 and over. The DHA supplement is meant to provide fatty acids which are believed to improve brain and eye function.

## **EARS - DEAFNESS, and LANGUAGE**

by Yuka Persico

Keith, who is now almost 10, has been trached since he was six weeks old. Because he had difficulty voicing around the trach, when he was about a year old, I decided to begin signing with him. Because I believed at that time that he had hearing but couldn't voice, I only was learning and using nouns, adjectives and verbs - children picture book language. His first signs were 'more', 'please' and 'read book'. By the time he was two he was using about twenty signs, and when he was three he had a vocabulary of over 300 signs, often stringing several words together to shape a concept. Since he was learning the signs so nicely, we were becoming quite detailed in our list of fragments to describe things.

When Keith was three his hearing was routinely tested for placement in special education preschool we found that he is profoundly hard of hearing, with a 90-120 dB loss. At that time, the infant assessment team had targeted Keith for the orthopedically handicapped classroom, as he had a trach, a gastrostomy, was not independently walking and had balance issues. I had contacted the deaf and hard of hearing (DHH) teacher because I was eager to place Keith in her class even before I knew he was deaf, because signing was his mode of expression. When the DHH teacher called me back, I had already absorbed the fact that Keith was deaf; she however had not heard the news yet. Our conversation was awkward, but revealing. She began by politely outlining for me all the reasons non-deaf children use sign. She praised the IEP presentation I had prepared for Keith and was impressed by his vocabulary and my documentation of it. She began to explain how being deaf is different than being hearing and utilizing sign when I interrupted her and said "But Keith is deaf. He has a profound hearing loss!" It seems odd to say but she was very excited about this, mostly because she is a wonderful teacher and exceptional at reinforcing language. Now she was attached to Keith and to his agenda - that he needed a complete language.

There was something in the beginning of the conversation we had, before she realized Keith was deaf, and she was talking about language and vocabulary that the green light went on in my head. I had given Keith a vocabulary, but not a language. I hadn't really thought about that difference yet. I frantically began to fill in all the blanks in my signing, trying to create whole sentences, sign all the words in a storybook, and even sign all I said to anyone. Since I was quite determined about this, it really limited my conversation for a while. I was frustrated by my limited progress, and how awkward at times it was to learn the signs from a book and use them with any conviction.

Keith started special day preschool in the orthopedically handicapped class with a full time sign language interpreter, who was a deaf education student from CSUN (California State University – Northridge). When I saw her comfortably and fluently sign a complete language, I knew this is what we needed, and I invited her to come work in our home, and just sign sign sign. She came three days a week. She signed storybooks. She sat in front of the television and signed videos. She signed when she talked to me and she patiently answered all the questions I had from literally "how do I sign this.." to questions about deafness and its culture. I put ads up at Moorpark College and CSUN, and found two more college signers to come into the home the other days. Our entire family social life was based on these wonderful students and at dinnertime our table was always full of hungry students signing signing signing. I wanted not only the direct language presented to Keith, but the richness of the passive receptive language that hearing children naturally benefit from by simply being in the room when adults are speaking. I wanted to bring Keith's language to that place of fluency that deaf children of deaf parents demonstrate.

It really took about 4 years for Keith to start signing language back. At five he was indicating his preference for certain nicknames - like any three year old might do, and at six he would dictate simple repetitive stories. At seven he began to be able to read, spell and speak intelligibly, and that's when the whole thing took off. He went from simply always doing his best, to doing his best and doing fantastically. When Keith had just turned nine, he attended Space Camp in Alabama, and won the "Right Stuff" Award, ran for Vice President of Student Council for the entire Elementary School where his DHH class is located and won, and has been on the honor roll every quarter since letter grades have been given. As much as language has been the key to his emotional independence and "coming into his own" he also seems to love language, playing with it, using it and reading it.

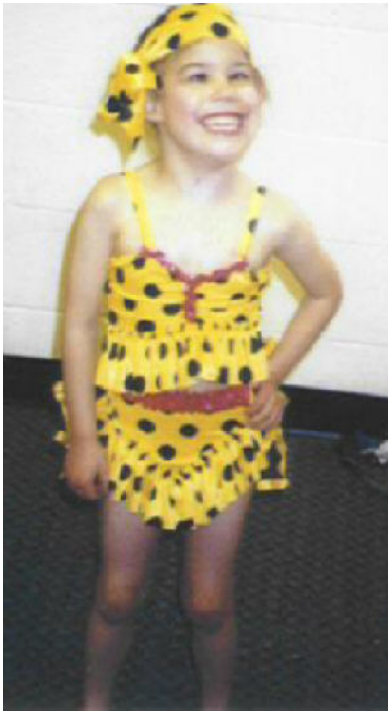
With language, not only could we now know our child intimately, but he could freely unleash himself upon the world with joy. It's not so much that Keith is "all caught up," because he isn't, but now his progress is no longer bittersweet and we no longer compare him to the "normal" life he would never have. He has complete ownership of his own person, just like anyone else. He has his own strengths and weaknesses just like anyone else. He has become completely equal and stands on his own without qualification or explanation.



Keith walking his dog, November 2001

## CAITLYN, AGE 6

Jeanne McMullen, mother and frequent contributor to the list  
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Caitlyn, 6, at dance recital

With Caitlyn's severe heart problems, respiratory problems, and developmental delays, I never thought she'd be able to take ballet class, unless it was designed for special needs children. Yet I found a teacher who was more than willing to have Caitlyn in her class. Though Caitlyn's abilities are very limited and she tended to wander during class, she DID take ballet class and did participate in the recital. It brought tears to my eyes every time I saw her in her leotard and in her costumes.

It has been a long road to this point. Here are some of my observations on what helped us with Caitlyn:

*"All of her hospitalizations have taken their toll on her nervous system."*

### SENSORY INTEGRATION

One of Caitlyn's most distressing issues have been related to sensory integration (SI). All of her hospitalizations have taken their toll on her nervous system. The bright lights of the ICU, the constant beeps and alarms of the monitors, repeated IV sticks, blood tests, and so on. This has left her a very anxious child who is easily overstimulated by her environment.

As an infant, Caitlyn didn't want to touch anything including her toys, nor did she want to be touched. She arched her back when we tried to hold her and cried when we caressed her skin. We soon learned it was only a firm, deep touch, and vigorous bounce that she found calming. She cried most of the day and night for most of her first two years. She refused anything in her mouth, teethingers or food. Due to her resistance to stimulation, her development was stagnant.

*"OT is NOT job training, but is fine motor therapy"*

What helped Caitlyn tremendously was occupational therapy (OT). I didn't realize until she was almost two years old that OT is NOT job training, but therapy for fine motor and daily life skills.

OT often includes SI therapy which is crucial to CHARGErs like Caitlyn. SI therapy helps desensitize a child to her world. Desensitizing her sense of touch, her mouth, her ears, and eyes. I would recommend that every CHARGEr have OT from birth, particularly those who have spent a lot of time in the hospital. SI therapy also covers vestibular disorders (a lack of balance) common in CHARGErs due to their ear abnormalities. Starting OT at age two was much too late because she had missed a lot of critical development periods, and it has proven impossible to catch up.

An important part of Caitlyn's SI Therapy is the brushing program. It must be done under the supervision of an OT and it must be done as prescribed to get the full effects. This means every two hours, for the initial two weeks, which can be difficult. A handy way to stay on top of it, is to do the brushing with every diaper change. Though there was a slight improvement with the implementation of the brushing, it was only after we made a concerted effort to do it every two hours that we saw a breakthrough and progress in her development began.

### **GROSS MOTOR**

Caitlyn has had low tone and vestibular problems, so early physical therapy has also been important. I want to stress that parents should consider using their private insurance to help pay for private services to supplement the early intervention (EI) services for their kids. The added therapy can make a difference in the gains a child will make. Often EI does not have as much equipment as the private programs do, and much of it is too large and bulky to be transported to the home. Also, often EI therapist may not recommend medical enhancements that may benefit the child such as orthopedics or walkers. By providing more intensive therapy during the critical stages of development, the child's prognosis may be far better.

### **ORAL MOTOR**

Oral defensiveness is also a common problem with CHARGErs, particularly those who are tube fed, even for a short time. For this problem we used a NUK brush found in Gerber's first toothbrush set, the brush with the round, rubber-nubbed tip. For starters she would only allow it in her mouth for a second. Progress was slow. Eventually we were able to massage her gums, tongue, and all around her mouth, and finally we were able to dip the tip in aversive textures in order to make great gains in this area. An occupational or speech and language pathologist can recommend a treatment program to assist in this area.

To stimulate her speech, we attempted a variety of games. We imitated her coughs and sneezes to make a game of it, then encouraged her to imitate our coughs and sneezes. We then were able to gain her cooperation in imitating animal sounds. Early on, we purchased a good sign language dictionary and signed to Caitlyn as we spoke, emphasizing the main words of the sentence such as "Bath" "Bedtime" "Eat" etc. Soon Caitlyn had created a few of her own signs and thus had started communicating her basic needs.

Sign language truly helped reinforce the concept that all words have meaning and the importance of communication. It did NOT discourage her verbal speech, but rather promoted it. This principal applies to BOTH hearing and deaf children, and was very beneficial to Caitlyn who has normal hearing, albeit with a delay in processing.

*"I think this helped her to keep from falling further behind"*

### **COGNITIVE**

In addition to working hard with Caitlyn on the skills she was lacking, we also took the extra time to walk her through age-appropriate activities that were well beyond her means. Using hand

over hand and other modifications, we eventually learned that though Caitlyn had very poor motor skills, her cognitive skills were indeed a strength. As her motor skills were almost stagnant, our efforts brought about great strides cognitively. The key was remembering that Caitlyn did not have to accomplish each milestone before moving onto consecutive ones that required alternate skills. Identifying letters and numbers were taught *and learned* before she mastered stacking blocks and doing puzzles. We then were able to build on her strengths to accommodate her weaknesses.

At age 3, though unable to manipulate many baby toys such as crib gyms and pop up toys due to her poor fine motor skills, Caitlyn knew her name, address, and phone number. By age 4, she knew her upper and lower case letters, all her numbers and shapes, and colors. At age 6, Caitlyn read her first book, *“Green Eggs and Ham”*. She has a memory that far exceeds her age, another strength. We build and take advantage of that.

*“Social skills are some of the most important skills for children”*

## **SOCIAL SKILLS**

I don't know what to say here other than if I had been a better advocate for my daughter at her doctor appointments she might not have spent most of her life acutely ill. Many of her medical problems went months and sometimes-even years out of control. This prevented her from participating in community activities and play dates. It seems as though the majority of Caitlyn's medical problems were addressed by the appropriate doctor until the problem had reached such a climax that it created irreversible and permanent damage.

Social skills are among the most important for children, particularly a disabled child. Relying on their peers for assistance will be critical as they mature. Yet these skills are often overlooked by doctors in the early years as being trivial compared with the severe medical problems these children face.

Caitlyn becomes very anxious around peers and is very disconnected from play. She appears mildly autistic in many ways. She prefers to spin around in circles and make odd vocalizations when children are playing nearby. She is unable to focus on any particular toy or activity instead always flitting about from toy to toy much more so than other children her age. I think the most important thing we did do for Caitlyn's social skills was to remind her of her strengths, of our love for her, and make the most out of nonclinical times to get a smile out of her. She is a very happy and social child among adults.

*“Even if vision services are inaccessible on a regular basis, go for evaluations”*

## **VISION**

Vision services should be available from birth through early intervention and through your closest School for Blind Children. Take advantage of these services with or without the presence of colobomas. Caitlyn has crossed eyes and it wasn't until 1-1/2 years that we started getting vision services. That was when we learned she couldn't track objects at all--a three-month skill. Tracking is critical to depth perception, which is necessary for safe mobility and locating objects. Some children also have visual processing difficulties, cortical visual impairment, or other vision issues. Vision therapists can often recognize and treat such conditions that may go unnoticed by the parents and other professionals.

Even if vision services are inaccessible on a regular basis, go for evaluations and consults every few months to learn what exercises can maximize your child's vision. Children can



compensate for even significant visual deficiencies particularly when taught proper techniques. Therapy can maximize what vision a child has, and also prevent delays in other areas. Therapists can recommend modifications and alternatives to traditional toys to make them interesting to a child with low or no vision. Vision services can be quite beneficial even when a child's vision difficulties originate in the brain.

Orientation and Mobility Therapy is also important to children with visual deficits. Some vision therapists are trained in O & M therapy, yet some are not. O & M carries it's own evaluation and therapy.

*Don't be afraid to sign your child up for "typical programs". Often you will face prejudice and ignorance and be turned away, but keep looking to find programs that will suit your child. He will benefit from the experience.*

*"It was only after five years of hounding that Caitlyn finally was referred to an immunologist"*

One final note:

Caitlyn always had either croup, bronchitis, pneumonias or other severe upper respiratory infections with only a few days between each illness, if that. Each illness was treated individually as it arose with nobody putting together that she was always sick, really sick. Only after five years of hounding her doctors was Caitlyn finally referred to an immunologist for testing which led to a diagnosis of a primary immune deficiency. Caitlyn is now thriving with her IVIg (intravenous immunoglobulin) treatments. Though I am thrilled to have found answers and a treatment for her, one that has changed and improved our lives drastically, I am saddened by all the years she has lost socializing, playing, growing, and developing. Immune deficiencies have rarely been mentioned in CHARGE literature, however, it has frequently been discussed how sick our kiddos get, even in the CHARGE manual. I would be very interested in seeing more parents whose kids are always acutely ill as Caitlyn was, pursuing this problem as a cause. Caitlyn's doctors didn't take her illnesses seriously, even though her temperatures frequently were well over 104 degrees, even though they resulted in a number of hospitalizations. I feel that many of her developmental and behavior problems are a result of her constant, acute illnesses. Parents must be proactive in order for their child to receive appropriate treatment.

***Editor's note: Here are two more stories from Jeanne and Caitlyn.***

## **A WEEK IN THE LIFE OF CAITLYN, AGE 6**

**What have we been up to this week? Let's see.**

**Monday A.M.:** Ophthalmologist recommends another eye muscle surgery, but advises that after 4 unsuccessful surgeries, her odds of success are poor. He is understanding when we decline.

**Monday P.M.:** Our regular cardiologist is on vacation, so we are forced to see an alternate one. Of course this means reinventing the wheel. We have come to terms with her prognosis after obtaining three previous opinions on her heart, all agreeing that she is not amenable to surgical intervention. Yet today, we are asked to strongly consider going ahead with a surgery attempt. This, in light of advice that if it is not successful, will greatly diminish her quality of life and possibly be fatal.

And these doctors wonder why I'm so cranky at appointments!

**Tuesday** A.M. Psychiatrist talks about adding Prozac type drugs to her already daily dose of 10 medications.

**Tuesday** P.M. pediatrician notices she has developed allergies and puts her on Claritin-- medication #11!

**Thursday** P.M. Pulmonary changes all her asthma meds because he confirms my suspicions that she's still aspirating and can feel that she is not exhaling fully anymore (again). Cannot increase GI meds because they're maxed out.

**Friday** A.M.: New Neurologist stands us up. Her secretary forgets to call us to cancel. I catch the neurologist on my way out whom I've seen before, but who has never met me. I chew her out ranting and raving for 15 minutes--so livid I was shaking. After all, I got my kid up at 5:30 in the morning and listened to her tantrum about it for an hour. To top it all off, she woke up that morning with pink eye! Then dragging her to the hospital (hoping to get antibiotic eye drops to save another trip back to the pediatrician) only to be stood up. At the same office that always makes patients wait at least three hours for a fifteen minute visit with the doctor. Fifteen minutes which last time were spent entirely on lecturing his accompanying students on the CHARGE facial features before them (in my daughter). I have to admit I just love it when they point out her colobomas--SHE DOESN'T HAVE ANY! Anyway, I can't BELIEVE they didn't call security!

**Friday** P.M. After three urgent phone calls to the pediatrician, I can wait for a prescription no longer. I leave for our GI appt and keep her on my lap the whole two hours in the waiting room to keep her from touching anything. GI doc increases her GI meds--even adding yet another medication (make the grand total 12!) and increasing the dosages that she's on.

**Saturday:** I hear back from the pediatrician who calls in a prescription for antibiotic eye drops. I sadly call to cancel the birthday party we were supposed to go to remembering the last one she was invited to she missed due to a hospitalization.

**Tomorrow** (Sun): She'll be getting her IV infusion so can't make that birthday party either. (Yes, I am very grateful that she's been invited to so many parties this year).

Editor's Note: A year later, Caitlyn is only on nine medications, only six of which are daily. Ten kids attended *her* birthday party, which was a grand success.

## **2) Caitlyn's Christmas story**

Every Christmas we receive a letter from a wonderful family. The story behind it is beautiful. It is with tears in my eyes and pride in my heart that I share this story.

When Caitlyn was admitted to the hospital for an emergency open heart surgery at 3 weeks of age, I sat in the cardiology waiting room crying. A woman approached me and gave me a big hug offering words of encouragement and prayers. She introduced me to her 9 year old granddaughter and we chatted briefly. During such a scary time, the kindness of this stranger left an impression on me that I wouldn't easily forget.

At 3 months of age Caitlyn was in severe heart failure and septic. She had arrested several times that day, and had to be in a medically induced coma to stabilize her. The surgeon said she had developed an aneurysm from the previous heart surgery and that he had to operate as soon as he could stabilize her enough to withstand surgery. He said for the first time in his



career, he called the Ronald McDonald House and told a family that drove in from out of town that their surgery would have to be canceled to accommodate Caitlyn. He said he didn't even know if she'd make it till morning.

Caitlyn's surgery was very rough as she suffered a lot of complications. It was very touch and go for days. That Sunday, as I sat in the ICU waiting room with my husband, a woman greeted us warmly with a big hug. She saw my lost look and asked if I remembered her. She recounted the experience in the cardiology waiting room several months earlier, and I slowly recalled the experience. She said her daughter, Karlye, had just had open heart surgery that day that had been rescheduled from the previous week due to an emergency. She said, "A baby was dying and needed the surgery right away." My husband and I said shocked--Dr. Siewers at 7:30am Thursday? She said, "How did you know?" We said "It was our baby." She nearly fainted.

The woman described what had happened Wednesday night after getting the call from the surgeon. She told us how Karlye was so upset about having to drive back home several hours away only to come back and get worried all over again but her mother said, "You're 9 yrs old and have lived with this heart condition just fine your whole life and you can wait until Sunday. This baby can't wait until Sunday, she'll die if she doesn't have the surgery tomorrow."

The girl said "Then if I wait until Sunday so she can have my surgery slot tomorrow then would I be helping to save this baby's life?" Her mom said yes. The girl asked if the baby would still be at the hospital on Sunday and her mom assured her most probably. The girl asked if she could go shopping for a get well present for the baby and her mom agreed.

The woman showed us the stuffed bear Karlye picked out for Caitlyn. I asked them for their address to send a thank you note to the girl. A year later, I sent them a Christmas card with a picture of Caitlyn and a note thanking them for their kindness and telling them how much it moved me. I wrote that though they probably don't remember me, I'll never forget how deeply they touched me. They replied with a letter detailing how much their daughter talks of Caitlyn as the highlight of her life. How she says she "helped save Caitlyn's life".

It's been five years since our two brief encounters in the hospital. This past year marked the first time that the girl, now 14 wrote me herself. Karlye wrote that Caitlyn was an angel, a true angel and a miracle child. I asked Caitlyn if she knew what that meant because so many people say that about her all the time. After some thought she said, "I think it means to know me is to love me."



Caitlyn, age 5-1/2 helping mom make birthday cake

## **HUMOR**

Casey Fisher, mom, Founder of the list serve, and a major advocate for CHARGE California

Humor has been our family's saving grace. Aari uses it too. When people ask him what happened to his ear his standard reply is that "Daddy forgot it at the hospital." When he had his first choanal atresia repair, the doctor used a bright red-orange tube for the stent. After six months of getting asked the what the devil happened, I started replying to the obnoxious ones that his doctor got stuck in an old Laugh-In episode (boy am I telling my age on that one).

At a meeting with the school one day, I said that Ari was a FLK. The school psychologist began writing furiously, saying she was gonna have to look that one up as she hadn't heard of a syndrome or medical condition called FLK. Ari's teacher and I got to laughing so hard it took a while to explain to her that FLK meant Funny Looking Kid.

Once when we were out somewhere, Aari was reading a book. Being so little, about the size of a 4-year-old (he was about 7 at the time), some lady asked him if he would like her to read it to him. He looked up and told her "Thanks, but I'm quite able to decipher written language." The look on her face was priceless.

JULIA, age 18

Marie Patterson, mom  
Australia

## **TRANSITIONS**

We have Julia, now 18 and what an experience it is raising a teenager who we think cannot do lots of things, and it brings us to reality about their ABILITIES not their disabilities. Julia is doing three day a week work experience, and next term she will be going to work experience at McDonald's, clearing tables, wiping up, etc. This is just one of the milestone we enjoy. With all the down sides of a child with CHARGE, it is nice to get some light stuff. Our problem looming is what happens aft school. There are very limited opportunities for Julia working in an environment without support staff. Finding a uniform to fit her is also a problem (she is only as tall as an eight-year-old). Another problem in Australia is the Proof of Age Card does not necessarily admit her to a club where her size says NO GO. We are working on the discrimination process and believe me I will find an answer. Because of the mileage barrier for clubs in New South Wales, we have to prove she lives outside the area but without a driver's license or residence proof, it is difficult.

## MY FAMILY IS A WONDERFUL INFLUENCE IN MY LIFE

Andy Prouty, age 19

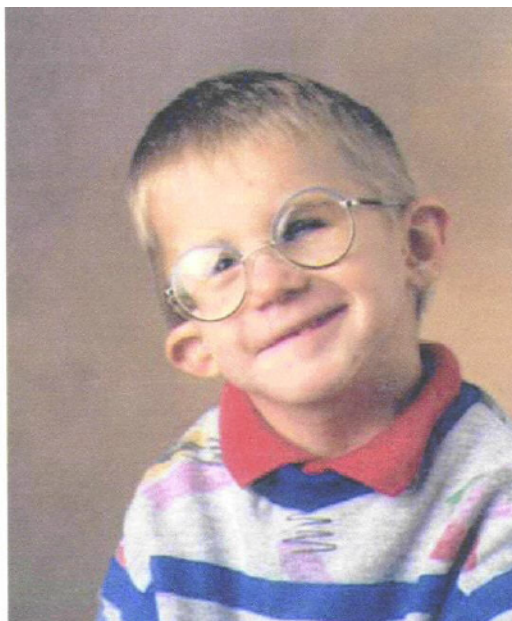
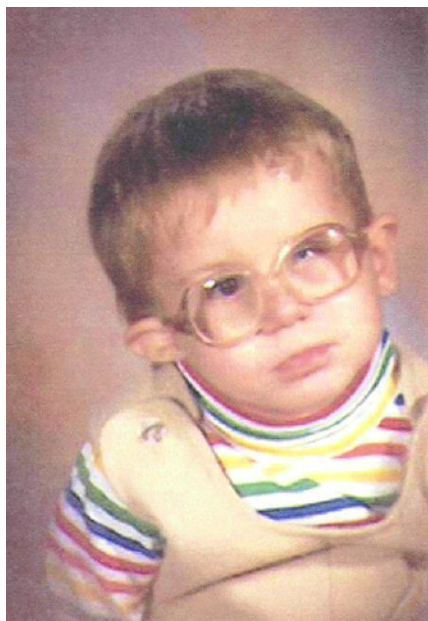
When I was born, my parents learned that I was deaf and had vision loss. They wanted me to be successful in my life. My family also treated me equally to my siblings. I have the same responsibilities and education needs.

The most important thing my parents learned was communication. Since I am deaf, they need to learn to sign so they went to programs that teach sign language. My siblings learned how to sign by copying my parents. Also my closest relatives have learned to sign to communicate with me.

Besides communicating with me, my parents give me best services possible. I get many services from State Service for the Blind such as monocular to see faraway objects, magnifiers to see fine prints, and orientation and mobility training. My elementary, Junior, high schools, and Vector, a program in Hennpin Technology College, for people with special needs, also give me good services such as 1:1 interpreters teachers who are well aware about deaf-blind culture. Finally, I went to a deaf church, Bread of Life. This church has high technology that allows me to see the pastor far away with a video camera and a monitor, and everyone signs!

My parents also want me to have many fun opportunities in the community. They let me fly to New York City alone to visit a friend. I also traveled to North Carolina and Ohio to go to American Association Deaf-blind. I attended group activities such as Teen Group, Children Linking Families, and Summer Transition Program. Also I took karate classes when I was young. Now I am taking pottery classes every Saturday afternoon. All these experiences help teach me independent living skills and social skills. Those activities are useful for my future.

I have many support services and activities in my life because my family wants to help me become an independent adult in the future.



## LET ME TELL YOU A LITTLE ABOUT MYSELF

Andy Prouty, age 19

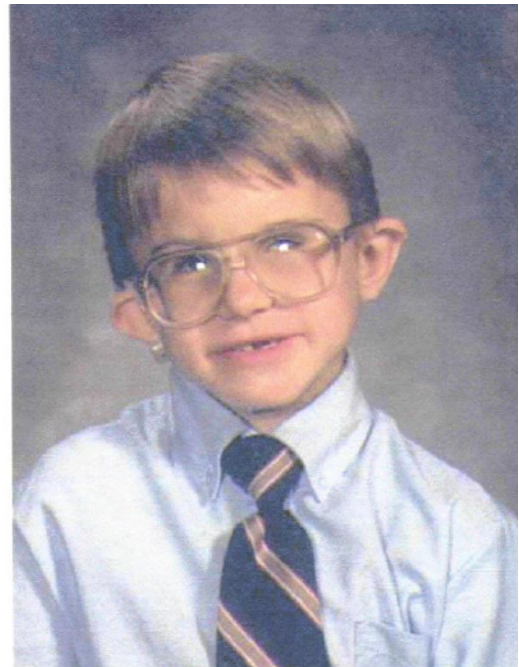
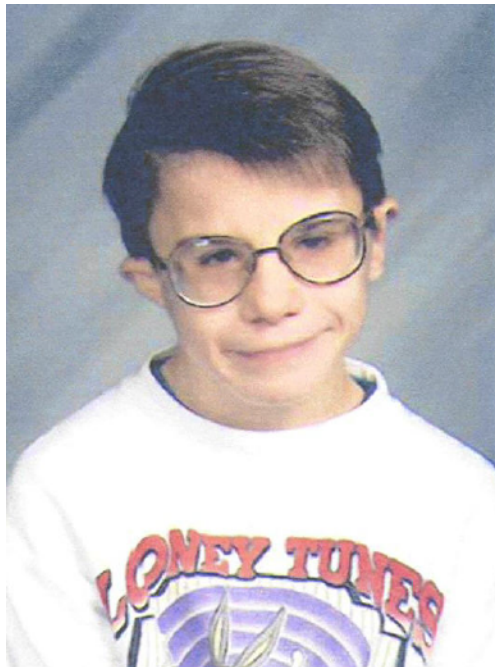
I was born in Oregon in 1981. While I was growing up, my family moved to many different states including Oregon, Washington, Utah, Maryland and Minnesota to find the best services. Our final stop was in Minnesota 13 years ago.

I am deaf and legally blind. I cannot see with my left eye but can see with my right eye. I can see general shapes, sizes, colors far away but hard time seeing details such as small features of an object. I wear glasses to help me see things more clearly. I only take my glasses off when reading and using the computer.

We live in the suburbs of Minneapolis-St. Paul, Minnesota. My parents both come from large families so I have lots of cousins who live all over the United States. When we get together at my grandparent's cottage in the summer we have so much fun. They try to sign with me but we also write notes back and forth.

I have many interests and hobbies. My outside activities include jumping on the trampoline, swimming, water skiing and kneeboarding. My indoor activities and hobbies are pottery, reading books, playing video games, watching movies, drawing and playing games on the computer.

When I was little I enjoyed reading teenager mutant turtles and the Berenstain Bear books and I had EVERY ONE of them. Then I liked to read Disney Books. Every time I went to a new Disney movie, we bought the book about the movie so I could understand the story better.





I also love comics, especially Garfield. I think I have 10 of those books on my shelf. I liked short books until I began reading Harry Potter stories. I've read all 4 books and can't wait for the next one to be printed. While I wait, I read "Girls to the Rescue" Books. I think those books are great.

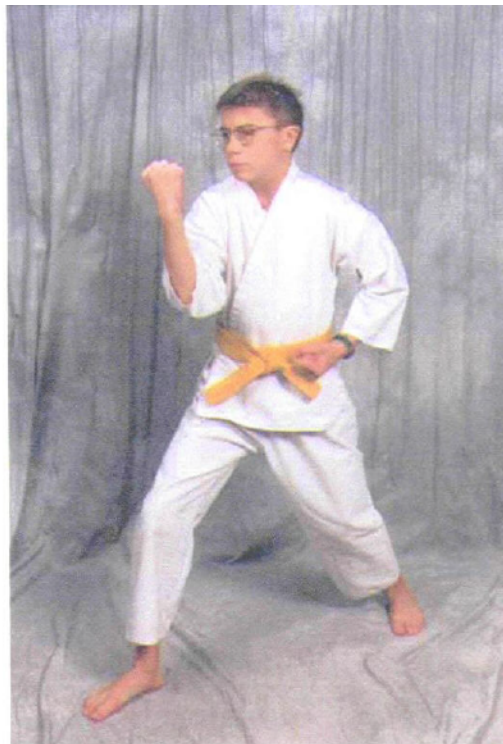
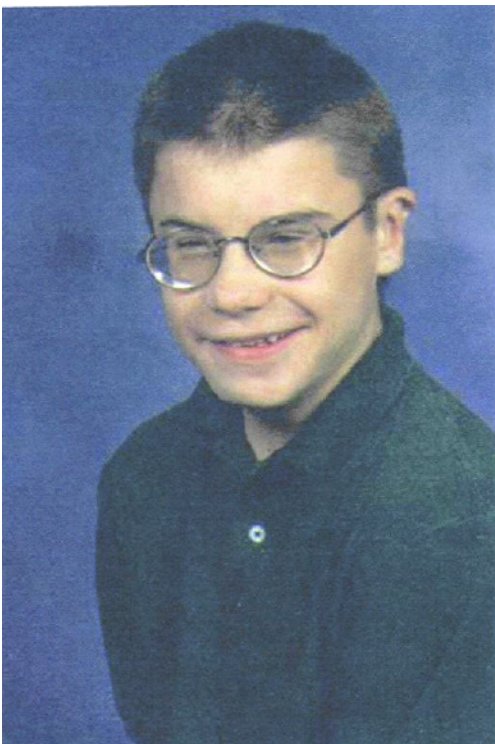
I took pottery classes in high school and when I graduated, the school gave me an old potters wheel that I use in my basement. I have taken a pottery class in Minneapolis with an interpreter and it was fun.

## **Interveners**

Andy Prouty, age 19

I have had many interveners though my life. My two first interveners were my parents! I have had interveners in school and in the community. All of my interveners were wonderful influence on me because they taught me many things I need to know. After my parents, I had other interveners. When I was a little boy, age 2, my parents hired two teenager girls who helped me learn. Wendy was in high school and Kim was in college. They both knew a little sign language.

When I was 2 years old I started school. My parents made sure my teachers and interpreters understood how to work with me because I couldn't see as well as my deaf friends. They always made sure the DeafBlind Technical Assistance Project came and gave an inservice to the staff before each school year.



I have interveners in the community too. First, it was in our home and then in our neighborhood. As I got older, we started going to stores, museums, fairs and other fun places. My interveners not only taught me and gave me access to my community, but we also had fun! We often go to the community to do various activities such as Mini Golf or Valleyfair. As we had fun, I learned how to buy clothing, foods, and many different kind of life skills I need to know.

One intervenor I had was a real neat guy. He was about 25 years old, deaf, and a professional social worker. Keven was my favorite intervenor. He was also a role model, and a good friend.

In the future, as I become more and more part of the adult world, I might not need an intervener. But I can have good friends that can give me good advice, and support me. I already have two very good adult friends that help me now.

Maybe some day I will become an intervenor for some young person who needs my help. Maybe I can be a role model for someone. People have helped me all my life; I want to help them when I can.

Well, this is kind of a short speech, but I hope I will have a chance to visit with you all weekend. I look forward to trying to answer any questions you have. I love family weekends in Minnesota and Wisconsin. This is my first North Carolina family weekend. I hope I can make it more interesting and informative for you!

## HOW MY SON HAS INFLUENCED MY LIFE

Sally Prouty

Before I get started, I want to say, the past 19 years have been incredible. The journey has been incredible, I have learned so much about myself, advocacy (both medical and educational), and “the system”, I would do it all over again. It hasn’t been easy, but nothing really good ever is easy. Please feel free to read the article my husband and I wrote about our journey. You may have read the analogy of going to Holland in the past, but we matched it to our experience and how it proved true for us. (I did spend two weeks in the Netherlands last year and it REALLY IS a wonderful place!)

Here is a synopsis of our family’s experiences and how we coped.

When our son Andrew was born 19 years ago, we felt like we were the only people on earth (except Helen Keller’s family) who was having the experience of a child who was DeafBlind. If I saw then the audience in front of me today, I would have felt much differently about those early years. I have learned over the years that there are many devoted, caring, and knowledgeable people like you, but finding them is like a needle in a haystack. Let me tell you about my family’s journey.

I was living what I thought was the American dream with my husband Mike, (who by the way is my Rock of Gibraltar), and son Billy. I was a stay-at-home-mom and Mike had a job working for the United States Forest Service that allowed us to live where people vacationed. We literally lived at the end of the road at the base of Mt. Adams in Washington State. Bill was a happy 2-year-old who we knew would have a perfect, carefree life.

In September of 1981 our second child, Andrew, was born. Within two days, Andrew was diagnosed with CHARGE syndrome. What a shock! First we were told he was profoundly deaf; then we were told he was blind. We had never met a Deaf person or a blind person. What were we going to do? We cried, we hoped for a cure and we cried lots more, but fortunately for Andrew we did our best to face reality and realized within those first months that we were wasting precious time feeling sorry for ourselves and our lost dreams for Andrew. We had to pull ourselves up and provide a meaningful life for him.

Once the major medical issues were taken care of we knew we needed to focus on his communication and future independence. We wanted to know that when we were old, we could feel satisfaction in knowing we had provided Andrew every opportunity to be as independent an adult as possible. Our goal has always been to develop a tax-paying citizen.

Fortunately for Andrew we were hooked up with “the system” by the time he was 2 months old. Don’t get me wrong. I wasn’t enthusiastic about it, **I was scared** – this wasn’t what we expected. We didn’t know where to start; we had never done this before. I occasionally remind professionals I work with that they purposefully got into the profession, parents DIDN’T! Andrew says he is like a pop-quiz to me.

Within this system, we were fortunate to meet great people early on and have positive experiences. Getting connected to resources early was very important to us and crucial for Andrew’s future.

The twice a week, 200 mile round trip drive to Portland’s Infant Hearing Resource Center was wearing on us. We knew we needed to move closer to services but to where? Strangers loaned us a motor home, we researched programs for DeafBlind kids across the country and set



off to find “Nirvana” - you know, a classroom of little DeafBlind kids. Guess what? No such thing!

As a family, we knew we needed to find the right balance of three priorities:

- 1) medical and educational services for Andrew
- 2) a lifestyle and living environment we could enjoy and
- 3) Mike's career. We kept these priorities in our mind continually.

First, we transferred to Southern Oregon where we were blessed with the talents of a DeafBlind specialist. We discovered the McInnes' Canadian model of Intervention was the answer for Andrew. His eyes and ears did not permit him to access his world so we approached our Lady Lioness club to help fund an Intervenor for the summer. They did and through that experience became convinced that the only way Andrew could access his environment was by 1:1 learning. The school provided a 1:1 Intervenor in the classroom as well.

By the time Andrew was 3 years old we heard about an Intervenor program in Utah so we packed up and transferred there. Again, we had experiences and met people who impacted our lives. Andrew and my husband Mike successfully lobbied the state legislature to fund a DeafBlind specialist. Three years later a job opportunity opened in Washington DC. After researching our other priorities of education and medical as well as lifestyle for the family, we decided to move on.

I should interject that I would NEVER recommend this nomadic life to any family. It worked for us but we also gave up a lot. We left wonderful professionals who also became our friends with every move. Fortunately, our oldest son, Bill, was young enough to think each move was an adventure. I often wonder what life may have been like if we had stayed in our small town of Trout Lake.

One year was all we lasted until our move to St. Paul Minnesota 13 years ago where, I am happy to say; we still live and have actually grown roots. We were happy to be back in the Midwest where we were born and raised and to provide extended family for our children and ourselves. Before buying a house, we researched school districts, looking for the one that would best suit Andrew's needs and provide the services required. Remember the three priorities we had as a family? Good education and medical supports for our children (Andrew) Lifestyle we enjoyed and Career. We finally found the right combination in Minnesota.

Andrew has always attended programs for Deaf students with adaptations including an Intervenor, large print, Orientation and Mobility training, technology such as a closed circuit TV and adaptive physical education to name a few. Convincing his school he needed an Intervenor was a struggle. They worried about dependency, but we finally persuaded them his vision required it. The term Intervener was always questioned, so we played the game and said Interpreter/Intervener..... that way, we figured they knew he needed something extra.

We have always tried to be involved in Deaf Culture. We go to a Deaf Church, I went through an Interpreter Training Program, used only Deaf or children of Deaf People to baby-sit, and helped start the Children's Discovery Project at DeafBlind Services Minnesota.

I'd like to share some lessons we have learned in our journey:

- Parents, please develop a communication system with your child as soon as possible. Don't waste precious time!
- Parents, maintain high expectations for your children and let professions know you do. They may buy in.
- Encourage decision making with your kids on day 1 and continue this practice all their life, it will make encourage independence.
- Parents know their child better than anyone. Trust your instincts. You know what's best. You will be with your child longer than anyone. Professionals will come and go but you are permanent and you must make decisions that will impact your child's life. Don't give up.
- Parents, remember Dorothy in the Wizard of Oz? Remember how she searched relentlessly for the Wizard to help her find her way back to Kansas? She was awestruck by his power. We did that for a while with Andrew. We went from doctor to doctor and teacher to teacher looking for magical solutions for his education and medical needs. We were looking for the answer. But guess what, like Dorothy, we had the answer all along ---trust yourselves.
- Professionals, please be willing to admit when you don't know the answer. Like all parents, I can't tell you how many professionals (doctors, teachers, therapists and others) we have trained over the years. I appreciate and respect hearing, "Gosh, I don't know, but I will try to find out." Don't feel you need to be a Mary Poppins drifting in with your umbrella to give a spoonful of sugar and make everything OK. It is not fair to you and it is not fair to families. It is really refreshing for a family to hear, "I don't know, but I'll work with you and we will learn together!" That REALLY builds good teamwork.
- Parents, develop the skill to be "Respectfully Demanding" when you have a need. A demand without respect puts people on the defensive.
- Parents, encourage active participation among people who work with your child. They will be much more enthusiastic meeting you for a breakfast meeting than a school meeting. They appreciate and remember an occasional thank you note or acknowledgment that you appreciate them. It's easier for them to say "yes" next time you have a request.
- Sometimes this doesn't work, sometimes people don't "get it." Twice we have taken our needs to the state legislature asking them to appropriate money for DeafBlind and both times we received it. Andrew testified at both, once when he was 4 years old and again a few years ago.

Andrew has always been full of curiosity so that he is in to EVERYTHING! When he was younger we were exhausted just trying to keep up with him.. It was also exhausting, but exciting to teach him about his world.

I'm delighted to say Andrew has far exceeded our early expectations. Fortunately he had enough vision to allow him to be a visual learner and attend Deaf Education programs where he has done well. People who see him think nothing is wrong with his vision until they really look. I would encourage you to use the term deaf-blind when it can get improved services for your children, and be careful in other situations; it is really scary for people. Learn to "work" the system and develop your skills to become "*respectfully demanding*".

## **PATTY IS A MIRACLE! SHE IS A GIFT!**

Bonnie Hagerty

*"When Patty was born we were told she would never walk, talk or be independent in any way"*

Patty (age 16) is developmentally delayed both medically and physically. She is about two years behind. Sometimes more, sometimes less. When Patty was born we were told-as everyone knows-she would never walk, talk or be independent in any way. They were wrong. I found right from the beginning that although doctors should listen to us, they often didn't. She was born full term. As soon as she entered the world I felt that I should do something, there was something wrong. I actually felt that she was born too early, or wasn't done developing yet but the doctors said it was just my emotions. They should have listened. I found the colobomas the next day. The doctors did the other tests, but apparently she came out fine and they sent us home. I still felt that something wasn't quite right. The next day at home after I fed her, my milk had just come in, and I gave her a bath. She just lay there, turned blue and went limp. My mother did CPR. The ambulance came, but on the way to the hospital she did it again, then again and again in the hospital. She ended up in Newborn ICU. It was a long night. They didn't think she was going to make it at first.

*"I realized Patty would live and if I held her close to my heart she would do her best to "be whatever she could be."*

After a while they told us to go home and get rest because we would need it. My world had turned upside down and I was walking without being inside my body. My mother took my husband and me home. On the way out of the hospital there was this statue of a mother holding a child. When I saw that statue a feeling came over me and I realized Patty would live and if I held her close to my heart she would do her best to "be whatever she could be." I still see and use that statue as comfort when she is in that hospital.

There were all these doctors coming and going who did not have anything at all positive to say. In my heart I knew they were wrong but had no way to show them. Patty didn't even hold her head up until immediately after her first choanal surgery around 9 months. All she would do is lay there. I do not really know if what I did that was right. I just went with what I felt she needed. I remember that as an infant, Patty needed to be wrapped tightly in her blanket, and held closely. Soft touches and caresses would make her distant and quiet. I held her often. I would also move her little arms and legs for her- again not softly but surely.

*"The first and absolute best thing that we did for her was to have early intervention right from the start"*

*"professionals spent most of their time working on her little body instead of what was going on within her little mind"*

*"I knew there was a mind and a will in there that had to be let out."*

Patty was such a sick little baby that professionals spent most of their time working on her little body instead of what was going on within her little mind. The first and absolute best thing that we did for her was to have early intervention right from the start. Our hospital identified her from the PICU and they came to the house once a week from the time she was sent home. When she was 10 months old I was already looking at the future for her. It was incredibly hard but we

sent her to a school for the disabled. Putting her on the bus broke my heart. I would spend much of the day in terror without her. But I saw the little gains she made here and there. She even learned to walk around age 2. I could see how determined this child was. Just when you would feel like giving up on her doing something, she would surprise you and accomplish the task. It just took her longer. But she was still like a porcelain doll. She seemed as though she would break sometimes. She had numerous illnesses and surgeries, but she also had this drive, this determined look in her eyes. I knew there was a mind and a will in there that had to be let out.

*"This was the first time that professionals listened to me"*

This first school was great for what she needed at the time. But there were children there with many different disabilities. I noticed she would model other behaviors and actions. I did not like some to the behaviors she was exhibiting. But at this school she learned how to grasp things and walk. She was not talking and they taught her some signs. That was good at first. She was able to communicate her wants and needs but somehow I felt that wasn't enough. Patty made guttural sounds only. As a matter of fact her voice box wasn't fully developed when she was born and it took a long time for her to make audible sounds. By the time she was 2 we knew she was hearing impaired. It seemed at the time she was making these guttural sounds as a way of trying to be verbal. The next BEST thing we did was to send her to a school for the hearing impaired. It was tough but she learned how to talk. It was an oral school that was intensive. This was the first time that professionals listened to me and worked with me and showed me how to help my daughter. That was SO important. They actually had extremely high standards for her. This was a first too. They pushed her to her highest level and yet they mixed that with love and compassion. They also made sure she was happy with herself. It seemed they found a joy to celebrate every day. Finally, Patty learned how to communicate! I feel that the educators in this school gave her life, to be part of the world we were in. They opened the door for her. She was still having her health problems but she was learning. They did not put limitations on her expectations. Without them I do not know where she would be now.

*"We were all a part of Patty!"*

I could not give my daughters much. Especially because with all the medical bills, we were now poor. But I could help give them an education. That is what I have continued to focus on. This school also provided all the other special needs, speech, hearing, PT, OT. We were all a part of Patty! There was one teacher and one aid to about 5 kids half of the day, and the other half there were "normal" children who these "hearing impaired" kids could model. This school was about an hour away from home.

*"From the beginning we have had to fight to keep "professionals" from limiting her"*

Our home district continued to "show us" how they could provide the same services for her. We brought her back to home district in kindergarten. (My father and grandfather had died that year and I was weak in fighting) They wanted to put her in the contained Special Ed classes but since at the other school she spent time in the Regular Ed classes there (it was a great program!), they did the same here. That's when the battles began. We have had to fight to keep her in Regular Ed. I feel it was their decision to bring her to our home district because it was cheaper for the town. I also believe they thought she would fail in Regular Ed. They probably felt they would show us they were right and we were wrong. They still to this day do not understand Patty. There were many wonderful educators who worked with Patty though. But from the beginning we have had to fight to keep "professionals" from limiting her.

*"Patty exhibited illnesses by behavior."*

Then the behavior aspect came in. We were often told about her negative behaviors. It was often that I had to tell them when her misbehavior increased she was getting ill. By now her kidney problems were diagnosed. When Patty's behavior was worse, I would take her to the doctor and she would have ear infections, bladder infections, phenomena, always something. Patty exhibited illnesses by behavior. I didn't allow misbehavior; I just tried to understand it. There were patterns.

*"They learned consistency was important for her"*

There were also times I would have to go in and tell them how to do things. At one point she was getting in trouble. I found out that one specialist would let her do something which another would reprimand her for. They learned consistency was important for her. So was modeling!!!! Patty continued to make gains but continued to be about two years behind. Again, it was a battle to keep her in Regular Ed but we did. I also discontinued intellectual testing because I felt there was no way they could accurately measure her on this. I still believe so. Again, this puts limitations on her. She has great gaps, especially in math. But she continues to thrive. And there continues to be modifications made which can overcome these gaps. Patty has continued in Regular Ed.

At one point I thought she had stopped growing intellectually. It was the same year she grew about 5 to 6 inches. I think her brain needed to rest while her body grew. Anyway, the special services she was receiving lessened. PT and OT were stopped, actually mostly on consult because they felt they had taken her as far as she could go. Preschool and early schools were good because they were children-based. By adolescence school is more curriculum-based. That gets harder. Still the fight was and continues to keep her in Regular Ed and to modify things to meet her needs.

*"Patty went from being social with everyone in elementary school to having very few friends."*

The greatest lack in her life has been her social needs. Adolescence is hard for anyone. Add having multiple disabilities and it gets harder. Patty went from being social with everyone in elementary school to having very few friends. Actually at one point she had no friends at all. It has been my large family that has kept her busy and feeling a part of society. She has been lonely, for years. Patty has problems with correct social behavior. She doesn't get the little nuances of language. She perseveres though. Middle school was extremely hard for her. And one must remember that there are few children in Regular Ed with as many needs and differences as she has. We now had to fight for almost everything; both medically and educationally. I went from being the concerned mom to being the, I guess you could call it "irritable" mom who thinks she knows more than they do. Well, I DO. There were all these specialist who "thought" they knew what they were doing with my daughter and what her needs were. Who was I, just her mom?

We have had no community involvement unless we put her into something ourselves. By this I mean she was in our town's sports program as a child and people included her because she was so cute. For example, we put her in basketball, the kids were great, and they would run and not run into her. She was tough. In softball they would just pitch and pitch to her until she hit the ball. The sports got to the point where it was beyond her abilities and became competitive and dangerous (around grade 2) so we had her involved in Special Olympics. She even did the torch run but then she wasn't "appropriate" for that either. I have put her in community theatre

once, again she was so cute, but now that she is older she auditions for it but she does not make it, yet her sister does. We tried the youth group in our church but because of the number of students in one room -teenagers- all talking at once she ends up alone and frustrated. There is nothing in our town that involves her outside of school so we have now given her guitar lessons. Children need to be involved in something, so we do it ourselves and always have. Otherwise, Patty is lonely or bored.

*“They took one look at her and said CHARGE.” “There were little Pattys everywhere”*

Then, Patty lost all hearing in one ear. I was desperate to find out why because I feared her losing her hearing all together. Months later we ended up in a geneticist office. They took one look at her and said CHARGE (Patty was 14 at the time!). That was the best thing that could ever have happened. Because of that diagnosis everything came together and started to make sense. We attended the CHARGE conference. Our daughter was no longer a misfit. Previously she didn't fit Regular Ed nor did she fit in Special Ed. No one was like her. Until then. There were a hundred babies and children there just like her. It was overpowering. There were little Pattys everywhere. Even the way these babies held their heads, rolled on the floor, were like Patty. It was spooky.

There were also a hundred parents just like us. I would look in the eyes of the parents of these babies and see the fear, longing and the hurt we had. I wanted to tell them it was all right. It gets better and they get older. But sometimes, that hasn't been true for everyone. I do not know why Patty has come so far. She has all the CHARGE characteristics and a few more. They call her mild but I honestly feel that is only because she has overcome so much and appears mild. They didn't see the sick little baby who I had to feed every two hours, took 45 minutes to feed and everything would come out her nose. This child has almost died on us many times yet here she is. She is our miracle. She continues to be. She also continues to be one tough cookie and is why I have so many gray hairs. I have had to be strong with her but I have also always just followed her lead.

*“These delays do not mean unable, just delay”*

With the CHARGE diagnosis there is finally an understanding about her delays. These delays do not mean unable, just delay. I do not think anyone should ever put limitations or labels on physical or mental expectations for our children with CHARGE. No one really knows what is inside them. You must let them reach their highest potential, whatever that is. I feel there are often higher possibilities than are ever said. With this CHARGE diagnosis we have been helped by professionals and other parents. We also try to help others. I get tons of information just from the parents alone. We also looked further at some medical aspects. She had her heart PDA surgery at age 13. I can't even think about how close she came. She had strep in her blood system twice. Had chicken pox in her blood system. We also got medical help that is more coordinated now instead of having one physical look at one thing while another looks at something else. We understand more now. We can embrace more in life. She continues to grow and seems to be closing some gaps. Others will always be there.

*“We now have medical professionals and professionals outside of our school district that understand CHARGE and all its impacts”*

There have been doctors and educators who still try to limit our daughters potential. It is a battle that we don't feel we should have to fight but we must. We now have medical professionals and professionals outside of our school district that understands CHARGE and all

its impacts. That is important and will help all the younger children. Patty's transition into life and adulthood is a battle for us. We have found that transitions are incredibly difficult for Patty. Some have been horrendous. The worse was when she has transitioned from one school to another with only one person having any experience with her, and her equipment (her FM unit) was malfunctioning too. You can go to meeting after meeting but when there are 10 specialists, then add other staff, meetings really don't do as much good as they are supposed to. They only hear the problems; not what Patty has really been about. After staff lacking experience with her, she has lacked experience of them. It was incredibly hard when she was younger to have different expectations from different people. This included her daily schedule. She also had a hard time transitioning with different schools. It takes her a while to get into the swing of things, when younger I would even dare to say almost half a year. Setting up for these transitions have been minimal after around 4th grade. It would have been wonderful if she could have attended the school she was transitioning to 9or even the next grade) while classes were going on, experiencing it. And this should be done more than once. It would have been good to have her and her educators meet, and more than once.

*"This transition will be the hardest."*

I have to say the transition into adulthood is where we are at and is what terrifies me the most. Again, finally we have brought in specialists to teach those who work with her about CHARGE and Patty's unique qualities. They have even started a Futures Personal Plan. Then they leave, then what-nothing. One of the problems is I have different expectations of and for than some specialists" in our district. I almost feel like they think I do not understand my child. They don't understand her. The transition planning should happen well before age 14, but we did not do that. We are now battling this. If nothing changes Patty will have a terrible time transitioning to adulthood. She needs to "experience" more. She needs more experience socially, vocationally. She needs technology to be independent. She needs someone to help her work to her goals, yet not limit them. She needs to be able to reach for the stars. She needs compassion and understanding, which is something she almost always gives but seldom receives. This transition will be the hardest.

We fight limitations and want her to be able to be the best she can be. We finally have people who understand this. She cannot go further without them. They will probably be with her for the rest of her entire future. But the main point is that she has a future. I, and now finally there are others, will do our best to let that future be healthy, happy and fulfilling for her. We have great dreams for her. It is not an easy task but one that is well worth every moment. Patty is a miracle. She is a gift.





## PRIORITIES FOR A CHILD WITH CHARGE

Cathy Lyle, M.Ed.

Educational Consultant to the Minnesota DeafBlind Project

What is most important?

1. Oxygen and food.
2. COMMUNICATION AND BONDING.
3. Everything else.

### COMMENTS

by Sandra L. H. Davenport, M.D.

While medical issues occupy the energies and thoughts of the parents (and the child, but usually to a lesser extent) during the first two years of life, communication and bonding are, in fact, the keys to helping the child become a happy and independent adult. Therefore, the "cares" (medical procedures like suctioning) need to become *routine* and incidental to the learning process. The flow of conversation and learning should be interrupted as little as possible. If nurses or personal care attendants are present during the home schooling hours or are at school with the child, they should get clear instructions from the physician about how much intervention is absolutely critical.

Too often I see sudden interruptions of the teaching process for suctioning or other procedures that happen without warning to either the child or the teaching staff. Since medical personnel are an integral part of the child's life, they need to learn not only how to communicate with the child that something is going to happen, but also to help enhance the communication process. *In effect, they should become teachers, too.*

My bias is that parents, teachers, medical staff, and others need to work on  
communication, communication, communication.

When they are done with that, the next step is to work on  
communication, communication, communication.

After that, they need to work on  
communication, communication, communication.

Without a formal system of communication, the child will have a hard time making you understand what he or she wants and you will have a hard time getting across what you want. *For a DeafBlind person, access to communication is a lifelong issue.* The earlier good communication starts and the more people that develop good communication with the child, the happier and more successful child (and parents, etc.) will be.



# INTRODUCTION TO MEDICAL ASPECTS AND MANAGEMENT OF CHARGE SYNDROME:

## **What this section contains, and how to use it.**

Meg Hefner, M.S.

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The Medical section of the CHARGE Syndrome Management Manual is divided into multiple sub-sections according to areas of the body. We intend for these to be useful both from the point of view of the families and of the professionals who work with your children. Each sub-section is has two parts: a DOCTOR section and a PARENT section. ***You may want to make copies of the DOCTOR pages for the specialists working with your child.*** Also make copies of the **OVERVIEW** page (just past the Visit Pad info at the front of this section) to give to each specialist. DO NOT GIVE OUT THE ORIGINALS - KEEP THOSE IN THE MANUAL

**DOCTOR section:** Each section contains a short summary intended for physicians. As you may already know, most specialists do not want or need to know everything about CHARGE. They are interested in the features of CHARGE within their specialty and other features which may have an impact on how the child is treated. We have attempted to cover the range of severity of features found in CHARGE, how they may be different in CHARGE than in other children, and other factors which may need to be considered, including cautions about care for children with CHARGE. For example, it is important for the cardiologist to know about tracheomalacia and anesthesia risks before planning surgery. The DOCTOR sections are written in standard medical jargon and include medical references and contact information for the author. We have tried to include the medical terms in the **glossary**.

**PARENT section:** In each PARENT section, we have tried to start out by describing the normal structure and function of the area of the body covered. Then we list some of the most common problems or abnormalities often seen in CHARGE, how the problems may be diagnosed, what tests may be done, and some of the management issues. Combined effects of multiple problems may be discussed or you may be referred to another section for the discussion.

### **AUTHOR CONTACT INFORMATION AND COPYRIGHT NOTE:**

Authors' names and contact information are on the top of each section (physician and parent). All pages are dated, so you will have an idea if the information may be incomplete or outdated. The CHARGE Syndrome Foundation, Inc. has retained copyright on all pages of the Manual. Please feel free to photocopy pages for use by anyone involved with your child. Contact the Foundation for permission before using the information in any publication, including web pages or other Internet uses.

## **MEDICAL VISIT PAD / RECORD KEEPING**

The typical child with CHARGE is followed by an average of 17 different medical specialists and will have more than 20 surgical procedures before he or she is 10 years old. Although the ideal would be to have a Patient Care Coordinator for your child, in reality it is the parents (most often the mother) who take on this role. Organized record keeping will help keep track of what is going on, aid in coordination of appointments, and facilitate communication among all concerned (parents, physicians, and other specialists).

Organization of Medical Records is an important skill for you to learn, if you haven't already. To help with this, we have designed a Medical Visit sheet. Your Manual includes a separate pad containing about 50 of these sheets. Take a moment to look this over. The purpose of the visit pad is to help you keep track of office visits. An example of a completed sheet follows this description.

The top of each sheet has a place for your child's name and date of birth. The date of the appointment, name of the doctor being seen and his or her specialty should be completed, as well as where (name of hospital or clinic) the visit took place. Before each appointment, think about what you hope to get out of it. Jot down any questions you have on the upper section. During the visit, be sure your questions are addressed.

Keep the pad with you during your visit and make notes about what you are told. You may want to ask the doctor to write a short summary on the sheet for you. If any tests are done at the visit, make a note of them and when you should expect to hear any results or findings from the tests. Be sure write down your next appointment and any referrals made by this doctor.

The sheets have holes punched in them so you can keep them in a three-ring binder. You may want to keep your child's records/notes in your Manual or establish a separate binder for records. You may decide to keep all records in chronological order, or establish separate sections for each specialist (see also the Minnesota Developmental Timeline in Development section).

Some parents have found it helpful to keep a master list where they list every visit (and every hospitalization or procedure) in chronological order. You will be amazed at the number of appointments and the variety of specialists you and your child see. Keeping such a list can also help coordinate appointments and even medical procedures. For example, if your child needs an other set of PE tubes in her ears, the ophthalmologist may want to take advantage of her being under anesthesia to get a better look at her retina at the same time. This will take advantage of one general anesthesia to perform multiple tasks.

**CHARGE Syndrome Medical Visit****Child****Birth Date**

<b>Date of Visit:</b>	<b>Next Visit:</b>	<b>Time:</b>
<b>Specialist:</b>	<b>Referral to:</b>	
<b>Specialty:</b>	<b>Date/Time:</b>	
<b>Place:</b>	<b>Place:</b>	
<b>Our questions: what we would like to discuss at this visit:</b>		
<b>Information discussed at this visit:</b>		

\_\_\_\_\_ APPOINTMENTS from \_\_\_\_/\_\_\_\_/\_\_\_\_ to \_\_\_\_/\_\_\_\_/\_\_\_\_

Date	MD	Specialty	What was done	RTC

\*Return To Clinic, e.g. 3 mo, 6 mo, 1 yr

# OVERVIEW OF CHARGE SYNDROME FOR PHYSICIANS

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## DIAGNOSIS & MANAGEMENT

The acronym CHARGE was proposed in 1981 by Pagon, et al.<sup>1</sup> Since then much has been learned, particularly about the influence of cranial nerve anomalies on major medical crises and long-term morbidity in CHARGE syndrome. Revised diagnostic criteria were published in a review for physicians in 1998.<sup>2</sup> Morbidity can be greatly diminished by understanding the interaction of these multiple anomalies. In particular, involvement of cranial nerves IX and X cause neurologic impairment of swallowing, leading to reflux and recurrent aspiration pneumonia. Addressing this problem could prevent many hospitalizations. When failure to thrive is present, one needs to take the swallowing difficulty into account along with possible lack of olfaction and the many other anomalies like heart disease which traditionally cause poor nutrition.

Each affected system is addressed in this CHARGE Parent Manual with particular caveats related to each specialty.

## DEVELOPMENT

Older literature states that most, if not all, of these children are mentally retarded. ***That is not true.*** A majority of children with CHARGE have vision and/or hearing loss, which together can constitute the designation of “deafblindness” even when residual hearing and vision is present. Children who are deafblind are “**input-impaired,**” which means they need to learn alternative modes of communication and different ways to explore and interact with their environments. In addition many children have Mondini malformations which lead not only to hearing loss but also to vestibular dysfunction. The developmental effects are unusual motor milestones including a “five-point crawl” and delayed age of walking. Without congenital vision deficits and other anomalies, children with vestibular dysfunction (e.g. Usher Type I) walk on average at 18-24 months. Children with CHARGE who are multiply affected walk at 3-4 years. Clearly any infant who has delayed motor milestones, does not speak, and may not look you in the eye because of a macular coloboma will be considered mentally retarded. However, measuring output in such cases does not necessarily reflect true mental processing.

As more children are put into appropriate deafblind programs with adequate input, much improved output is being observed. Early diagnosis of vision and hearing loss is important but *referral to appropriate educational programming is vital.*

## FINDINGS DESCRIBED BY THE ACRONYM

### **C- Coloboma (ocular)**

Cleft affects the globe but not the eyelid. The coloboma can involve the iris, retina (with or without involving the macula) or disc. An upper visual field cut can range from insignificant to major.

Microphthalmos or even anophthalmos can be part of the coloboma spectrum.

### **C- Cranial nerve anomalies**

I – the olfactory nerve may be involved with arhinencephaly. Lack of smell can have a major impact on feeding and, later, on socialization.

II – the ocular nerve is usually involved only if a coloboma involves the disc

VII - facial palsy is usually unilateral and present at birth. Facial asymmetry without facial palsy can also be seen as can asymmetric crying facies.

VIII – the acoustic nerve may be involved separate from malformations of the ear itself

IX & X – the major early problems poor or incoordinated swallowing with gastroesophageal reflux and aspiration pneumonia. These tend to improve over weeks, months or years.

#### **H - Heart malformations**

Any of the common types may be involved but tend to be of the conotruncal variety.

Vascular rings and aberrant subclavian arteries may cause tracheal compression.

**A - Atresia or stenosis of choanae** UL or BL, bony or membranous

#### **R - Retardation of growth and/or development**

Height and weight are usually normal at birth. Loss of growth milestones in the first two years is often associated with failure to thrive due to swallowing problems, heart disease, hospitalizations and recurrent illnesses.

Growth hormone deficiency may be present.

Developmental delay is due to many causes, the most important of which are vision and hearing loss combined with vestibular dysfunction due to anomalies of the inner ear.

Acute medical illnesses and hospitalizations also contribute to the delays. Mental processing can be normal.

#### **G – Genitourinary anomalies**

Male genital anomalies include small penis, hypospadias, undescended testes

Female genital anomalies include small labia

Hypothalamic hypogonadism may account for the genital anomalies and delayed or absent puberty

Genitourinary problems also involve malformations of the kidneys and ureters (tubes to the bladder)

#### **E - Ears anomalies: outer, middle, inner**

Mixed hearing loss +/- vestibular dysfunction

Canals may be narrow

Acute and chronic otitis media is common

Anomalies of the middle ear ossicles may be present

Mondini defects vary in severity

### **OTHER FINDINGS**

Floppy cartilage in ears and trachea

TE (tracheoesophageal) fistula

Esophageal atresia

Cleft lip/palate

DiGeorge sequence with poor immune response

High pain threshold

Resistance to some forms of anesthesia

### **REFERENCES:**

<sup>1</sup> Pagon RA, Graham JM, Zonana J, Young SL. Congenital heart disease and choanal atresia with multiple anomalies. J Pediatr 1981;99:223-227.

<sup>2</sup> Blake KD, Davenport SLH, Hall BD et al. CHARGE association: an update and review for the primary pediatrician. Clin Pediatr 1998;37:159-174.



## DIAGNOSIS, GENETICS AND PRENATAL DIAGNOSIS IN CHARGE

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### HISTORY OF CHARGE ASSOCIATION AND CHARGE SYNDROME

The collection of features which came to be known as CHARGE was first recognized by Dr. Bryan Hall, who has been collecting information about choanal atresia and related anomalies since 1968. He saw a pattern emerging in children with choanal atresia and multiple anomalies and first published his findings in 1979. In 1981, Drs. Pagon, Graham, Zonana, and Young published a series of patients with similar findings and coined the acronym "CHARGE" as an easy way to remember the condition. The name is easy to remember and catchy. Unfortunately, it does not adequately cover some of the most important features seen in CHARGE syndrome.

Many physicians are still using the 1981 criteria (below) to make or rule out the diagnosis. Revised diagnostic guidelines were published by the CHARGE Syndrome Foundation Medical Advisory Board in 1998 (Blake, et al.) The patterns of defects in CHARGE syndrome can still be difficult to diagnose, even by specialists.

### Syndrome or Association?

A "syndrome" is a recognizable pattern of birth defects or malformations, typically with one recognized cause (single gene or chromosome abnormality, for example). An "association" is a nonrandom collection of birth defects which is less specific than a syndrome. Until the cause(s) of CHARGE are identified, the debate about CHARGE syndrome vs. association is likely to continue in the medical genetics community. Those of us who have been most closely involved with CHARGE over the years (and some others, see Lubinsky ) feel "syndrome" is a better fit for CHARGE than "association." But don't sweat it, the difference is largely semantic.

As with any condition, the most involved, most severely affected cases are more likely to come to medical attention and be diagnosed most easily. This means the severity of the condition may be over-estimated and the frequency underestimated (because milder cases are not yet recognized and counted). As we get better at diagnosing the milder cases, we must also revise the incidence and prognosis to better fit the entire spectrum.

### ORIGINAL "CHARGE" FEATURES (1981)

- C - Coloboma of the eye
- H - Heart defects
- A - Atresia of the choanae
- R - Retardation of growth and/or development
- G - Genitourinary anomalies
- E - Ear anomalies and/or deafness

The diagnostic criteria set out in 1981 required that 4 of the 6 "CHARGE" features be present to make a definite diagnosis. However, even the 1981 paper which coined the term "CHARGE" recognized these criteria were preliminary and that the acronym did not cover all the significant findings (e.g. facial palsy) in these children.

## REVISED CHARGE DIAGNOSTIC CRITERIA (1998)

The revised diagnostic criteria take into account the fact that there are several features which are extremely common in CHARGE but very rare in other conditions (Major Diagnostic Criteria, Table 1) and features which are common in CHARGE but are also seen in several other conditions (Minor Diagnostic Criteria, Table 2). In addition, children with CHARGE may have a variety of other features (Common Findings, Table 3) which may not be particularly helpful in making a diagnosis, but which can mean a lot to the family and medical community in terms of management. All of these features are described in more detail in following Medical sections.

A diagnosis of CHARGE should be considered in any newborn with any "Major" criterion (coloboma, choanal atresia, facial palsy, or classic CHARGE ear) in combination with any other significant birth defect. See Differential Diagnosis for overlapping syndromes.

As of this writing (2002), there is no specific test that can be done which will definitively diagnose or rule out CHARGE. CHARGE syndrome is a clinical diagnosis. That is, it is made based on physical findings along with the best judgment of the Medical Geneticist. A diagnosis of CHARGE should be made or confirmed by a Medical Geneticist who is familiar with CHARGE.

### CHARGE SYNDROME: MAJOR DIAGNOSTIC CRITERIA

Features seen commonly in CHARGE, rarely in other conditions

CRITERION	INCLUDES	FREQUENCY
Coloboma	Coloboma of iris, retina, choroid, or disc Microphthalmia, anophthalmia	80 - 90%
Choanal Atresia	Unilateral (UL) or bilateral (BL); Bony or membranous; Stenosis or atresia	50 - 60%
Cranial Nerve Dysfunction	I: lack of smell VII: facial palsy (UL or BL) VIII: sensorineural hearing loss or vestibular problems IX/X: swallowing dysfunction	Frequent 40%+ 70-85% 70 - 90%
Characteristic CHARGE Ear ++	External Ear: Short, wide ear with little or no lobe, snapped off helix, prominent antihelix discontinuous with tragus, triangular concha, decreased cartilage, asymmetric, often protruding laterally  Middle ear: abnormalities of stapes, absent stapedius tendon, cochlear anomalies	90%  ?90%

Tables modified from Blake, et al., 1998, with permission

++The external ear abnormalities can be so specific as to suggest a diagnosis of CHARGE based on the ears alone.

\*\*\* see glossary and/or other medical sections for definitions of medical terms and diagrams

## CHARGE SYNDROME: MINOR DIAGNOSTIC CRITERIA:

Features less specific to CHARGE and/or not consistent enough to be considered major

CRITERION	INCLUDES	FREQUENCY
Characteristic CHARGE face	Square face, broad prominent forehead, arched eyebrows, large eyes, occasional ptosis, prominent nasal bridge with square root, small nares, prominent nasal columella, flat midface, small mouth, occasional small chin; larger chin with age. Facial asymmetry even without facial palsy	> 50%
Characteristic CHARGE hand	Small thumb, broad palm with "hockey-stick" palmar crease, short fingers	50%
Genital hypoplasia	Males: micropenis, cryptorchidism Females: small labia Both: delayed or incomplete pubertal development	70 - 85% Frequent ?50%
Congenital heart defects	Most common: tetralogy of Fallot, VSD, AV canal, aortic arch anomalies	70 - 85% have CHD
Cleft palate or Cleft lip	Unilateral or bilateral cleft lip +/- cleft palate Isolated cleft palate, including submucous cleft palate Can even occur with choanal atresia or stenosis	20 - 30%
TEF	Tracheo-esophageal atresia or fistula Esophageal atresia	20% 15%
Middle ear	Frequent ear infections Many sets of PE tubes	>80%
Hypotonia	Upper body hypotonia, sloping shoulders	Frequent
Renal anomalies	Hydronephrosis or reflux; Horseshoe kidney; Small or absent kidney	40%
Growth deficiency	Short stature Growth hormone deficiency	Common Rare

Tables modified from Blake, et al., 1998, with permission

**CHARGE SYNDROME: OTHER COMMON FINDINGS:**

May be important for management, but not very helpful in making diagnosis

FINDING	INCLUDES	FREQUENCY
Brain abnormalities	Microcephaly, Agenesis of corpus callosum, Dilated ventricles	Rare
Apnea	? central (brain)	Rare
Seizures		Rare
Laryngomalacia	Can result in weak cry	Frequent
Nipple anomalies	Extra, poorly formed or misplaced nipples	Rare
Floppy cartilage	Includes tracheomalacia, floppy ears	Frequent
Thymic or parathyroid hypoplasia	DiGeorge sequence without chromosome 22 deletion	Rare
Webbed neck	Often looks like sloping shoulders	Rare
Abdominal wall defects	Omphalocele Umbilical hernia	Rare 15%
Scoliosis	Younger children Older children	Common Frequent
Limb/skeletal anomalies	Absent thumb, Polydactyly (extra fingers), Split hand	Rare
Autistic-like behavior	Often noted in childhood	Occasional
Behavior problems	Often not noted until school age	Common

Tables modified from Blake, et al with permission

**REFERENCES**

1. Hall BD: Choanal atresia and associated multiple anomalies. J Pediatr. 1979(95)395-398
2. Pagon RA, Graham JM, Zonana J, Young SL: Congenital heart disease and choanal atresia with multiple anomalies. J Pediatr. 1981(99): 223-227
3. Blake KD, Davenport SLH, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JM: CHARGE Association: An update and review for the primary pediatrician. Clin Pediatr. 1998(37)159-174
4. Lubinsky MS: Properties of associations: identity, nature, and clinical criteria, with a commentary on why CHARGE and Goldenhar are not associations. Am J Med Genet 1994(1)21-25

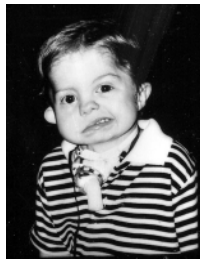
## CHARGE FACE



(1)



(2) iris  
coloboma



(3) R facial  
palsy

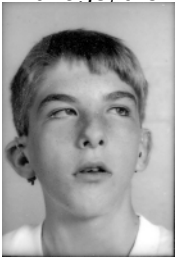


(4) R facial  
palsy



(5) BL facial  
palsy

Square, often asymmetric face, round eyes, flat cheekbones, wide nose with broad nasal bridge, and small chin. Unilateral facial palsy increases the asymmetry. Note that (3) and (4) are different children. With age, the face gets longer and the chin larger. (8) and (9) are the same child.



(6) teen



(7) teen



(11) **Hockey-stick palmar crease:**  
Upper crease on palm goes  
between index and middle fingers



(8) baby



(9) child



(10) teen

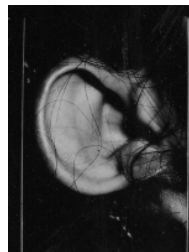
## CHARGE EARS



(12) R  
ear



(13) L  
ear



(14) R  
ear



(15) L  
ear



(16) R  
ear



(17) L  
ear

Right and left ear of three individuals. Floppy, small, wide ears with little or no lobe, often an unfolded or clipped-off appearance to the helix (outer fold), and a prominent antihelix creating a triangular concha (center of ear, esp. 17) The two ears usually are different.

## DIFFERENTIAL DIAGNOSIS: What else can look like CHARGE?

### **Chromosome abnormalities**

A variety of chromosome abnormalities can result in features which overlap with CHARGE. Most have different ear anomalies and facial features. The chromosome abnormalities which overlap with CHARGE may give us clues about where to look for an abnormal gene. See also the discussion below on VCF (22q deletion syndrome). Children with CHARGE would be expected to have normal chromosomes, including FISH for 22.

### **VCF (velocardiofacial) syndrome - the 22q deletion syndrome**

VCF had a number of names in the past, depending on the features seen in the individual child. Now that a test is available, we know that all of these “separate” syndromes have one cause, a microdeletion of chromosome 22 detected by a special chromosome test called FISH. Some of the terms that used to be used are: Shprintzen syndrome, VCF, DiGeorge and CATCH-22.

Features of VCF include

- Velo - cleft palate and occasionally cleft lip
- feeding difficulty – usually gets better in a few months
- Cardio - heart defects – exactly the same heart defects as seen in CHARGE
- Facial - typical face: long and slender, often with overfolded ears
- kidney abnormalities – similar to CHARGE
- small or absent thymus - leading to immune system problems
- hypocalcemia (low calcium), sometimes causing seizures
- learning disabilities
- psychiatric problems
- 22 - deletion of chromosome 22q11

"DiGeorge sequence" technically refers to children with heart defects in combination with thymus abnormalities, including low blood calcium levels. Although most common in VCF, DiGeorge sequence can also be found in children with CHARGE or as an isolated finding.

Velocardiofacial syndrome (VCF) can include DiGeorge sequence and other CHARGE-like features including palate problems, renal abnormalities, ear abnormalities and even occasionally colobomas and/or hearing loss. VCF is caused by a tiny missing piece (microdeletion) of chromosome #22. It can be confirmed by a special lab test called FISH (fluorescent in-situ hybridization) for deletion 22q11.2. Children with possible CHARGE should also have FISH for 22q to rule out VCF.

The heart defects and swallowing problems seen in VCF can be similar to those seen in CHARGE. However, the characteristic ears, face and hands are distinctly different (e.g. long and slender hands in VCF vs. short and broad in CHARGE; long face in VCF, square face in CHARGE). Only about 5% of children with CHARGE have DiGeorge sequence. Conversely, of all the children with DiGeorge sequence, about 85% have a chromosome 22 deletion, 5% have CHARGE and 10% have something else. To date, we are not aware of a single individual with definite CHARGE who had a FISH test which was positive for the 22q11 deletion.

## **VATER or VACTERL association**

VACTERL is an acronym for a collection of findings which overlap with CHARGE:

- V - vertebral (backbone, spine) anomalies
- A - anal atresia (referring to the anus, not the nose)
- C - cardiac (heart) defects
- TE - tracheoesophageal fistula or esophageal atresia
- R - renal (kidney) anomalies
- L - limb, especially lower arm bone anomalies

Vertebral anomalies, limb anomalies and anal atresia are each very common in VACTERL and rare in CHARGE. Children with VACTERL are unlikely to have any of the Major Diagnostic Criteria of CHARGE. Neither do they have the typical physical features (face, ears, hands) associated with CHARGE. In some cases, especially in the newborn period, VACTERL and CHARGE may be difficult to distinguish from each other due to overlapping birth defects.

## **WHAT TESTS TO DO?**

All individuals with a suspected diagnosis of CHARGE should be evaluated for chromosome anomalies, VACTERL and VCF, including chromosome analysis with FISH for the 22q11 VCF/DiGeorge locus. "FISH for 22q" is a specialized test which must be specifically requested separately from routine chromosome analysis.

## **PAX2**

PAX2 is a rare condition with features which overlap with CHARGE. Individuals with PAX2 abnormalities may have colobomas, renal (kidney) anomalies and hearing loss. They do not have the facial features or ear shape associated with CHARGE. A recent study showed that children with definite CHARGE syndrome do not have any mutations in the PAX2 gene.

## **Retinoic acid (Accutane)**

Accutane (a drug used to treat cystic acne) taken in the first two months of pregnancy has a very high chance of causing birth defects, including unusual ears and heart defects. The ear abnormalities seen in prenatal retinoic acid exposure can be very similar to the CHARGE ear. However, the other problems caused by retinoic acid are different. Accutane taken **before** pregnancy is not known to cause any birth defects. Retin-A skin creme does not cause birth defects.

## **WHAT DOESN'T CAUSE CHARGE**

It wasn't anything you did during the pregnancy. Other than retinoic acid, no specific pregnancy exposures have been linked to the features seen in CHARGE. Exposures to pesticides, smoking, alcohol use, and/or other drug use do not appear to play a role. Because the organs involved in CHARGE are developing over a number of weeks of pregnancy, it is unlikely that any one single event (car accident, food poisoning, whatever) could cause CHARGE. There is almost never any history of CHARGE or CHARGE-like features in other family members.

## **HOW OFTEN DOES CHARGE HAPPEN?**

The incidence of CHARGE at birth is estimated to be about 1 in 12,000 births. Many infants and young children with CHARGE do not survive due to the major medical complications. Many children with a milder expression of CHARGE may not be diagnosed until they are older or missed entirely. Therefore, the number of children with CHARGE in the general population is difficult to estimate.

## **HOW LONG DO CHILDREN WITH CHARGE LIVE - LIFE EXPECTANCY**

Life expectancy is decreased in children with CHARGE. Infants with CHARGE have complex medical problems and many of them do not survive. The highest mortality is in the first three years. Infants with bilateral choanal atresia, a complex heart defect, and/or tracheo-esophageal fistula appear to have the lowest survival rates and poorest outcomes. There is a relatively high post-operative mortality with CHARGE, possibly due to reactions to anesthesia and/or breathing or aspiration problems. The more surgeries (and anesthetics) a child must undergo, the greater the risks. Even beyond infancy, many children with CHARGE require multiple surgeries and are medically fragile.

Children with CHARGE are also at very high risk for aspiration - sucking food or liquid into the lungs (due to TE fistula, tracheomalacia, and swallowing problems), which often leads to pneumonia. There are reports in the medical literature of as many as 30-40% of children with CHARGE not surviving to five years. However, reports in the medical literature include children with the most severe cases of CHARGE. Less severely affected children are often not diagnosed in the newborn period, and may not be included in the "survival" numbers. Nevertheless, as many as 20-25% may not survive beyond two years of age despite our best efforts. Sometimes parents are faced with very difficult decisions about how aggressively to treat their seriously ill child. Such parents are not alone. The Foundation can put you in touch with others who have been through this process.

Beyond early childhood, the mortality remains higher than in children who do not have CHARGE. This may be due to a combination of factors, including residual heart defects, continued swallowing problems, anesthesia risks and general medical fragility. Because CHARGE is a relatively recently recognized syndrome, long-term life expectancy is unknown. There are many adults with CHARGE who appear to be in good health and have relatively few remaining medical concerns.



## RECURRENCE RISK

### WILL CHARGE HAPPEN AGAIN IF I HAVE ANOTHER CHILD?

For parents with one child with CHARGE, the recurrence risk is low, probably around 1-2%. There are only a handful of families with more than one child with CHARGE. The 1-2% recurrence risk is based on surveys of over 300 families with a child with CHARGE and extensive review of the medical literature. Most of the "familial" cases reported in the older medical literature probably would not be diagnosed as CHARGE today based on the revised criteria. Even using a very loose definition of CHARGE (i.e. anyone diagnosed as CHARGE, regardless of the expertise of the person making the diagnosis, the certainty of the diagnosis or the criteria used), the recurrence risk is still only about 1-2%. Using strict diagnostic criteria, the risk would probably be even lower.

### WILL CHARGE HAPPEN AGAIN TO ANYONE ELSE IN THE FAMILY?

Aunts, uncles and siblings of individuals with CHARGE probably are not at increased risk for having a child with CHARGE, although this information may change as we learn more about the cause of CHARGE.

Some individuals with CHARGE may be capable of having their own children when they become adults (many may not, due to hormone abnormalities). If CHARGE syndrome is caused by a single gene or chromosomal microdeletion, the risk to children of affected individuals could be as high as 50%. Information in this area may change very quickly, so be sure you have current information before making family planning decisions.

## FUTURE PREGNANCIES/PRENATAL DIAGNOSIS

The recurrence risk is low, but it is not zero. Parents are understandably worried about attempting another pregnancy and want to know what can be done to look for signs of CHARGE during a pregnancy. **There is nothing that can be done to prevent CHARGE.** However, as much reassurance as possible during the pregnancy that things appear to be going well is helpful.

The way to get the most information about CHARGE during a pregnancy is to have a directed ultrasound to look for features associated with CHARGE. This should be a **Level II ultrasound exam** and should be performed at a **tertiary care center** by an experienced ultrasound technologist using state-of-the-art equipment. This is not a procedure that can be done in the typical obstetrician's office. The ideal would be two ultrasound exams, the first at 18-20 weeks (post LMP) and the second about a month later, at 22-24 weeks. Take along the following checklist to give the sonographer the best information possible about what to look for.

## **ULTRASOUND EXAM FOR FEATURES OF CHARGE SYNDROME: THIS WILL NOT DIAGNOSE OR RULE OUT CHARGE**

The ultrasound evaluation should include a **complete standard anatomic survey** with particular attention to the following:

- Amniotic fluid measurement:  
Look for polyhydramnios (excess amniotic fluid) associated (especially in late pregnancy) with choanal atresia, esophageal atresia or poor swallowing.
- Cardiac evaluation:  
Many centers can do a formal fetal echocardiogram. Heart defects most common in CHARGE include tetralogy of Fallot with or without AV canal, and right-sided anomalies, including VSD and aortic arch anomalies.
- Kidney:  
Any kidney anomaly can be associated with CHARGE, including hydronephrosis (excess fluid in the kidneys), small or absent kidney, horseshoe kidney, posterior urethral valves.
- Brain:  
Dilated ventricles, absence of the corpus callosum or any other structural abnormality of the brain.
- Face: cleft lip or cleft palate
- Ear: abnormal shape or placement (take along a photo of CHARGE ears)
- Genitalia: small penis in a known male fetus

**Many abnormal findings would not be present early on and/or would be undetectable until later in pregnancy.** Don't be shy about asking how confident they are about the accuracy and completeness of the ultrasound exam. Every exam is different and none will detect every birth defect during pregnancy. The accuracy will depend on a number of things, including how far along you are, the position of the baby, your weight (the image is not as clear when it has to travel through a lot of maternal tissue before it reaches the fetus), the quality of the equipment, and the expertise of the sonographer. What can be seen one day may not be visible another day.

Remember, **none** of the major diagnostic criteria for CHARGE (coloboma, choanal atresia, cranial nerve abnormalities, characteristic CHARGE ear) can be definitively diagnosed by prenatal ultrasound exam. Although finding evidence of some problem or potential problem through the ultrasound exam would certainly raise the suspicion of CHARGE, and can help parents be prepared for that possibility, it is not diagnostic. And remember, **even a completely normal ultrasound exam cannot rule out CHARGE.**

# THE BRAIN IN CHARGE SYNDROME: FOR THE NEUROLOGIST

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## Brain anomalies seen in CHARGE

### Cranial nerve anomalies (70-90%)

- I - olfactory - (arhinencephaly in 42%)
- II - optic (colobomas in 80%)
- III, IV, V, VI rarely affected
- VII – facial palsy, UL or BL (43%)
- VIII - acoustic (60 - 80%+)
- IX, X glossopharyngeal/vagus (50%+)
- XI accessory - rarely affected
- XII hypoglossal - rarely affected

### Structural anomalies (less common)

- agenesis of the corpus callosum
- arhinencephaly
- holoprosencephaly
- hydrocephalus
- cerebral dysgenesis
- Mondini defect
- hindbrain defects
  - cerebellar hypoplasia
  - Dandy-Walker malformation

### Other

- seizures
- behavior abnormalities
- learning disabilities

Development: although it has been reported in the past that a majority of children with CHARGE are mentally retarded, this is not necessarily the case. It is extremely important for parents to realize that their child may have significant (even near-normal) potential, given accurate diagnosis and appropriate management of sensory deficits and other complications of CHARGE.

### Diagnostic tests/referrals

- MRI of brain
- CT
- EEG (if suspect seizures)
- Evoked potentials (vision/hearing)
- Other hearing tests (otolaryngology)
- Dilated eye exam (ophthalmology)
- Developmental testing (developmental pediatrics/deafblind specialist)

#### Medical treatment caveats

Possible unexpected reaction to anesthesia: Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.

Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.

Reflux, facial palsy and other cranial nerve anomalies may additionally compromise feeding in these children. There is a very high risk for repeated aspiration pneumonia.

If MRI is being considered, also consider a few more cuts to view the inner and middle ear, as there is a very high frequency of ossicular malformations and/or Mondini defect.

#### Non-medical management:

Cranial nerve IX, X, and VII anomalies can have an enormous impact on feeding. Feeding issues and common aspiration pneumonia are among the most important day-to-day issues for families. Often a feeding specialist can help the family cope with these issues.

#### References

Blake KD, Davenport SLH, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JM: CHARGE association: an update and review for the primary pediatrician. *Clinical Pediatrics* 1998;37:159-174.

Byerly K, Pauli RM: Cranial Nerve Abnormalities in CHARGE Association. *Am J Med Gen.* 1993; 45:751-757

Harvey AS, Leaper PM, Bankier A. CHARGE association: Clinical manifestations and developmental outcome. *Am, J Med Genet.* 1991; 39:48-55

Lin AE, Siebert JR, Graham JM: Central Nervous System Malformations in the CHARGE Association. *Am J Med Genet.* 1990; 37: 304-310.

## THE BRAIN IN CHARGE: PARENT INFORMATION

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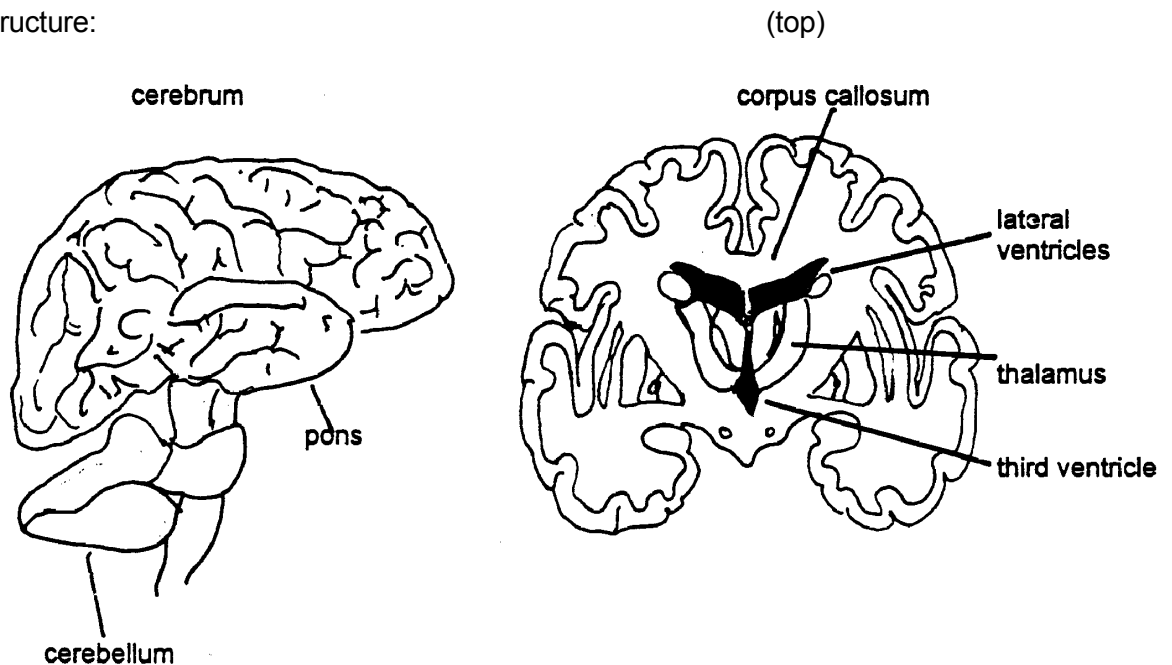
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### NORMAL BRAIN STRUCTURE AND FUNCTION:

Brain structure:



Cranial nerves: there are 12 cranial nerves, which begin in the brain and extend to structures in the head and neck. These nerves provide both motor control and sensation and include nerves involved in the senses of smell, taste, hearing, and vision, as well as movements of the eyes, face, tongue, palate, and swallowing. Some cranial nerves are also involved in the control of heart rate and movements of the gastrointestinal tract. Cranial nerve anomalies are very frequent in CHARGE.

## BRAIN ABNORMALITIES SEEN IN CHARGE

### Cranial Nerve Anomalies:

Anomaly	Frequency	Tests Used	Specialist
I – olfactory (smell)	? 40%	Clinical assessment	neurologist/otolaryngologist
II – optic nerve Coloboma	80-90%	Dilated exam Visual evoked responses	Ophthalmologist
VII – facial palsy	42%, usually one sided	Clinical assessment	neurologist
VIII – sensorineural hearing loss	60-80%	ABR Other hearing tests	pediatric audiologist otolaryngologist
IX, X swallowing and oromotor problems	50%+	Barium swallow Laryngoscopy	otolaryngologist OT, PT, speech pathology
XII – tongue	rare	Clinical assessment	neurologist

### Structural brain anomalies:

A variety of structural malformations of the brain have been reported in children with CHARGE. Pretty much any brain anomaly is consistent with CHARGE; none are extremely common. A neurologist may order brain imaging such as MRI or CT scan to look for possible structural brain anomalies. Because not all children have had imaging performed, it is not possible to list the frequency of each anomaly.

1. Arhinencephaly: absence of the olfactory lobes
2. Holoprosencephaly: abnormal formation of the cerebral hemispheres.
3. Agenesis of the corpus callosum: lack of fiber tract connecting the two hemispheres.
4. Cerebral dysgenesis: abnormal formation of the cerebral cortex.
5. Hydrocephalus: increased fluid in the ventricles of the brain

### Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG
2. Behavior abnormalities: See DEVELOPMENT sections
3. Learning disabilities: SEE Development section

## EFFECT OF ABNORMALITY ON CHILD

Cranial nerve abnormalities: cannot be surgically corrected. Only a few (hearing loss, swallowing problems) are amenable to management.

Olfactory nerve (I):

This nerve controls the sense of smell. If it is abnormal, the child's sense of smell (and therefore taste) will be absent or abnormal. This can complicate feeding problems.

Optic nerve (II):

A majority of children with CHARGE have colobomas. Optic nerve colobomas can severely affect vision. See EYE section.

Facial palsy (VII):

Droopiness or weakness of one (unilateral, UL) or both sides (bilateral, BL) of the face. This may affect feeding, with drooling on the affected side. If bilateral, there may be problems keeping food in the mouth. Facial palsy can have an effect on facial expression. In some cases, the eye on the affected side may not close completely and need to be kept moist with artificial tears. In some cases, facial nerve palsies get better over time. See FEEDING section.

Acoustic nerve (VIII):

Abnormalities of cranial nerve VIII will result in sensorineural hearing loss (see EARS AND HEARING).

Glossopharyngeal and vagus nerves (IX and X):

These nerves are involved in coordination of suck and swallow and with some of the mouth movements involved in speech. Feeding and swallowing problems are very common in children with CHARGE (see FEEDING). Many of these problems get better over time (often years), due to a combination of maturation of the nerve and use of appropriate therapies.

Structural brain abnormalities: most are not treatable

1. Arhinencephaly will cause lack of smell. This can be difficult to diagnose, but has been described very often at autopsy in CHARGE.
2. Holoprosencephaly is a serious brain abnormality, which is likely to result in significant mental retardation.
3. Agenesis of the corpus callosum is often (but not always) associated with learning disabilities and/or mental retardation.
4. Hydrocephalus can be associated with learning disabilities or mental retardation. Hydrocephalus is treated by placing a shunt.
5. Cerebral dysgenesis: many children with CHARGE have structural brain abnormalities noted on MRI or CT scan. The significance in terms of functioning is not always clear.

Other brain abnormalities described in CHARGE

1. Seizures: diagnosed by EEG, often effectively treated with medication
2. Behavior abnormalities: See DEVELOPMENT sections.





# THE INFLUENCE OF SENSORY LOSS ON DEVELOPMENT

*See also the Manual sections on Development*

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## INPUT IMPAIRMENT

In terms of intellectual development, the most important anomalies in CHARGE involve the eyes and ears. Vision and hearing are the most important “distance” senses. Some people have estimated that 70% of what we learn is through our vision and yet language develops most quickly with hearing unless the child is born into a signing environment. The combination of hearing and vision loss has been called “dual sensory impairment” or, more simply, “deafblindness.” I also like to use the term “input impaired.” Most measures of intelligence rely on output, i.e. how well the children use their hands, walk, talk, etc. From these outputs, an estimate of mental processing is reached. However, if input is inadequate, how can processing and output be optimal? As a result, the intelligence of children who are deafblind is routinely underestimated. While some children with CHARGE may have brain malformations which limit intelligence, *I believe the majority of these children have normal or near-normal abilities to process input.* However, that input has to be presented in a manner that the children can understand.

## DETERMINING THE COMMUNICATION BUBBLE

Programs for those individuals who are blind/visually impaired usually count on hearing as a compensatory skill whereas those for the deaf/hard-of-hearing use vision. What happens if both are limited? Different approaches to learning must be found. Finding a program with at least some staff who understand dual sensory loss will be vital for the optimal development of these children. These staff can then teach those with more traditional backgrounds to adapt their programs. Using the sensory inventory to be published in a manual for interveners<sup>1</sup> can be useful for families and educators in determining the “communication bubble.” This bubble is the area within which a child can comfortably communicate with another person or get input from the environment. The area will be different for each of the five senses: vision, hearing, smell, touch and taste.

## VISION (also see section on Eyes)

If a coloboma is present just behind the iris, that section of retina will not function properly, creating a blind spot in the child’s vision above the eyes. This is normally small and does not interfere much with seeing. If the cleft is large and involves a significant portion of the lower retina, then the blind spot may also be large. That child will not be able to see anything above the level, for instance, of the top of the eyeglasses. He/she will have to look up or tilt his/her head up in order to see what is up there. More significant is a cleft involving either the optic disk, which is the nerve coming into the eyeball, or the macula, which controls central vision. In either of these cases, the child may not be able to see objects clearly and distinctly no matter what kind of glasses are used.

Many children with CHARGE have different vision in each eye. In addition, some of them may have amblyopia. This occurs when the vision in one eye is better than in the other or when the eye muscles are weak on one side so the child cannot focus well using both eyes. The brain suppresses the image from the poorer eye so they do not have to contend with double vision. Sometimes the children have eyes that are of unequal size as well. A smaller eye usually means that the cleft inside is larger or more significant; the vision on that side is usually worse.

The effect of these anomalies is an upper visual field cut and/or uncorrectable blurry vision. Such a visual field cut means the child will bump into object above him/her such as tables or open cupboard doors, or will have to tip the head back in order to look up at an adult (this can be hazardous if balance is not good).

If the macula is involved, the central vision may be very blurry or even absent. With central vision loss, the child will look above the target. For instance, in order to see a person's eyes, he/she would look at the forehead or hairline. Such children may be labeled autistic because they do not appear to make eye contact in addition to having poor communication skills because of deafness.

*A vision consultant can help adapt materials to fit the vision needs. An orientation and mobility specialist may be needed to help the child move safely about the environment.*

**HEARING** - Please see the section on Hearing in CHARGE for more details.

The hearing losses in CHARGE can be anything from mild to profound. Most children with profound hearing loss, however, do have better residual hearing than is recognized during the early hearing evaluations. Recent evidence shows that any kind of language program (oral/aural or sign) initiated for a deaf/hard-of-hearing infant within the first six months of life can lead to significantly improved language development.<sup>2</sup> This is clinical confirmation of the basic science evidence that the brain pathways for hearing (as well as other sensory modalities) do not develop if the input is not presented while the brain is developing.

## **BALANCE**

When the vestibule of the inner ear is involved, the children are born without the balance sense that comes from the inner ear. This balance sense tells the child where his/her head is in space: up down, tipped sideways, or in a diagonal direction. When this sense is not present at birth, the child feels unsteady as he/she raises the head. If that same child does not see very well, there is very little motivation to raise the head. Therefore, these children have very delayed gross motor development. They prefer to keep their bodies flat on the ground or in a stable condition. They may roll to where they want to be, combat crawl, crawl with their head down on the ground (a 5-point crawl), or sit and bounce forward or backwards to where they want to be. When they get up and walk, they hang on for longer periods of time and they also keep their feet wider apart. When both vision and hearing are affected, the average age of walking in CHARGE is 3-4 years. Unsteadiness may persist into adulthood if vision is significantly impaired. The CHARGE Syndrome Foundation has a videotape of children with inner ear balance problems which shows gross motor development from infants to young adults. Children learn to trust their muscles and joints and do learn to walk and run, though on a delayed schedule.

## **TONE**

Many children are also reported to have low muscle tone (hypotonia). In some cases, the upper body is more involved than the lower body. This has not been well studied. However, we think that the muscles themselves are normal and that the major problem is muscle control from the nervous system.

## **OTHER SENSES**

**Touch** becomes the major means of input if vision and hearing are limited. This can be with the hands, feet (preferred in some cases), face and tongue. Young children routinely mouth objects. This becomes socially unacceptable in older children and, yet, mouthing objects is a legitimate way of getting further information when other input is not available. Most children with CHARGE have a good sense of light touch. Pain sensation, however, is often altered. Many have very high thresholds for pain. Since they do not experience pain normally, they may inflict pain on others without realizing what that pain is. This can become a social problem.

**Smell** is frequently diminished or absent in CHARGE. The consequences are important for nutrition since smell constitutes the major part of flavor. When your nose is stuffed up from a cold, food does not taste as good. In addition, children may not understand why others move away from them when they pass gas or, as teenagers, when they take off their shoes or have just come in from vigorous exercise. They simply do not smell what others do.

**Taste** is probably normal but this has not been investigated. Taste buds detect only four qualities: salty and some sweet on the front of the tongue, sour on the sides and bitter in the back. Therefore, children without sense of smell may be more likely to prefer salty and spicy foods. Interestingly sweetness is not as prominent when smell is absent.

## **OTHER FACTORS**

The child who has spent many days, weeks, or even years in the hospital does not have the normal experiences developmentally. If he/she cannot hear well, see well, or balance properly, then all phases of development are delayed. This includes gross motor, language, personal-social skills, and even fine motor skills. Understanding what is happening in the eyes and ears is, therefore, a very important task, especially in the first year of life. This rarely happens, however, because the doctors and families are more concerned with the structural problems with the nose, the palate, the heart, the kidneys, the esophagus, etc. Filling out a Developmental Timeline can be helpful in showing the interaction of the many medical concerns with developmental milestones.<sup>3</sup>

## **SUMMARY**

In the long run, the measure of intelligence and the ability to develop into an independent and contributing adult depends on the level of communication that can be achieved. If a child cannot tell you how he/she thinks, you cannot measure his/her intelligence. Routinely, therefore, tests of intelligence conclude that children with CHARGE are retarded. In some cases, certainly, children have difficulty with cognition and could be called mentally retarded. In most cases, however, it is my contention that children are developmentally delayed to a very significant degree, but we cannot accurately measure what their cognition is. As we learn more

about adolescents and young adults, it appears that some of them have significant learning disabilities. Exactly what these learning disabilities are is just now being studied.

It is important to understand the interaction of the multiple anomalies present in CHARGE. From a functional point of view, it is crucial to find a way of establishing a formal communication system, both at home and at school, so these children can reach their maximum potentials.

## REFERENCES

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1. Davenport SLH, et al. Evaluation section in Understanding Deafblindness: Issues, Perspectives and Strategies. Developed by SKI-HI Institute, 6500 Old Main Hill, Logan UT 84322-6500. 435-797-5598. To be published by Hope, Inc. 1856 North, 1200 East, North Logan UT 84341 435-752-9533
2. Yoshinago-Itano C, et al. Language of Early- and Later-identified Children With Hearing Loss. Pediatrics 1998; 102:1161-1171.
3. Davenport SLH, Kloos E, Prouty S: The Minnesota Developmental Timeline. Requests to Dr. Davenport, 5801 Southwood Drive, Bloomington MN 55437-1739. 952-831-5522.

## THE EYES IN CHARGE: FOR THE OPHTHALMOLOGIST

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### OPHTHALMOLOGIC ANOMALIES SEEN IN CHARGE

Coloboma (80-90%)  
Iris  
Retina  
Optic nerve  
Microphthalmia  
Facial nerve palsy (40%, unilateral >>bilateral)  
Visual acuity abnormalities (90%)  
Strabismus or amblyopia (frequent)  
Ptosis  
Cataracts  
Retinal detachment  
Photophobia (frequent)

### DIAGNOSTIC TESTS

Dilated funduscopy examination

### MEDICAL CONSEQUENCES

- ❖ Colobomas of the iris typically do not affect visual acuity or visual field
- ❖ Colobomas of the retina cause visual field defects in the upper visual field. They also predispose the patient to retinal detachment.
- ❖ Colobomas of the macula and/or optic disk usually affect visual acuity significantly.
- ❖ Facial palsy can result in lack of blinking and resultant dry cornea which can lead to corneal scarring.

### MEDICAL MANAGEMENT WITH CAVEATS

- ❖ Accurate description of visual acuity and visual field are of paramount importance for educational and communication purposes, particularly since most children with CHARGE have mild to profound hearing loss as well.
- ❖ Glasses (spectacles) to correct refractive error
- ❖ Tinted glasses for photophobia
- ❖ Occlusive patching for treatment of amblyopia
- ❖ Surgery for strabismus, cataracts, retinal detachment, as appropriate
- ❖ Artificial tears or gel to treat corneal exposure associated with facial palsy
- ❖ Regular (yearly) ophthalmologic evaluations to assess changes in visual acuity, refractive error, and potential for retinal detachment. Parents should be informed of the risk of retinal detachment and the importance of immediate medical assessment if there is any change in the vision status of the child.

## NON-MEDICAL MANAGEMENT ISSUES

- ❖ A diagram of the visual fields should be given to families and vision teachers or therapists so that communication programming will be optimized. The visual field may be a crescent-shaped area of the lower visual field. Some children will tilt their heads back in order to compensate to see. Accurate assessment of the most comfortable head position for viewing objects is important.
- ❖ Low vision aids such as magnifying bars, televisions and binoculars may be helpful
- ❖ Many children with CHARGE are sensitive to bright lights. Sunglasses can be very helpful in making the child more comfortable.
- ❖ Often the child appears to see better than would be predicted based on results of formal acuity and visual field testing. Many children who are legally blind function quite well visually. The parents and teachers usually can provide an excellent description of what the child can see.
- ❖ Demonstrate for parents what the vision is with best correction to help them understand what the child can and cannot see. For instance, parents frequently misunderstand that children with high myopia can see object moving at a distance when lighting and contrast are adequate, but cannot see detail clearly. In this situation, parents often have the impression that the child is not significantly visually impaired, when that is not the case.
- ❖ Most children with CHARGE have multiple anomalies, especially hearing loss. Significant vision problems combined with facial palsy, deafness, and inability to communicate may result in autistic-like behavior. However, once vision and hearing have been accurately assessed and an appropriate communication pattern established, such behaviors are often extinguished. Evaluation by a deafblind specialist (not simply a low-vision specialist) is essential.

## REFERENCES

- Blake KD, Davenport SLH, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JG: CHARGE association: an update and review for the primary pediatrician. *Clinical Pediatrics* 1998; 37:159-174.
- Blake, K. D.; Russell-Eggitt, I. M.; Morgan, D. W.; Ratcliffe, J. M. and Wyse, R. K. H.: Who's in CHARGE? Multidisciplinary management of patients with CHARGE association. *Arch. Dis. Child.* 65: 217-223, 1990.
- Chestler RJ, France TD: Ocular findings in CHARGE Syndrome. *Ophthalmology* 95:1613-1619 (1988)
- Chestler RJ, France TD: Ocular findings in CHARGE Syndrome. *Ophthalmology* 95:1613-1619 (1988)
- Russell-Eggitt IM. Blake KD. Taylor DSI. Wyse RKH. The eye in the CHARGE Association. *Brit J Ophthal.* 1990; 74:421-426.

## THE EYES IN CHARGE: PARENT INFORMATION

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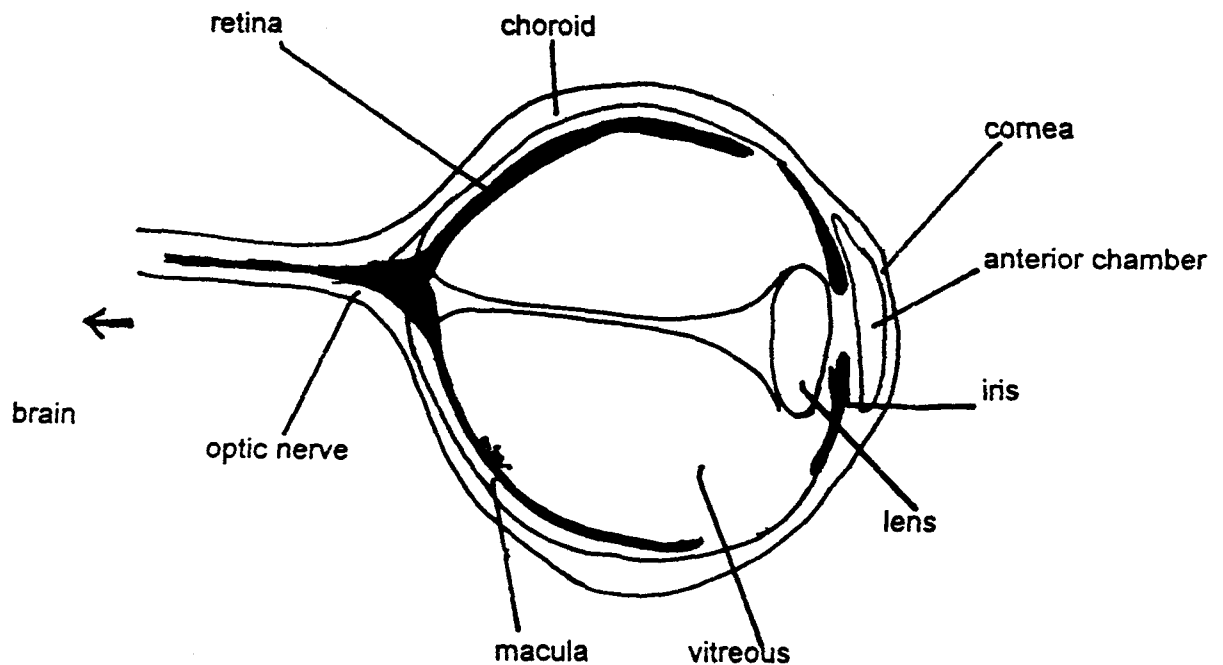
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### NORMAL STRUCTURE AND FUNCTION OF THE EYE

#### Structure: Parts of the eye



#### Function

Transmission of light

cornea

aqueous humor

lens

vitreous humor

Conversion of light to electricity: retina

Transmission of electrical signals to the brain:

optic nerve (cranial nerve II)

optic tract

Interpretation of electrical signals: occipital cortex and surrounding tissues (brain)

Problem List: eye problems seen in CHARGE

Problem	Test(s)	Specialist
Coloboma of the iris (keyhole pupil)	External examination	Ophthalmologist Pediatrician/Family physician
Coloboma of retina, optic nerve	Dilated eye examination	Pediatric ophthalmologist
Visual acuity (blurriness)	Eye charts or cards	Pediatric ophthalmologist
Visual field defects (blind spots)	Dilated eye examination Visual field testing	Pediatric ophthalmologist
Retinal detachment	Dilated eye examination	Ophthalmologist - retinal specialist
Corneal exposure secondary to facial palsy	External examination	Ophthalmologist Pediatrician
Cataracts	External examination	Ophthalmologist
Ptosis (droopy lids)	External examination	Ophthalmologist
Strabismus or amblyopia (weak eye)	External examination	Ophthalmologist

EFFECT OF PROBLEM ON CHILD

Iris coloboma:

This does not affect vision, but may make the child more sensitive to light (photophobia).

Retinal coloboma:

This will result in large blind spots, usually in the upper field of vision, (as if the child were wearing a baseball cap). Many children with retinal colobomas prefer to be upside down and to bottom-shuffle, in part because that way they can best make use of their available visual field. .

Coloboma of the macula or optic nerve:

This often results in blurry vision as well as large blind spots. Children with extensive colobomas are often legally blind (20/200 acuity or worse). They may not look directly at objects or establish eye contact because of poor central vision.

Microphthalmia (small eye)

This can be associated with large colobomas of the retina.

***Any coloboma of the retina or disk puts a child at increased risk of retinal detachment. Any sudden change in vision should be treated as a medical emergency.***

Strabismus

Loss of vision can result if not corrected.



## DEVELOPMENTAL EFFECTS OF VISION LOSS (also see Development sections)

Infants with decreased vision will have delayed motor milestones. This is especially true for children with CHARGE, who often also have hearing loss, vestibular (balance) abnormalities and serious medical problems requiring multiple hospitalizations and surgeries.

Communication can be complicated by vision problems in children with CHARGE. Because of the hearing loss, sign language, speech reading and other visual communication is often used. Decreased visual acuity can make this more of a challenge.

## MEDICAL TREATMENT OPTIONS

There is no medical or surgical treatment for coloboma

Retinal detachment can often be treated surgically if it is correctly diagnosed shortly after it occurs. Suspected retinal detachment should be regarded as a medical emergency.

Amblyopia may be treated with patching; strabismus may be treated with surgery.

Refractive errors that reduce visual acuity can often be helped with glasses.

## NONMEDICAL MANAGEMENT

Photophobia can be helped by using tinted glasses, indoors as well as outdoors.

It is important to know the extent of your child's visual field. If there is very little vision in the upper portion of the visual field, sign language and objects the child may want to see must be placed in the lower half of the visual field (in the lap). Many children compensate for small visual fields by adopting a certain head position or body position.

### Education

If your child has both hearing loss and vision loss (even if she or he is not "deaf" or "blind"), the educational team for your child should include a specialist in deaf-blind children. Such specialists exist in every state in the U.S. Input from such an expert is important even if the hearing loss or vision loss is not "complete."



# THE EARS IN CHARGE – FOR THE PHYSICIAN

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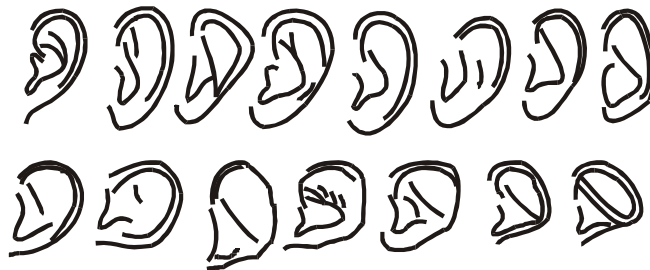
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## I. Outer ears & canals

### A. Distinctive shape of CHARGE Ears<sup>1</sup> (see diagram)

1. Ear shape can be so distinctive as to tip off the examiner to a diagnosis of CHARGE:

- Helical fold: thin, "clipped off" along the inferior edge or absent
- Antihelix: may extend out to the helical rim rather connecting smoothly to the antitragus
- Tragus: Usually intact
- Antitragus: Usually present or thinned but may not connect to antihelix to form the posterior rim of the concha
- Concha: Takes on a more triangular appearance as the antihelix extends farther out
- Lobules are frequently small or absent.



2. Lop ears are fairly common but in and of themselves do not constitute "CHARGE ears." When the ear is put into the normal position, look for the distinctive CHARGE features.

### B. Floppy ears due to soft cartilage

Typically, the pinnae flop forward or can be bent forward too easily even after the newborn period. The reason for the apparently abnormal cartilage is not known. It does firm up somewhat over months or years but even adults may have somewhat soft pinnae.

### C. Reconstruction

Taping back the ears during the few months of life can partially correct a lop-ear deformity.<sup>3</sup>

Though otoplasty can serve a cosmetic purpose in older children, it has been done primarily to provide better support for behind-the-ear hearing aids and eye glasses. However, the weight of the aids may push the ear back into the preoperative position. Additional means of securing the aids should be used until the cartilage is firm enough to support the weight.

### D. Canals & Tympanic Membranes

Significantly stenotic canals are rare in CHARGE. TMs appear normal unless change in position or shape reflects ossicular changes or severe Eustachian tube trouble.

## II. Middle ears

### A. Ossicular malformations

The possibility of ossicular malformations is frequently overlooked when chronic serous otitis media is present. Therefore, audiometry after PE tube placement is important. CT scan of the middle and inner ear is important in all cases where any hearing loss is present in order to rule out malformations.

### B. Chronic serous otitis media

CSOM is common with or without accompanying cleft palate. Because of the detrimental

effect any additional hearing loss has on development, *do not delay PE tube placement*.

### III. Inner ears

#### A. Mondini malformations

Significant malformations are common but much underappreciated. Early confirmation by CT scan is helpful for medical/habilitation prognosis and management.

#### B. Cochlear involvement

Hearing losses range from none to profound. Sensorineural loss is usually stable and aidable so management is the same as in other conditions. Cochlear implants have been done successfully and can provide at least environmental information for deafblind children. Keeping hearing aids on and working is very difficult. The reasons include not only the floppy cartilage mentioned above but also the frequent presence (90%) of impaired swallowing so that copious oronasal secretions run down the face of the supine child into the ear. The secretions may loosen the ear mold or block the air hole. In addition, the secretions may keep the canal moist setting up the conditions for chronic otitis externa.

#### C. Vestibular involvement

Congenital loss of vestibular function is probably the single most important factor in delayed gross motor milestones so early imaging of the inner ear is indicated.<sup>5</sup> This will decrease the need for extensive neurologic testing and falsely low estimations of intelligence IF the implications are communicated to the primary physician and neurologist.

Few tests other than tomography are available to confirm vestibular involvement in early childhood. Caloric testing may help though many children have PE tubes or TM perforations by the time this question comes up. Placing the baby in the mother's lap in a rotatory chair and comparing their eye movements is possible only if the child does not have a significant coloboma affecting the macula (central vision). Posturography is usually not possible until middle childhood because walking is often delayed to 4-5 years, visual fixation may be impaired by the colobomas, and the child will not cooperate if he/she does not clearly understand what to do.

#### D. Retrocochlear involvement

Very little firm data is known though some suggestion of central hearing loss has been made.<sup>2</sup> Since surrounding cranial nerves, such as VII, IX, X are clearly involved, it would make sense that the VIII nerve nuclei could be involved also. We know of no postmortem confirmation of this.

### IV. CAVEATS: What make CHARGE different from other syndromes?

Multiple cranial nerve involvements produce many ENT concerns: olfactory, facial, glossopharyngeal and vagus nerves may be involved. Choanal stenosis/atresia, cleft lip/palate and TE fistulas may also be present. The ENT consultant is an early and very important member of the team.

Dual sensory loss is present in over 3/4 of cases. Therefore, typical hearing habilitation measures may be inappropriate since they rely on good vision. INTELLIGENCE IS ROUTINELY UNDERESTIMATED in these children. Early identification of hearing and vision loss is often made without early referral to the appropriate education team, which should include educators familiar with dual sensory losses. This referral should be made as ASAP since communication is challenging to establish and yet is the key to assessing intelligence and optimizing quality of life.

## **V. BIBLIOGRAPHY**

1. Davenport SLH, Hefner MA, Thelin JW: CHARGE Syndrome. Part I. External ear anomalies. *Int'l J of Pediat ORL* 12:137-143, 1986
2. Thelin JW and Davenport SLH: CHARGE Syndrome. Part II. Hearing loss. *Int'l J of Pediat ORL* 12:145-164, 1986.
3. Matsuo K, Hayashi R, et al.: Nonsurgical correction of congenital auricular deformities. *Clin Plast Surg* 17(2):383-95 1990.
4. Shah UK, Ohlms LA, et al.: Otologic management in children with the CHARGE association. *Int'l J of Pediat ORL* 44:129-147, 1998.
5. Blake KD, Davenport SLH, et al.: CHARGE Association: An update and review for the primary pediatrician. *Clin Pediat* 38:159-174, 1998.
6. Tellier AL, Cormier-Daire V, et al. CHARGE Syndrome: Report of 47 Cases and Review. *Amer J Med Genet* 76:402-409, 1998.



## THE EARS AND HEARING IN CHARGE SYNDROME: PARENT INFORMATION

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Abnormalities of the ears, ear infections, and hearing loss are very common features in CHARGE. Together these problems can affect health and the ability to speak and learn. As with other aspects of CHARGE, not every individual has every problem. The number of problems each individual has and the severity of each problem varies greatly -- within individuals and across individuals. As a result, each individual's set of abilities and disabilities is unique.

### HEALTH CARE PROVIDERS FOR EARS AND HEARING

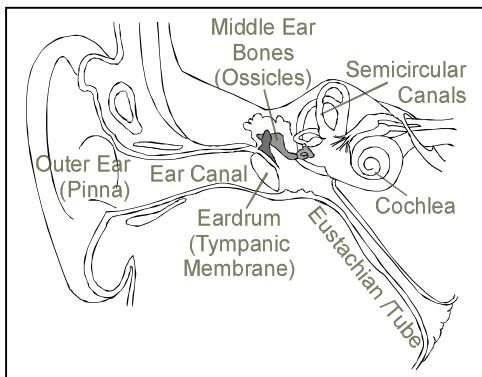
Medical care for ears and hearing in CHARGE is best provided by a physician who specializes in ears: an otologist, otolaryngologist, or ear-nose-and-throat (ENT) physician. Though ear problems in childhood are often diagnosed and treated by the primary medical care provider (family physician or pediatrician), the problems in CHARGE are so complex that they need to be diagnosed by an otolaryngologist who makes an otologic (ear) diagnosis and recommends a plan of treatment to the primary care provider.

Hearing is assessed by an audiologist. The results of the hearing assessment are used in the medical diagnosis of the hearing loss, in determining the success of medical/surgical treatment, and in developing an aural habilitation plan. This is a plan for overcoming hearing loss that cannot be corrected medically or surgically. It can include selection and fitting of hearing aids, speech-language evaluation/therapy, teaching of sign language, and placement in educational programs.

It is very difficult to assess hearing accurately on persons with CHARGE – especially infants and young children. In some cases, detailed hearing evaluation may require many test sessions. Until hearing is assessed with confidence, it is often difficult to establish the otologic diagnosis, to select hearing aids, or to make optimal educational recommendations.

Children with CHARGE need care from otolaryngologists and audiologists who specialize in the disorders of children and especially children with unusual problems. Many audiologic test facilities are not equipped to perform the types of tests needed for children with CHARGE. Selection of the appropriate otolaryngologist and audiologist is crucial to the accurate diagnosis and successful treatment of ear and hearing problems in CHARGE.

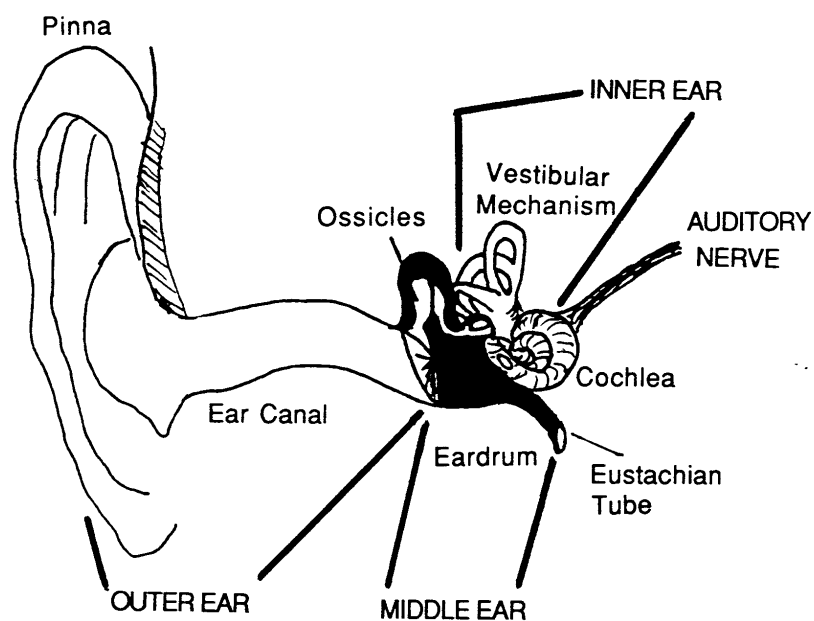
### EAR AND HEARING ABNORMALITIES IN CHARGE SYNDROME



The structures of the ear are shown in the diagram to the left, and on the following page. They are described on page 3.

**FIGURE : STRUCTURES OF THE AUDITORY SYSTEM**

**OUTER & MIDDLE EARS**  
*(Conductive Hearing Loss)*



**INNER EAR & AUDITORY NERVE**  
*(Sensorineural Hearing Loss)*



## **OUTER AND MIDDLE EARS**

The outer and middle ears conduct sound to the inner ear. Hearing loss due to problems in the outer or middle ears is called “CONDUCTIVE HEARING LOSS.” Conductive hearing loss only makes the sound that reaches the inner ear softer. Amplifying the sound overcomes the hearing loss by restoring loudness and clarity. Conductive loss is common in CHARGE, but it is usually only part of the total hearing loss.

### **Outer Ear (Pinna)**

Pinnae can be slightly or significantly deformed. Unless the ear canal is blocked by tissue, deformities of the pinna have almost no effect on hearing. However, if a hearing aid needs to be worn, the pinna may be too soft or not large enough to accommodate a behind-the-ear hearing aid that is commonly fitted on a child. Special devices may be required to keep the hearing aid in place. In some cases, pinna shape can be improved with surgery.

### **Ear Canal (External Auditory Canal)**

The ear canals in CHARGE can be narrow (stenotic). This does not usually affect hearing, but it may make it difficult to examine the eardrum and middle ear, to clean wax (cerumen) from the ear, and to fit a hearing aid earmold in the ear canal. If the ear canal is not present (atresia), it causes a large conductive hearing loss. In some cases, surgical construction of an ear canal may be attempted.

### **Middle Ear**

The middle ear is an air-filled space behind the eardrum (tympanic membrane). Sound moves the eardrum and the three tiny bones (ossicles) which transfer the motion to the inner ear. In CHARGE, it is common to have malformed ossicles that cause significant conductive hearing loss – especially for low pitch or low frequency sounds. Usually, no attempt will be made to correct this problem with surgery.

At the bottom of the middle ear cavity is the Eustachian tube that connects the middle-ear cavity to the back of throat. When the Eustachian tube functions normally, it permits fresh air to enter into the middle-ear cavity and it balances the pressure on either side of the eardrum so that it can work optimally. If the Eustachian tube does not open regularly, air in the middle-ear cavity is resorbed into tissue and a vacuum is formed in the middle ear that causes the eardrum to be retracted. This causes a slight to mild low-frequency conductive hearing loss. If the Eustachian tube ventilates the middle ear, the loss vanishes. If the Eustachian tube does not open, fluid may collect in the middle ear (otitis media with effusion) and cause conductive hearing loss that is painless. If the fluid becomes infected, the retracted eardrum becomes a bulging eardrum that is reddened and painful (acute otitis media). In these cases, immediate medical treatment – usually with antibiotics – is needed to prevent rupture of the eardrum. Successful treatment of the infection eliminates pain but does not necessarily eliminate fluid behind the eardrum or the temporary hearing loss caused by the fluid.

Pressure-equalization tubes (PE tubes) are often inserted surgically in the eardrums to temporarily overcome the problems brought on by poor Eustachian tube function. PE tubes are shaped like sewing bobbins and are a little bigger than the tip of a ball-point pen. They allow air to flow into the middle ear when the Eustachian tube will not open. They typically remain in place from several months to a year before they fall out. Many children with CHARGE require several sets of PE tubes. A marked improvement in hearing is often noted after the insertion of PE tubes.

## **Inner Ear (Cochlea)**

Abnormalities of the inner ear or cochlea are a major cause of permanent hearing loss in CHARGE. The cochlea is named for its shape – snail shell in Latin. It is a normally fluid-filled structure that is smaller than the tip of the little finger, and it is embedded in the hardest bone in the body. It is the sensory end organ for hearing. It changes motion into neural impulses that can be processed as sound by the brain. Damage to the inner ear and the nerves that carry the messages to the brain is called “SENSORINEURAL HEARING LOSS.”

When the shape of the cochlea or the vestibular mechanism (which is joined to the cochlea) is abnormal, it may be detected using radiographic procedures (CT or MRI). However, there may be sensorineural hearing loss even though the shape of the cochlea is normal.

Sensorineural hearing loss creates several problems. Sounds may not be heard unless they are amplified. However, when they are amplified, they may be distorted in a way that they cannot be understood. For example, speech may always sound muffled or garbled even with the best hearing aid. Lastly, there may be “loudness recruitment” in which amplified sounds become intolerably loud. In fitting hearing aids on individuals with sensorineural hearing loss, it is necessary to amplify enough so that speech may be heard but not too much so that speech and other sounds become intolerable. If either goal is not met, the hearing aid may be rejected.

## **Neural Hearing Loss and Central Auditory Processing**

Abnormalities of the nerves leading from the cochlea to the brainstem and from the brainstem to the brain do not cause hearing loss in the conventional sense. If a person had this type of loss, they may have normal hearing thresholds but a poor ability to understand speech under any conditions and especially in background noise. These are problems that receive audiologic and educational treatment rather than medical or surgical treatment.

## **Mixed Hearing Loss**

When both conductive and sensorineural hearing loss is present, the loss is called “MIXED HEARING LOSS.” This is the most common type of hearing loss in CHARGE. It is usually composed of permanent conductive loss due to malformation of the ossicles, fluctuating conductive loss due to the intermittent presence of fluid in the middle ear, and sensorineural hearing loss that usually was present at birth. In general, the conductive losses are greatest for the low frequencies and the sensorineural losses are greatest for the high frequencies. Often there is marked asymmetry in hearing between the two ears.

## **Progressive Hearing Loss**

Progressive hearing loss (conductive and/or sensorineural) is an uncommon finding in CHARGE, but has been reported. However, there is probably insufficient audiological documentation over time on a group of children to reach a good conclusion. In cases where the loss has progressed, it has usually been a small increase superimposed on a large existing loss.

As the child matures and communication improves, behavioral hearing thresholds often improve slightly because of the child's improved ability to concentrate or to perform the listening task, rather than a change in hearing. Thresholds obtained with ABR (BAER) tests should not improve markedly with maturation.

## **HEARING ASSESSMENT**

### **Pure-tone Audiogram**

The most important information to be obtained in an audiologic evaluation is the determination of the thresholds of hearing for specific frequencies for the two ears measured independently. The information is recorded on a form called the "AUDIOGRAM." An example of an audiogram is shown in Figure 1 (next page). The thresholds of hearing can be measured in behavioral tests in which the individual is required to give an active response to sound or using an auditory brainstem response (ABR) test which requires that the person be asleep or sedated. Usually, attempts are made to obtain responses to pure-tone signals over a range of test frequencies that is important for understanding speech (500, 1000, 2000, and sometimes 4000 Hz). This information is very important to the otologic diagnosis and the decisions related to hearing aids.

Other tests may be done, but this is the most important. The audiologic evaluation process should be repeated until there is confidence in the pure-tone hearing thresholds that have been measured.

### **Tympanometry**

This is a test of the mobility of the eardrum and middle ear. It requires only that a child sit still for a short period of time, and it does an excellent job of detecting the presence of Eustachian tube dysfunction and fluid in the middle ear. This test is used regularly in the management of middle-ear disease – however, it is not a test of hearing. "TYMPANOGRAMS" give information about the middle ear and not the overall ability to hear.

## **AURAL HABILITATION**

### **Case Manager for Aural Habilitation**

The aural habilitation program needs to be managed by a professional who is in constant contact with the child with CHARGE. This may be an audiologist associated with the treating otolaryngologist or a professional associated with the educational system (audiologist, speech-language pathologist, special educator, teacher, etc.). If there is a significant hearing loss, the educational progress needs to be monitored closely and the amplification system needs to be checked daily.

## **Amplification**

When the hearing has been measured adequately, hearing aids need to be selected and fitted by the audiologist. It is important to remember that the sole purpose of hearing aids is to improve hearing, and that the aids have little value if they do not achieve that goal. In children with CHARGE, this is very difficult to achieve but it is the goal. The hearing aids must stay on the head, the ear molds must not hurt or irritate the ear canals, there must be adequate amplification but not overpower the child, and the frequencies amplified must improve the ability to understand speech.

In addition to hearing aids, there are amplification devices called assistive listening devices or auditory trainers. These devices have a microphone that is located close to the talker so that the listener has a better chance to hear. Assistive listening devices are valuable when there is background noise – especially in classrooms.

The type(s) of hearing aids selected will depend on a large number of factors. In-the-ear (ITE) aids are usually not appropriate for children. Behind-the-ear (BTE) aids are often chosen because they have power and flexibility. Body-worn aids are also considered if the hearing loss is very large. Cochlear implants are surgically implanted devices that bypass the cochlea and electrically stimulate the nerves directly; they are only considered when the losses are in the severe to profound category. If there is any measurable benefit from hearing aids, it is unlikely that cochlear implant surgery would be performed. There are strict FDA guidelines for who may be considered for implantation. Very few children with CHARGE have received cochlear implants.

## **DEVELOPMENT AND EDUCATION**

For many children with CHARGE, the first years of life are spent treating life-threatening problems. After this period the parents have the chance to focus on development and education. It is important to note that most children with CHARGE pass through critical developmental periods for the development of speech and language in a state of sensory deprivation. Usually there have been significant hearing and vision problems regardless of attempts to overcome them. As a result the child with CHARGE begins the educational process at a disadvantage. In whatever educational setting, a child with CHARGE is placed, there needs to be a coordinated effort of all the professionals who have knowledge of the child's abilities and disabilities to ensure success.

# CHOANAL ATRESIA IN CHARGE SYNDROME: FOR THE PHYSICIAN

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## ANOMALIES SEEN IN CHARGE

A little more than 50% of children with CHARGE have some form of choanal atresia. The range is complete - from bilateral bony choanal atresia to unilateral choanal stenosis. About half of the patients with choanal atresia have bilateral choanal atresia. In the general population, the incidence of choanal atresia is approximately 1 in 5000-7000 live births. A large percentage of these probably represent children with CHARGE.

## DIAGNOSTIC TESTS

### Physical Exam

Once the diagnosis of choanal atresia has been made, it can be confirmed initially on physical exam by failure to pass a # 6 to 8 French plastic catheter through the nares into the pharynx. (a typical solid feeling will be encountered at the level of the posterior choanal approx. 3-3.5 cm from the alar rim). Passage of soft metal probes has also been proposed.

### Imaging Studies

Plain films and tomograms of the skull with radiopaque dye instilled into the nasal cavities can confirm choanal atresia. An axial noncontrast high resolution CT scan with thin sections (2-5mm) has become the single radiographic study of choice. The CT scan has proven invaluable in the accurate assessment of both the normal and abnormal anatomy of the nasal cavity, posterior nasal choanae and nasopharynx. The knowledge obtained from the CT scan is valuable in the preoperative planning of the method and design of the repair.

When planning a CT scan or MRI in a child with suspected CHARGE, consult with otolaryngology: often a few more cuts can yield important information about the inner ear abnormalities seen in CHARGE.

## CONSEQUENCES OF CHOANAL ATRESIA IN CHARGE

### Bilateral choanal atresia

Bilateral choanal atresia causes complete nasal obstruction - immediate respiratory distress and even potential death due to asphyxia (because newborns are obligate nose breathers until approximately 4 to 6 weeks at which time mouth breathing is learned). The respiratory obstruction is cyclic - as the child falls asleep the mouth closes and a progressive obstruction starting with stridor followed by increased respiratory effort and cyanosis. Either the observer opens the child's mouth or the child cries and the obstruction is cleared.

Initial feeding is often the alerting event - as the child starts with inability to eat and breath at the same time, there is a progressive obstruction of the airway and subsequent cyanosis and choking due to aspiration of the milk. This can mimic a tracheoesophageal fistula (TEF). TEF and/or esophageal atresia is common in CHARGE.

### Unilateral choanal atresia

Unilateral choanal atresia rarely causes any acute respiratory distress. The most common finding is a unilateral mucoid discharge. Unilateral choanal atresia does not require any immediate surgical attention, but may eventually require treatment because children with CHARGE have numerous other potential respiratory complications, which can be exacerbated by UL choanal atresia.

## **MEDICAL MANAGEMENT**

An oral airway of some sort must be implemented very early on in the treatment of newborns with choanal atresia. A typical anesthesia oral airway is often sufficient, however if not then orogastric tube may be considered. A large nipple can be modified by having its end cut off and then ties are attached to the nipple and placed around the occiput. This type of airway is called a McGovern nipple and provides an airway through which the baby can breathe. A very small feeding tube can then be passed either through another hole in the nipple or along side the nipple for gavage feeding. This is the preferred method of establishing an oral airway.

### **Tracheotomy and caveats for CHARGE**

This is a controversial issue and many physicians conclude that there is never a need for this drastic a step in the initial management of infants with choanal atresia. Patients with CHARGE have a high propensity of airway instability. These children's early repair of their choanal atresia is rarely successful, primarily due to their abnormal anatomy of their nasopharynx and upper aerodigestive tract. Many children with CHARGE have at least one cardiopulmonary arrest prior to their definitive procedure. Therefore, some investigators have proposed that children who have CHARGE should have early tracheotomy to protect their brain from anoxic injury and delay the repair of their choanal atresia until they are at least 2 years of age. This is still a very controversial subject.

### **Feeding**

Gavage feeding is usually required until the child has learned to mouth breath. Then once the child learns the mouth breath, oral feeding can be attempted. A majority of children with CHARGE have significant feeding problems, possibly related to abnormalities of cranial nerves IX and X. Many of these children require gastrostomy feeding for a number of years. See sections on Swallowing and Growth.

## **REFERENCES**

Asher BF et al. Airway Complications in CHARGE Association. Arch Otolaryngol Head Neck Surg. May 1990;116:594-595.

Blake KD, Davenport SLH, Hall BD, Hefner MA, Pagon RA, Williams MS, Lin AE, Graham JG: CHARGE association: an update and review for the primary pediatrician. Clinical Pediatrics 1998;37:159-174.

Crocket DM et al. Computed Tomography in the Evaluation of Choanal Atresia in Infants and Children. Laryngoscope. 1987; 97:174-183.

Ferguson JL and Neel BH. Choanal Atresia: Treatment trends in 47 patients over 33 years. Ann of Otol Rhinol. Laryngol. February 1989; 98:110-112.

Kaplan LC. The CHARGE Association: Choanal Atresia and Multiple Congenital Anomalies. Otolaryngologic Clinics of North America. June 1989; 22:661-672.

## CHOANAL ATRESIA: PARENT INFORMATION

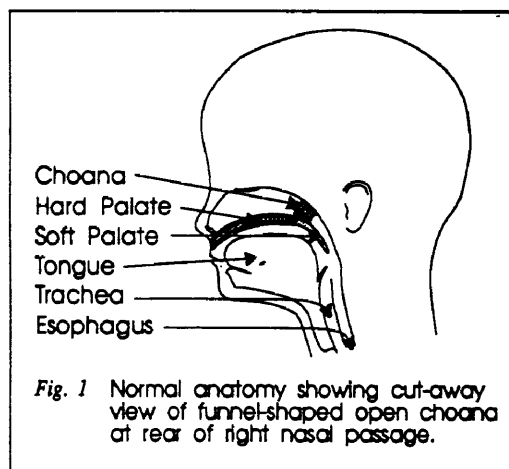
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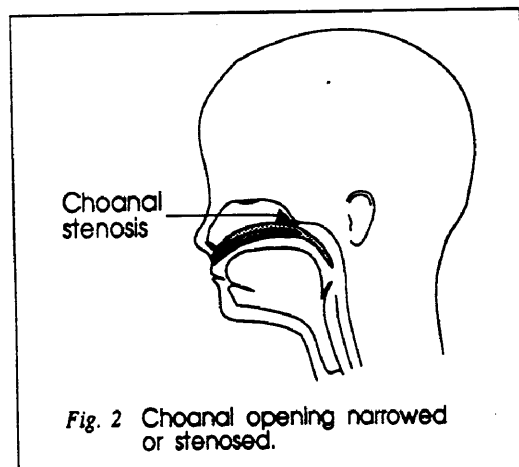
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### NORMAL STRUCTURE AND FUNCTION

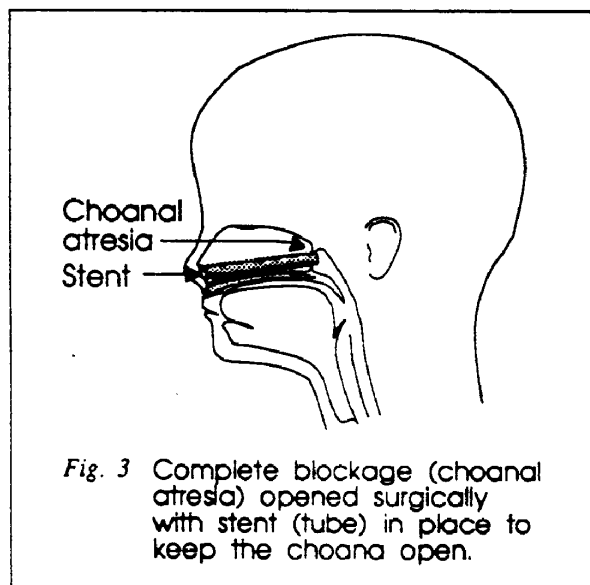


The choanae are funnel-shaped openings at the back of the nasal passages which connect the nose with the throat. They are located just above the roof of the mouth, where the hard and soft palate meet (figure 1).



In normal fetal development, the choanae open when a membrane breaks down at about the seventh week of pregnancy. If this membrane does not break down, choanal atresia results. If the blockage is only partial, the term choanal stenosis (meaning tight or narrow) is used (figure 2). Choanal atresia is total blockage between the nose and the throat (figure 3).

The choanae are critical in the newborn because newborn babies are obligate nose-breathers. If both nasal passages are totally closed off (bilateral choanal atresia), the newborn child almost always has trouble breathing. Babies cannot instinctively switch from nose breathing to mouth breathing. This can lead to lack of air (asphyxia) with lack of oxygen (anoxia), which can result in respiratory distress, brain damage, or death if not recognized and treated quickly.



**Fig. 3**  
Complete blockage (choanal atresia) opened surgically with stent (tube) in place to keep the choanal open.

### **FORMS OF CHOANAL ATRESIA IN CHARGE**

About half of all children with CHARGE have some form of choanal atresia. It is very rare in other conditions, and therefore very helpful in making a diagnosis of CHARGE. Children with CHARGE can have choanal atresia (blockage) or choanal stenosis (narrowing). It can affect both sides (bilateral, BL) or only one side (unilateral, UL). Choanal atresia can be bony (bone or cartilage blocking the passageway) or membranous (soft tissue membrane blocking the passageway).

Choanal atresia can be present even if there is a cleft palate. It may be more difficult to diagnose, as air can move through the cleft. Unilateral choanal atresia and choanal stenosis can be very difficult to recognize.

### **DIAGNOSIS OF CHOANAL ATRESIA**

Polyhydramnios (excess amniotic fluid) during pregnancy is often the first sign of choanal atresia. Normally, the fetus breathes amniotic fluid during pregnancy. If the nose is blocked, and the fetus can't "nose breathe," the amniotic fluid is not "recycled" by fetal breathing motions and excess fluid may build up in the uterus.

A tube should be passed through each nostril and nasal passage of any infant who has unexplained breathing difficulties in the first month of life. This will establish if the nasal passages are open (patent). X-rays can be performed using dye in the nasal passages to verify they are not open. Sometimes other imaging (MRI, CT) is used to confirm the diagnosis and determine the extent of the problem.



## **TREATMENT**

### Bilateral choanal atresia

Bilateral choanal atresia is life threatening in the newborn period, and treatment is urgent. Emergency treatment usually consists of placing a plastic airway (tube) into the mouth to keep the mouth open. This allows the baby to mouth-breathe. Other newborns may require intubation: passing a breathing tube through the mouth and down into the windpipe (trachea) so oxygen goes directly into the lungs. Occasionally, it is necessary to do a tracheotomy: surgically putting a breathing tube directly into the trachea through a hole in the lower front of the neck.

The above procedures are all temporary. Soon after, surgery will be performed to open the bone or membrane covering the nasal passage. A stent (plastic tube) is placed in each nasal passage to keep it open and guarantee adequate air entry (figure 3). The stents may have to stay in for a few weeks or even months. Children with CHARGE are more likely to have complications following choanal atresia repair than other children with choanal atresia. A few require repeat surgery later because the choanae close up again when the stents are removed.

### Unilateral choanal atresia and/or choanal stenosis

Lesser problems usually occur when one nasal passage is totally obstructed (UL atresia) or when one or both nasal passages are narrowed (stenosis). Air can pass in varying amounts through the nose into the lungs. The child with UL atresia or stenosis often does not have obvious respiratory symptoms, but he or she may have a constant runny nose on the side that is blocked or narrow. Recognizing choanal stenosis or unilateral atresia can be important in the treatment of these respiratory complications.

## **OUTCOME**

A major long-term complication of choanal atresia or stenosis is recurrent ear infections, which can lead to a conductive hearing loss. Because children with CHARGE also have a high risk for sensorineural hearing loss, these ear infections should be treated aggressively.

Children with CHARGE may require more than one surgery before the choanal atresia is permanently repaired. Often, difficult decisions must be made about surgery - should the heart be fixed first or the choanal atresia? Is the baby strong enough for surgery, given the heart problems, etc? Any newborn with CHARGE should have the choanae evaluated before any surgery is done because of possible complications. Choanal atresia and the resultant lack of oxygen increases the risk for mental retardation due to brain damage.



## OROFACIAL CLEFTING IN CHARGE SYNDROME: FOR THE PHYSICIAN

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### CRANIOFACIAL ANOMALIES SEEN IN CHARGE

Orofacial clefting occurs in about 20% of children with CHARGE syndrome. These children may have cleft lip with or without cleft palate or isolated cleft palate, especially sub-mucous cleft palate.

### DIAGNOSTIC CONSIDERATIONS:

In general, patients with CHARGE are more likely to have cleft lip, while those with velo-cardio-facial syndrome (VCFS) are more likely to have a cleft palate. When orofacial clefting is present in patients with CHARGE, the choanae are usually patent, so this finding (clefting) may substitute for choanal atresia in the diagnostic criteria, particularly if the remaining findings are otherwise characteristic of this condition.

Other anomalies common in CHARGE which may affect treatment and/or management of orofacial clefts:

- TE fistula or esophageal atresia
- Cranial nerve IX/X palsies
  - Laryngotracheomalacia
  - Velopharyngeal incompetency
  - Reflux
- Facial palsy, unilateral or bilateral
- Heart defects
- Ossicular malformations with or without Mondini anomaly
- Choanal atresia or stenosis

### DIAGNOSTIC TESTS:

While cleft lip is obvious to the casual observer, the diagnosis of submucous cleft palate in CHARGE can sometimes be delayed. Many cases have been confirmed only when tonsils or adenoids are removed. **An evaluation looking for cranial nerve IX/X involvement is crucial prior to beginning a feeding program in order to avoid multiple aspiration pneumonias.**

## **CONSEQUENCES OF OROFACIAL CLEFTING IN CHARGE:**

### **FEEDING:**

Orofacial clefting can interfere with feeding in any individual. Children with CHARGE often have additional feeding problems due to neurologically-based velopharyngeal incompetency and/or reflux. Cleft lip or cleft palate can exacerbate this situation. Children with CHARGE (with or without orofacial clefting) often require g-tube feeding for significantly longer than children with other clefting syndromes.

### **EAR INFECTIONS/HEARING LOSS:**

Children with facial clefts are prone to ear infections and possible hearing loss. Children with CHARGE typically have more ear infections and require PE tubes more often and for a longer period of time than other children with clefts. Children with CHARGE often have sensorineural hearing loss and/or conductive hearing loss due to malformed ossicles. Close follow-up by ENT and audiology is critical to maximizing the hearing in these children.

### **SPEECH:**

An undiagnosed submucous cleft palate can interfere with speech development, already complicated in these children due to hearing loss and facial palsy.

## **MEDICAL MANAGEMENT AND CAVEATS**

Team management approach: It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. The cleft team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. When following a child with CHARGE, it is important to communicate with other specialists who are following the child. The feeding team members should understand the neurologic basis for incoordinated swallow and reflux. The members of the team and the approach to the child will vary from one institution to another. The specific management plan for orofacial clefting in a child with CHARGE will vary depending on the specific problems of that child as well as the protocols for a particular institution.

Early simple surgical repair maximizes optimal speech, aids in feeding, assists in reducing middle ear infections, and re-establishes normal separation of the oral and nasal cavity while minimizing growth disturbances of the upper jaw.

Post-surgical complications such as infection, dehiscence, oro-nasal fistula, and malocclusion may be more frequent in children with CHARGE. Therefore, a 23-hour stay planned for children with isolated clefts may need to be extended to an inpatient stay for children with CHARGE.

**Consider insertion of PE tubes and the taking of dental impressions at the time of surgery if indicated.**

Bilateral cleft lip and palate is rare in CHARGE syndrome. Although late treatable complications such as dental malocclusion, velopharyngeal incompetence, oro-nasal fistulae, and distortion of normal anatomy can often occur, initial planning during the first twelve months of life mirrors that of unilateral cleft lip and palate.

## **SPECIAL CONSIDERATIONS IN TREATMENT OF FACIAL CLEFTS IN CHARGE:**

- 1) Heart defects (present in 2/3 of children with CHARGE) may be complex. These may affect timing of surgery and overall stability of the patient.
- 2) Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.
- 3) Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications.
- 4) Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years, with a few children having permanent problems. In addition, facial palsy is usually unilateral and may affect lip closure and may compromise ultimate cosmetic outcome of cleft lip.

## **NON-MEDICAL MANAGEMENT**

If a patient has CHARGE, orofacial clefting is likely to be only one of a number of serious medical problems for that child. The cleft may or may not be a high-level concern to the parents. It is critical to talk to the parents about the whole child, and not just about the cleft. Parents are often especially concerned with how the cleft correction fits into other surgical schedules.

The orofacial team, which includes ENT, audiology, and feeding specialists, can be a great help to parents (even if the child does not have a cleft) because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of appointments is appreciated by the parents.

## **REFERENCES:**

Blake K., Davenport S.H., Hall B.D., Hefner M.A., Pagon R., Williams M.S., Lin A.E., Graham J.M. Jr.: CHARGE Association - An update and review for the primary pediatrician. *Clinical Pediatrics*, 37:159-174, 1998.

Blake KD, Russell-Eggitt IM, Morgan DW et.al. Who's in CHARGE? Multi-disciplinary management of patients with CHARGE association. *Arch. Dis. Child.* 1990;65:217-223.

Tellier, AL, Cormier-Daire v, Abadie V, et al. CHARGE syndrome: report of 47 cases and review. *AmJ Med Genet* 76:402-409 (1998)



## CLEFT LIP AND PALATE (CL/P) IN CHARGE: PARENT INFORMATION

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### NORMAL STRUCTURE AND FUNCTION:

You know what the lips are! The **palate** is the roof of the mouth: the bony and muscular structure between the oral (mouth) and nasal (nose) cavities.

### EMBRYOLOGY:

The lips and palate develop between 5 and 7 weeks of gestation (pregnancy). They result from the growth, merging, and fusion of five "processes:" two maxillary (upper jaw) processes, two mandibular (lower jaw) processes, and one frontonasal (nose) process (see diagram). The lower lip is formed when the mandibular processes unite. The upper lip is formed from the two maxillary processes and the frontonasal process. They come together (two from the side, and one from above) to form the upper lip. The "cupids bow" in the center of the upper lip shows the borders where the processes meet.

The primary (hard) palate, the nasal cavity and the choanae (passages from the back of the nose to the throat) result from merging of the medial nasal processes at 5 weeks. At 6 weeks, the secondary (soft) palate processes develop and fuse. This creates the soft palate, the nasal cavity and the choanae. The lips, palate and nasal cavity should be complete by about 7 weeks gestation. If any of these processes fail to fuse or merge, a gap, or cleft, results.

### FREQUENCY OF OROFACIAL CLEFTING IN CHARGE:

Orofacial clefting (cleft lip or cleft palate) occurs in about 20% of children with CHARGE syndrome. Submucous cleft palate is often not diagnosed in the newborn period, so the frequency of clefting may be even higher. It is possible to have both choanal atresia and cleft palate.

### Diagnosis of Clefts in CHARGE

Cleft lip is obvious at birth. The newborn exam performed in the delivery room usually includes an evaluation of the palate. A cleft of the hard palate should be apparent as part of this exam. A submucous cleft is more difficult to diagnose and is sometimes not recognized until much later.

### CLEFT LIP (with or without cleft palate, CL/P):

Cleft lip can be **unilateral** (UL, one-sided) or **bilateral** (BL, two-sided). Cleft lip typically occurs between the side and center (cupids bow) portions of the upper lip. It may involve only the lip, or extend into the gum and even into the primary (hard) palate.

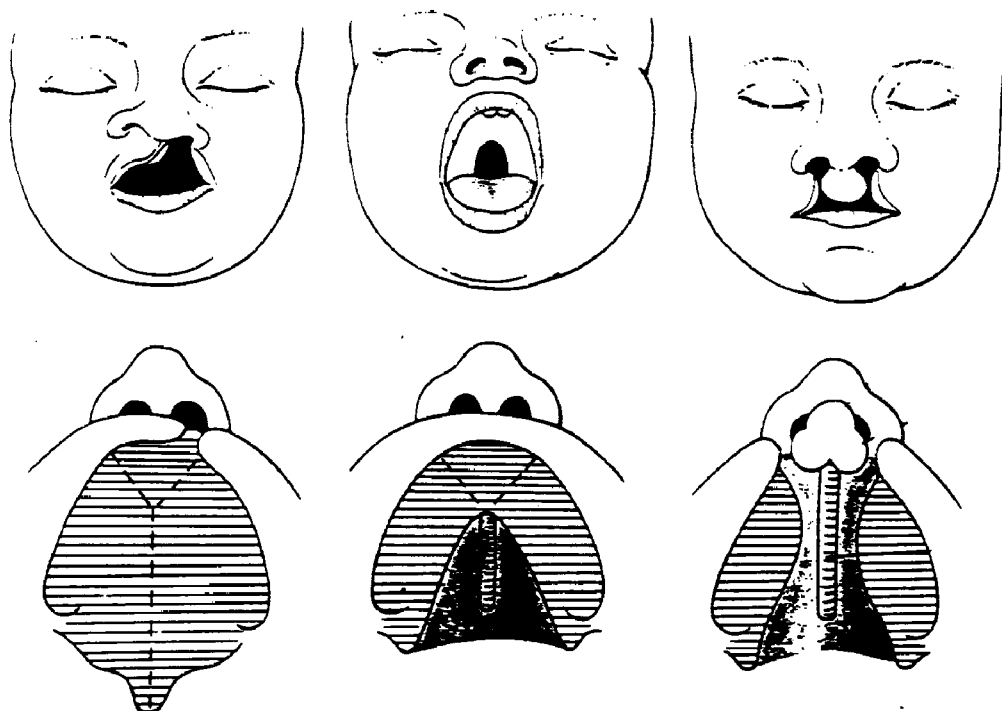
### CLEFT PALATE (CP):

Cleft palate results from a failure of fusion of the palatal shelves. Isolated cleft palate (without cleft lip) is in the back of the palate. It can involve the hard and soft palate or just the soft palate. Children with cleft palate typically have underdevelopment of the mid-face (flat midface) and often a small chin.

**Submucous cleft palate:** Submucous cleft palate involves the muscles (but not the bones) of the soft palate. Swallowing and speech are often affected.

Bifid uvula:

Bifid (split or double) uvula (little thingee that hangs down in the back of the throat) is considered a mild form of cleft palate. This alone does not usually create problems. It may be a sign to look more closely for a possible submucous cleft palate.



#### **EFFECT OF CLEFTING IN THE CHILD:**

Although cleft lip is a cosmetic problem, the more immediate concern is the effect on feeding. Special nipples may help the child suck from a bottle. Nursing is sometimes difficult or even impossible. Cleft palate makes feeding even more difficult, as the milk can pass through the palate into the nose. Children with CHARGE often have additional feeding problems due to facial palsy, reflux, and/or swallowing problems (see FEEDING section). **Nurses and other specialists in a cleft palate clinic often have extensive experience with feeding difficulties and can be a big help to parents.**

Swallowing and speech are affected by cleft palate, sometimes even after surgical correction. Children with a cleft palate are more prone to ear infections and aspiration. Children with CHARGE (with or without a cleft) typically have multiple ear infections and require multiple PE tubes (see EARS section). Children with cleft palate are prone to conductive hearing loss (see HEARING section).

Most children with CHARGE and a cleft (and many without clefts) are unable to take food by mouth for some time and require a gastrostomy tube (g-tube) for some time (see FEEDING section).



Children with cleft lip/palate often have orthodontic problems which will need to be addressed. Children with CHARGE may have particular dental problems as well, but these have not yet been well-described.

## **MEDICAL MANAGEMENT**

### **TEAM APPROACH:**

It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. Members of the Craniofacial team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. Every institution will have a slightly different team and perhaps a different approach to facial clefts. Make use of these team members! Many of them will have had experience with feeding problems in other children.

### **TREATMENT/SURGERY:**

The treatment for facial clefting involves a series of surgical procedures which may take place over many years, even through the patient's young adult life. The actual timing and type of treatment takes into consideration the child's growth, development, and other medical problems. Prior to surgery, it is often necessary to take dental impressions. Some children will need a pre-surgical orthodontic appliance and home therapy to prepare for surgery.

Surgery is not usually scheduled immediately for cleft lip and palate. Children with CHARGE often have more immediate surgical needs, such as choanal atresia or a heart defect. Many palatal clefts will be repaired between eight and twelve months of age in a single stage, although surgery on very wide clefts may be delayed until up to eighteen months of age.

### **OTHER TESTS/PROCEDURES:**

Audiological (hearing) testing should be done early. Children with CHARGE have an even higher risk of hearing loss than do other children with clefts. Insertion of PE tubes and the taking of dental impressions may be done during the same anesthesia as cleft surgery.

### **Non-medical management**

Most of the non-medical concerns related to facial clefts are feeding concerns. See the FEEDING section for more information.



## **CHARGE SYNDROME: ESOPHAGEAL ATRESIA AND TRACHEOESOPHAGEAL FISTULA: FOR THE PHYSICIAN**

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### **TYPES AND FREQUENCY OF TRACHEO-ESOPHAGEAL ANOMALIES IN CHARGE:**

Tracheo-esophageal fistula	20%* (or higher, often H-TEF is diagnosed late)
Esophageal atresia	15%

Related findings frequent in CHARGE which may affect treatment and/or management of tracheo-esophageal anomalies:

Polyhydramnios (prenatal)	frequent
Tracheomalacia	frequent
Laryngomalacia	30%
Gastro-esophageal reflux	50%
Esophageal dyskinesia	75%
Pharyngo-esophageal dysmotility	80%
Facial palsy	50%
Cleft palate	20%
Choanal atresia	30%
Cranial nerve IX/X anomalies	frequent
Abnormal Ba swallow	very frequent
Gastrostomy in patient w/o EA	36%
Fundoplication	frequent

### **DIAGNOSTIC TESTS:**

NG tube  
X-ray  
Barium swallow

The newborn with CHARGE who has TEF/EA will have copious oral secretions and a nasogastric tube will not pass into the stomach.

### **MEDICAL CONSEQUENCES OF FEATURES**

Children with CHARGE and TEF/EA have higher mortality than other children with CHARGE. This is especially true when combined with choanal atresia and/or a heart defect.

Many individuals with CHARGE have abnormal esophagus motility and/or gastro-esophageal reflux. A significant percentage also has difficulties with suck and swallow resulting from cranial nerve anomalies. These problems further complicate feeding problems in these children. Many children need gastrostomy feeds for years.

## MEDICAL MANAGEMENT AND CAVEATS

Treatment of EA/TEF in CHARGE may be similar to treatment of isolated EA/TEF. However, there are a number of special considerations in these cases:

1. Heart defects (present in 2/3 of children with CHARGE) may be complex. These may affect timing of surgery and overall stability of the patient.
2. Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.
3. Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications. The mortality rate in children with this combination of features is high.
4. Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years. A small number of children have permanent problems. Facial palsies are usually unilateral and may affect lip closure.

## NON-MEDICAL MANAGEMENT

A feeding specialist is a must for children with CHARGE with TEF/EA. All of them will have feeding difficulties, which are likely to last for years. Transition from gastrostomy feed to oral feeding can take years, as can transition from pureed to solid and liquid foods. Growth may be impaired by poor caloric intake.

If a patient has CHARGE, TEF/EA is likely to be only one of a number of serious medical problems for that child. It is critical to talk to the parents about the whole child. An orofacial team which includes ENT, audiology, and feeding specialists can be a great help to parents because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of medical appointments is appreciated by parents.

## REFERENCES

Blake KD, Davenport SLH, Hall BD, et al. CHARGE Association: An update and review for the primary pediatrician. Clin Pediatr 1998; 37:159-174.

Tellier AL, Cormier-Daire V, Abadie V, et. al. CHARGE syndrome: Report of 47 cases and review. Am J Med Genet 76:402-409, 1998.

Markert ML, Majure M, Harville TO, et al. Severe laryngomalacia and bronchomalacia in DiGeorge syndrome and CHARGE association. Pediatr Pulm 24:364-369 (1997).f

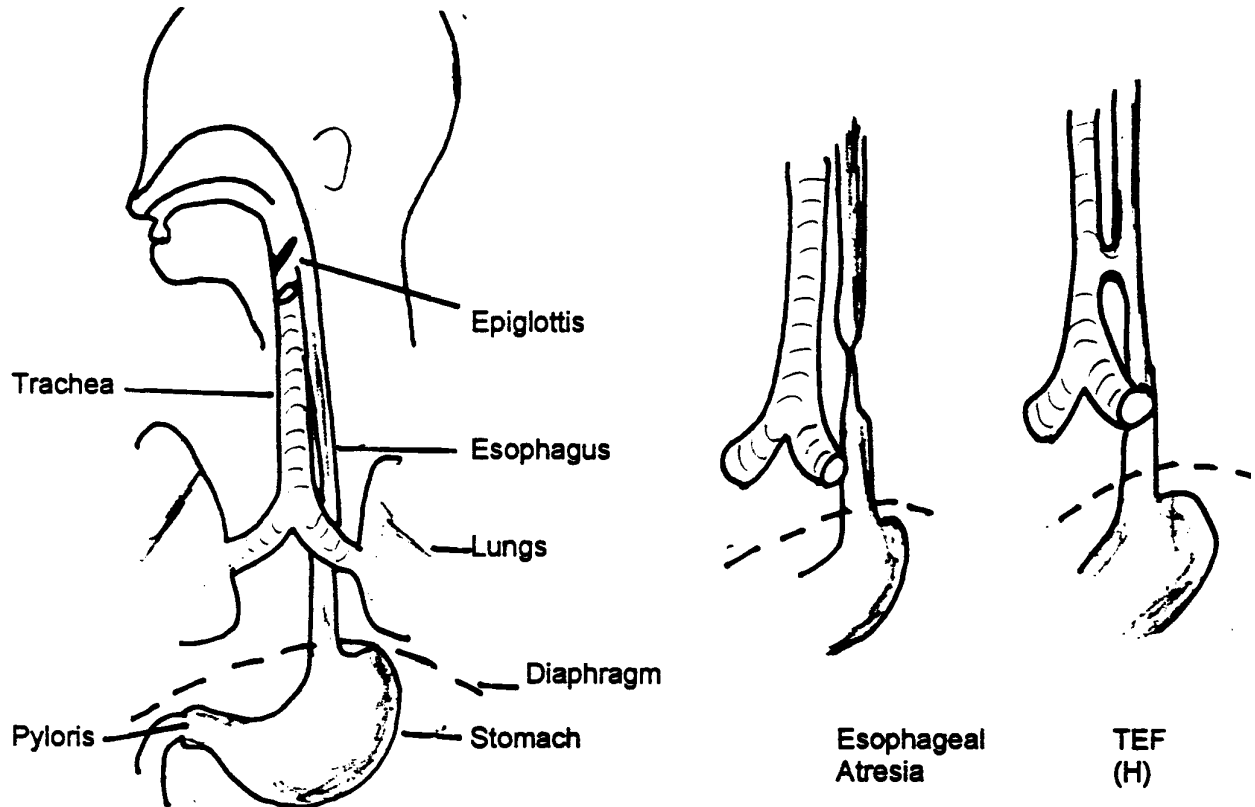
Fraser C, Baird PA, Sadovnick AD. A comparison of the incidence trends for esophageal atresia and tracheoesophageal fistula, and infectious disease. Teratology 36:363-369, 1987.

Kutianawala M, Wyse RKH, Brereton RJ, et. al. CHARGE and esophageal atresia. J Pediatr Surg 27:558-560, 1992.

## CHARGE SYNDROME: ESOPHAGEAL ATRESIA (EA) AND TRACHEOESOPHAGEAL FISTULA (TEF): PARENT INFORMATION

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### Structure and function



The **epiglottis** is a small flap in the back of the throat. The purpose of the epiglottis is to ensure that food goes down the esophagus and air goes down the trachea.

The **trachea** (windpipe) is the tube leading from the back of the throat to the lungs for breathing. The trachea runs along side the esophagus and then splits in two along either side of the esophagus, leading to the two lungs. The trachea is surrounded by rings of cartilage and muscle.

The **esophagus** is the tube leading from the back of the throat to the stomach. It is surrounded by a coat of muscle, which help the food move down into the stomach. Food normally moves down the esophagus into the stomach. Normally, when food or saliva is swallowed, waves of movement in the esophagus (peristalsis) propel the food into the stomach. The opening from the stomach to the intestines is called the **pyloris**. Swallowing normally begins in the first three months of prenatal development.

## Problems associated with CHARGE

**Esophageal atresia (EA)** refers to an esophagus which ends blindly before reaching the stomach. It is often found along with tracheo-esophageal fistula. Often, polyhydramnios (excessive amniotic fluid) was noted during pregnancy. Prenatally, the fetus normally swallows and recycles amniotic fluid. If the fetus cannot swallow due to EA, polyhydramnios can result. Also, the stomach may not be visible by ultrasound exam because it is not filled with amniotic fluid.

Once the baby is born, he or she will often have copious frothy secretions pouring out of their mouths which require constant suctioning until the EA is treated. A baby with EA cannot eat by mouth (food can't get to the stomach) until the atresia has been surgically corrected.

### Tracheo-esophageal fistula (TEF)

A fistula is a connection. TEF is a connection between the esophagus and the trachea. This connection allows food (from the esophagus) to enter the lungs (aspiration). Food in the lungs can cause pneumonia (referred to as aspiration pneumonia), which can be very serious. If there is a TEF with EA, all the food taken in will end up in the lungs, as the esophagus does not connect to the stomach, but ends blindly.

The hardest type of TEF to diagnose is **H-type TEF**, where the esophagus does not end blindly, but there is a fistula or connection between the trachea and the esophagus. In H-type TEF, air can enter the stomach (from the trachea) and food can enter the lungs. Air in the stomach can cause bloating of the baby's abdomen and can make the baby very uncomfortable.

### Symptoms of TEF/EA

If you had polyhydramnios or the fetal stomach could not be seen on ultrasound, TEF/EA may have been suspected during your pregnancy. After a baby is born, esophageal atresia may be suspected in the first few days if the baby doesn't tolerate feeding (chokes and spits), doesn't seem to swallow saliva (froths at the mouth), seems to aspirate the milk into his or her lungs, or has abdominal distention (bloated tummy).

H-type TEF may not be recognized until much later. The typical symptoms are choking with feeding, excessive gas in the stomach or intestines, and **frequent aspiration pneumonias**.

By now, you probably already know enough about CHARGE to recognize that many of these symptoms can also be caused by other problems associated with CHARGE. Choanal atresia, cleft palate, reflux, and cranial nerve abnormalities can all interfere with breathing and eating. Infants with CHARGE may not be fed right away due to other problems. And so on. This means that TEF and associated problems may not be diagnosed as quickly or as thoroughly in infants with CHARGE.

### Diagnosis of TEF/EA

Esophageal atresia is usually diagnosed by placing a tube either down the nose or through the mouth, down the throat and into the stomach. An X-ray is taken to document that the tube is in the esophagus (not the trachea) and that it has reached the stomach. If the baby has EA, instead of ending up in the stomach, the tube will curl up in the blind-ending pouch. If there is a tracheo-esophageal fistula (which is very common with EA), the tube may end up in the lungs.

Other tests, which might be done to diagnose or confirm the diagnosis, are bronchoscopy or esophagoscopy. Barium swallow may be needed to diagnose an H-type TEF, where the esophagus is attached to the stomach, but there is a connection to the trachea.

### **Treatment**

EA and TEF can be surgically repaired. Before the repair and while it is healing, the baby will need to be fed by a gastrostomy tube or button. This is a tube or opening which goes directly into the baby's stomach, bypassing the esophagus. How long the baby will require the G-tube is variable. Many children with CHARGE are fed by g-tube for years (see below and Feeding section)

Occasionally, EA will be repaired and yet a small H-type TEF is not recognized. The remaining TEF can still cause multiple aspiration pneumonias. A barium swallow test might be helpful in diagnosing the TEF.

### **Outcome and complications:**

Even after surgical correction, there can be some leakage at the site of the reattachment. The esophagus can tighten up. If this happens, it can be treated by dilating the esophagus. Many children with CHARGE have swallowing difficulties, gastroesophageal reflux, recurrent pneumonias, and poor growth. Some of this may be due to TEF/EA, but much of it may be due to other complications of CHARGE (see FEEDING section). Once the baby has recovered from surgery, oral feeding might be tried. However, individuals with CHARGE typically have other features which complicate oral feeding. Be careful not to push it!





## THE AIRWAY IN CHARGE: FOR THE PHYSICIAN

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Airway management is often one of the major problems in CHARGE. Many of the anomalies are well-known entities with well-established management protocols. However, CHARGE has several fairly unique features which make decision-making difficult, particularly whether or not to do a tracheotomy.<sup>1,2</sup>

### ANOMALIES

Choanal atresia – surgical management is not different from other conditions.<sup>3</sup>

Laryngo-tracheo-bronchomalacia – can be severe<sup>4</sup> leading to collapse of bronchi and decreased air movement.

Cleft lip and palate – surgical management is standard but feeding management is not. See gastroesophageal reflux below.

Gastroesophageal reflux related to neonatal brain stem dysfunction, which is the major cause of feeding problems in CHARGE. NBSC, previously reported in children with Pierre Robin sequence is a group of four types of symptoms involving the supranuclear region of the IXth, Xth and XIIth cranial nerves.<sup>5,6,7</sup> These symptoms, important in the first two years of life, seem to be related to a developmental defect of the suck-swallow central pattern generator in the solitary tract.<sup>8</sup> Recurrent aspiration pneumonia due to this problem is a major cause of morbidity and mortality.<sup>9</sup>

Heart disease – anomalous pulmonary venous return and vascular rings have been reported.<sup>10</sup>

Tracheoesophageal fistula – surgical management is the same but, again, feeding is not.

Other more minor airway anomalies and those above which are not recognized prior to anesthesia can lead to difficulty with intubation.<sup>11</sup>

Recent anecdotal report of asymmetry of diaphragm movement was noted on fluoroscopy of a one-year old.

### MANAGEMENT DECISIONS

Tracheotomy: While most physicians try to avoid tracheotomy in these patients, the swallowing problem can be so severe that the child does not swallow its own secretions. The secretions, then, become copious and are a management problem in themselves. They obstruct the upper airway and may be aspirated along with any feedings. After tracheotomy

is performed, secretions and any formula or food is usually seen coming out the trach with suctioning. Tracheotomy may be performed in a child who has NBSC until such time as it resolves spontaneously usually over several years. Rarely is there a need for permanent tracheotomy, but several cases are known.

Tracheal diversion: This procedure has been performed in older children whose NBSC has not resolved spontaneously, are unlikely to develop speech, and who show a strong desire to eat.

## REFERENCES:

- <sup>1</sup> Roger G, et al. The CHARGE association: The role of tracheotomy. Arch Otolaryngol Head Neck Surg. 1999;125:33-38.
- <sup>2</sup> Asher BF, McGill TJI, Kaplan L et al. Airway complications in CHARGE association. Arch. Otolaryngol. Head Neck Surg. 1990;116:594-595.
- <sup>3</sup> Samadi DS, Shah UK, Handler SD. Choanal atresia management: a twenty year experience. (submitted).
- <sup>4</sup> Markert ML et al. Severe laryngomalacia and bronchomalacia in DiGeorge syndrome and CHARGE association. Pediatr Pulmonol 1997; 24:364-9.
- <sup>5</sup> Abadie V, Chéron G, Couly G. Le syndrome néonatal de dysfonctionnement du tronc cérébral. Arch Fr Pediatr 1993;50:347-52.
- <sup>6</sup> Abadie V, Chéron G, Lyonnet S et al. Le dysfonctionnement néonatal isolé du tronc cérébral. Arch Pediatr 1996;3:130-6.
- <sup>7</sup> Abadie V, André A, Zaouche A et al. The oro-oesophageal dyskinesia : a cause of infant feeding disorders. J Ped Gastroenterol Nutr (submitted)
- <sup>8</sup> Jean A. Brainstem. Control of swallowing : localization and organization of the central pattern generator for swallowing. Neurophysiology of the jaws and teeth. Ed A. Taylor Mc Millan Press 1990 pp 294-321.
- <sup>9</sup> Sporik R, et al. Lung involvement in the multisystem syndrome CHARGE association. Eur Respir J 1997;10:1354-1355.
- <sup>10</sup> Lin AE, Chin AJ, Devine W et al. The pattern of cardiovascular malformations in the CHARGE association. Am J Dis Child 1987;141:1010-1013.
- <sup>11</sup> Stack CG, Wyse RK. Incidence and management of airway problems in the CHARGE Association. Anaesthesia 1991; 46:582-7.

# SWALLOWING AND FEEDING IN CHARGE SYNDROME: FOR THE PHYSICIAN

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## ANOMALIES SPECIFIC TO CHARGE

Swallowing difficulties, one of the major features of CHARGE syndrome, particularly in the first years of life, occurred in 96% of our children with CHARGE<sup>1</sup>. Other authors report between 31 and 88%.<sup>2,3</sup> The main causes are as follows:

### **Poor suck-swallow coordination. Neonatal Brain Stem Dysfunction (NBSD):**

Neonatal brain stem dysfunction is the major cause of feeding problems in CHARGE.

NBSD, previously reported in children with Pierre Robin sequence (4-6), is a group of four types of symptoms involving the supranuclear region of the IXth, Xth and XIIth cranial nerves. These symptoms, important in the first two years of life, seem to be related to a developmental defect of the suck-swallow central pattern generator in the solitary tract.<sup>7</sup>

- (i)      *sucking and swallowing skills disorder,*
- (ii)     *esophageal dyskinesia,*

Clinically, both of these symptoms result in poor suck, prolonged feeding time, milk aspiration, unexplained cries during bottle feeding, nasal reflux, regurgitation and ALTE (Apparent Life Threatening Event) during feeding. They may lead to pharyngeal congestion, aspiration pneumonia and failure to thrive. The esophageal dyskinesia is responsible for gastroesophageal reflux (GER) that is poorly managed by classical medical treatments.

- (iii) *glosso-pharyngeo-laryngomalacia*
- (iv) *sympathetic-parasympathetic cardiac rhythm dysregulation.*

The glosso-pharyngeo-laryngomalacia is responsible for obstructive apneas or hypopneas and subsequent hematemesis while the vagal dysregulation may result in ALTE, induced by all previously noted symptoms. Sucking and swallowing incoordination of NBSD resolves when corticalization of feeding occurs, after 6 mo. of life.

### **Facial palsy**

The facial nerve plays a role in face and lip movements which increase sucking problems. Facial palsy in CHARGE is almost always unilateral and peripheral due to dysgenesis of the VIIth nerve extra bulbar pathway. Determining the origin of facial palsy is important for prognosis and electromyogram may help. Facial palsy may show little or no improvement with age and may be responsible for further aesthetic problems.

### **Malformations of esophagus, larynx or choanae ; cleft lip and palate.**

Anatomical malformations of the organs involved in sucking, swallowing and breathing are also responsible for swallowing and are discussed in other sections of this manual.

### **Hyposmia**

Abnormal olfactory lobes have been noted in CNS autopsy of CHARGE patients<sup>2</sup> and recently, the common embryological origin of olfactory tracts and hypothalamus involved in hormonal defects (LHRH) led to a better MRI analysis of that brain region, showing frequent anomalies.<sup>8, personal data</sup>

Even at birth smell seems to be involved in feeding behavior and increases appetite. This is more evident after corticalization of feeding, i.e. at the end of the first year. The clinical consequences of hyposmia need to be further defined, even in young infants, by perfecting olfactometric scales for children before language acquisition. Olfactory competence in CHARGE has been poorly investigated for both technical and medical reasons: i) olfactometric scales requiring good language cannot be performed by a normal child before 10 years of age and ii) hyposmia is a minor problem that escapes notice.

### **Exogenous factors**

Finally, feeding difficulties may also be secondary to exogenous factors such as cardiac or pulmonary dyspnea, effects of initial nasogastric tube feeding whatever the reasons of the enteral nutrition and deleterious effects of the long initial hospitalization both on the child and on altering the precocious mother-child emotional bonds.

## **DIAGNOSTIC TESTS**

Sucking and swallowing disorders are mainly evaluated by clinical means. A guided anamnesis, a good history and observation of the child during feeding are the best tests. Cineradiography may be dangerous and is not sensitive enough in mild cases. It may help determine when to restart an oral feeding program when aspiration risks diminish. Succimetry may be interesting but, in our hands, remains a research procedure.

Neonatal brain stem dysfunction can be investigated by its peripheral effects.

- i) Esophageal dyskinesia may be investigated by an esophageal manometry, which provides specific information on abnormal esophageal motor control: hypertonia or achalasia of the lower esophagus sphincter, abnormal coordination of pharyngeal contraction and the upper esophageal sphincter. These features are not constant (95% of investigated children in our series had at least one abnormal manometric criterion, 60% a specific anomaly and the rest a less specific abnormal motility of the esophagus). Investigations of the neurologically-based GER are only necessary when surgical treatment is considered. X-ray barium transit is useful but pH meter readings are less so since results may be normal even if vomiting is evident.
- ii) Laryngeal endoscopy may show specific aspects of hypotonia of the tongue base, pharynx walls and larynx. It may also show salivary stasis and peptic inflammation of the larynx and trachea secondary to gastric acid reflux and feeding aspirations.
- iii) A 24-hour electrocardiogram recording (Holter) with ocular compression test may help to evaluate a vagal hyperactivity.
- iv) Evoked potentials of the brain stem may show abnormal delay in tracing, particularly during the first steps of the auditory evoked potentials (AEP) used in

assessing hearing loss. Swallowing disorders by themselves do not require this investigation in practical terms.

- v) Facial and endobuccal electromyography (EMG) as well as dynamic EMG (when it is not dangerous) during bottle feeding may be important to determine the origin of cranial nerves defects. Most often, recording of the 7th cranial nerve shows peripheral palsy. Recordings of 9th 10th and 12th nerves separately are normal but dynamic EMG during bottle feeding shows poor coordination between muscles innervated by the 9th and 10th nerves respectively.<sup>9</sup> This test mainly has theoretical interest but it is quite aggressive and needs a specialized operator.
- vi) Finally, investigations of olfactory abilities may be performed with adapted scales (personal data) or by MRI of the olfactory lobes. Again, these investigations are not required for proper treatment of swallowing difficulties.

## MEDICAL CONSEQUENCES

Medical consequences of swallowing disorders are numerous. They alter pulmonary status, induce chronic bronchitis with a risk of hypoxemia and hypercapnia and adversely affect cardio-pulmonary vascularization, quality of sleeping and psychomotor development. Feeding aspirations and stasis in the pharynx worsen respiratory obstructive syndrome. Discomfort and pain induced by swallowing disorders lead to a decreased intake and failure to thrive. Sucking and swallowing disorders alter mother-child relations and increase the risk of further anorexia even when organic problems are solved.

### Personal data. 30 children with CHARGE from 1 to 9 years

- |   |                                  |
|---|----------------------------------|
| ▪ Adapted diet and feeding procedures only        | 3 children                       |
| ▪ Nasogastric tube only                           | 7 children (3 precocious deaths) |
| ▪ Gastrostomy and GER surgical treatment          | 20 children                      |
| ▪ Mean age of artificial nutrition weaning        | 3 years (min 16 mo, max 7 yr)    |
| ▪ Mean age of normal eating recovery (if reached) | 5 years                          |
| ▪ Number who do not eat after 6 years of age      | 2 children                       |

## MEDICAL AND NON MEDICAL MANAGEMENT *primum non nocere*

Apart from surgical treatment of esophagus and upper airway anatomic malformations, swallowing disorders have no radical medical therapy. Spontaneous resolution of functional disorders of sucking and swallowing is generally good and the major challenge is to wait for their natural improvement in the best conditions. One should recognize feeding disorders early in order to prevent deleterious consequences, reduce all contributing factors, teach feeding by multisensory and physiotherapeutic means and support parents' psychologically:

- Ask the mother for sucking skills disorders, even in the neonatal period:
- Improve sucking and swallowing mechanic, when it is not too poor, by using soft and largely perforated nipples, thickened milk, small meals and avoiding forced feeding.

- If swallowing disorders have respiratory or growth consequences, enteral nutrition is required, either exclusively if aspirations are present or as a night supplement when possible. Enteral nutrition avoids the respiratory consequences of feeding aspiration and induces proper weight gain.
- The choice between nasogastric (NG) tube and gastrostomy depends on the age of the child and the severity of the symptoms. Initially, NG tube feeding is started but, after few months or earlier if the procedure is not well tolerated. If the troubles persist, NG tube should be changed to a gastrostomy combined with anti-reflux surgery.
- Good airway clearance is essential to allow swallowing. Tracheostomy is often necessary and does not alter swallowing. On the contrary, it provides better ventilation and allows tracheobronchial drainage.
- Non-medical management of feeding teaching is detailed in the parents' section.

## REFERENCES

1. Tellier AL, Cormier-Daire V, Abadie V et al. CHARGE syndrome: Report of 47 cases and review. *Am J Med Genet* 1998;76:402-9.
2. Byerly KA, Pauli RM. Cranial nerve abnormalities in CHARGE association. *Am J Med Genet* 1993;45:751-7.
3. Blake KD, Russel-Eggitt IM, Morgan DW et al. Who's in CHARGE? Multidisciplinary management of patients with CHARGE association. *Arch Dis Child* 1990;65:217-23.
4. Abadie V, Chéron G, Couly G. Le syndrome néonatal de dysfonctionnement du tronc cérébral. *Arch Fr Pédiatr* 1993;50:347-52.
5. Abadie V, Chéron G, Lyonnet S et al. Le dysfonctionnement néonatal isolé du tronc cérébral. *Arch Pédiatr* 1996;3:130-6.
6. Abadie V, André A, Zaouche A et al. The oro-oesophageal dyskinesia : a cause of infant feeding disorders. *J Ped Gastroenterol Nutr* (submitted)
7. Jean A. Brainstem control of swallowing : localization and organization of the central pattern generator for swallowing. *Neurophysiology of the jaws and teeth*. Ed A. Taylor Mc Millan Press 1990 pp 294-321.
8. Lin AE, Siebert JR, Graham JM. Central nervous system malformations in the CHARGE association. *Am J Med Genet* 1990;37:304-10.
9. Renault F, Raimbault J. Electromyographie faciale, linguale et pharyngée chez l'enfant : une méthode d'étude des troubles de succion-déglutition et de leur physiopathologie. *Neurophysiol Clin* 1992;22:249-60.

## **SWALLOWING AND FEEDING IN CHARGE SYNDROME: PARENT INFORMATION**

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### **NORMAL STRUCTURE AND FUNCTION**

Feeding is both one of the more essential and the more complex of the mammalian functions. Feeding of children can be divided into two periods :

1. The sucking and swallowing reflex period lasts from fetal life to the age of 6 months and essentially requires the good organization of sensory and motor pathways going and coming from the face and the mouth to the brain stem.
2. The corticalized feeding period, lasting from the middle of the first year to adulthood, involves voluntary processes for the feeding orally and are influenced by sensory, psychological and environmental factors. Adult feeding processes are matured between 2 and 3 years of age.

Moreover, appetite and caloric intake are regulated by several neural mediators coming from the intestinal tract and the CNS (hypothalamus, thalamus, cortex). For children with CHARGE syndrome, the main problems seem to occur during the first period, during the brain stem development and organization. We do not know specific treatment for these disorders so the aim is to wait for the second period when feeding improves on its own.

### **TESTS USED AND SPECIALISTS**

Swallowing disorders are generally very well evaluated by the mother. From their child's first days of life, mothers may observe poor sucking, inability for the baby to be breast fed, discomfort with bottle, unexplained cries, bottle refusal or prolonged feeding time, skin color changes during feeding, breathing difficulties or cough increased by feeding, frequent spitting up, nasal reflux or poor weight gain.

The only two obligatory investigations are, in my opinion, a chest x-ray to detect pneumonia aspirations and an examination of the airway by a competent pediatric otorhinolaryngologist (ENT).

Other investigations may be useful to characterize the origin, the mechanism and the severity of the symptoms. They are performed differently depending on the child and the medical team (see doctor section).

### **MANAGEMENT**

Swallowing difficulties are as badly tolerated by the child as by his/her mother. Young infants affected with sucking, swallowing and breathing difficulties are not able to grow, to develop and to explore their environment in a proper way. Moreover, swallowing and feeding disorders cause great parental anxiety, for both medical and psychological reasons.

Parents have an important role regarding swallowing difficulties in pointing out the signs to the doctor. Parents need to understand that what appear to be very drastic and aggressive techniques (nasogastric (NG) tube, gastrostomy, tracheostomy and no oral feedings) would improve their child's comfort. These medical options are not easy to accept, especially when the problems do not seem to be major ones. Performing surgery may make their child seem more gravely affected and give a feeling of failure to the caretaker. The advantages of these interventions are observed after they are done. Discussing options with other parents of children with CHARGE or looking at other children who have had these procedures may help the families during these periods.

Parents also have a great role in the non-medical management of these difficulties by learning how to prevent worsening of the feeding problems and learning early how to adapt the teaching of a normal feeding by working with a Feeding Team. First by their positive attitude without guilt that they are at fault, no forced feeding and no excessive anxiety. Second by their presence and involvement in medical care, using adapted bottles, breast exclusion, by handling, massage or any methods to keep close physical contacts with the baby.

### **Learning to feed by mouth**

After several months of poor coordination of sucking and swallowing, children may be frightened by food. The first step consists of avoiding bad experiences with food. Non-nutritive sucking with a pacifier is useful. Then, the child must get acquainted again with his feeding instrument, that is to say his face, lips, gums. These vulnerable areas have to be approached with soft massage, by mouth games, etc. and always with close contact with the child. The pleasure of feeding has to be taught using pleasant and intense smells (even if the child's sense of smell seems to be poor), colors or tastes.

Food with a smooth texture should be given with a soft plastic spoon. Problems tolerating solid food can last several years. From the end of the first year, solid pieces may be offered to the child, allowing himself the peace from his hand to his mouth, in a totally voluntary procedure.

After the period of aspiration risk, drinking must be taught too, playing with pure water in a glass. Chewing must be stimulated too by elastic texture, placed in the lateral part of the mouth.

The management of feeding problems is a subjective procedure, depending on team and culture and requiring multidisciplinary interveners. In our opinion, this management must be a medical concern from the first week of life of a children affected with CHARGE syndrome.



## CARDIOLOGY IN CHARGE SYNDROME: FOR THE PHYSICIAN

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### TYPE AND FREQUENCY OF CONGENITAL HEART DEFECTS (CHDS) IN CHARGE

- ◆ 75% - 85% have a CHD
- ◆ Almost every type of CHD has been described, including "typical" VSD, ASD, PDA. Many children have multiple, complex congenital heart defects.
- ◆ There are proportionally more conotruncal and aortic arch CHDs (30-40% of those with a CHD)

Type of CHD (below)	Lin	Wyse	Tellier	Round #
# patients studied *	53	50	40	
Conotruncal, aortic arch (1)	42%	47%	33%	30-40%
Simple shunts, R/L obstruction (2)	32%	50%	57%	40-50%
AV/endocardial cushion (3)	15%	10%	10%	10%
Miscellaneous (4)	10%		5%	5%

\* many had more than one CHD

(1) Conotruncal: tetralogy of Fallot, double outlet right ventricle, aberrant subclavian artery, right aortic arch, truncus arteriosus, interrupted aortic arch (type B), conoventricular VSD.

(2) Shunts: atrial septal defect, ventricular septal defect, patent ductus arteriosus, pulmonic stenosis/atresia, tricuspid stenosis/atresia, aortic stenosis, mitral stenosis, coarctation, hypoplastic left heart syndrome.

(3) AV canal: complete atrioventricular canal, atrial septal defect, primum.

(4) Misc: complex single ventricle, anomalous pulmonary venous return, others not specified.

### DIAGNOSTIC TESTS AND CAVEATS

1. Echocardiography . In addition to intracardiac anatomy, imaging should look for aortic arch anomalies, such as vascular ring and aberrant subclavian artery.
2. Catheterization provides additional information about pressures and anatomy. In one study, 1/4 of the children with CHD also had renal anomalies. Although renal ultrasound and IVP/VCUG are more definitive, **delayed abdominal films** at the time of a cath may be useful in screening for urinary tract malformations.
3. Electrocardiogram (ECG) to study electrical activity. May be supplemented by longer term Holter monitoring.
4. Occasionally: treadmill stress testing, MRI scanning.

## MEDICAL MANAGEMENT WITH CAVEATS

1. In one study of 50 CHARGE patients with CHD, 75% required surgery.
2. Children with CHARGE may be resistant to chloral hydrate sedation.
3. Anesthetic risk is increased in children with airway involvement such as **choanal atresia**, or **laryngotracheomalacia** (both common in CHARGE). Children with choanal atresia and complex heart defects have the highest rate of serious complications and/or poor outcome.
4. Swallowing problems with increased secretions (presumably due to involvement of cranial nerves IX and X) may present an additional risk of aspiration.
5. Prostaglandin administration may be risky because of a high mortality in CHARGE following prostaglandin administration.
6. Hypocalcemia may be due to the absence of the parathyroids in the small number of CHARGE children with classic DiGeorge sequence. However, hypocalcemia may also be due to hypoparathyroidism in other patients.
7. Heart problems are only one component of growth failure. Others include feeding problems, frequent illnesses (especially chronic otitis media and respiratory infections), and possibly growth hormone deficiency.

## NON-MEDICAL ISSUES

Many children with CHARGE syndrome have dual sensory impairment (hearing and vision loss) in addition to the medical problems. This combination (medical concerns along with sensory impairment) has a tremendous impact on development. All of these children will be developmentally delayed. With appropriate medical and educational intervention (including a deafblind specialist), many of these children will eventually function in the normal to above normal range of intelligence. Do not assume mental retardation based on early developmental delay.

## REFERENCES

- Blake KD, Davenport SLH, Hall BD et al. CHARGE association: an update and review for the primary pediatrician. Clin Pediatr 1998;37:159-174.
- Lin AE, Chin AJ, Devine W et al. The pattern of cardiovascular malformations in the CHARGE association. Am J Dis Child 1987;141:1010-1013.
- Blake KD, Russell-Eggitt IM, Morgan DW, et al. Who's in CHARGE? Multidisciplinary management of patients with CHARGE association. Arch Dis Child 1990;65:217-223
- Tellier AL, Cormier-Daire V, Abadie V, et al. CHARGE Syndrome: report of 47 cases and review. Am J Med Genet 1998;76:402-409.
- Wyse R, Al-Mahdawi S, Burn L, Blake KD. Congenital heart disease in CHARGE association. Pediatr Cardiol 1993;14:75-81

## CARDIOLOGY: THE HEART IN CHARGE SYNDROME: PARENT INFORMATION

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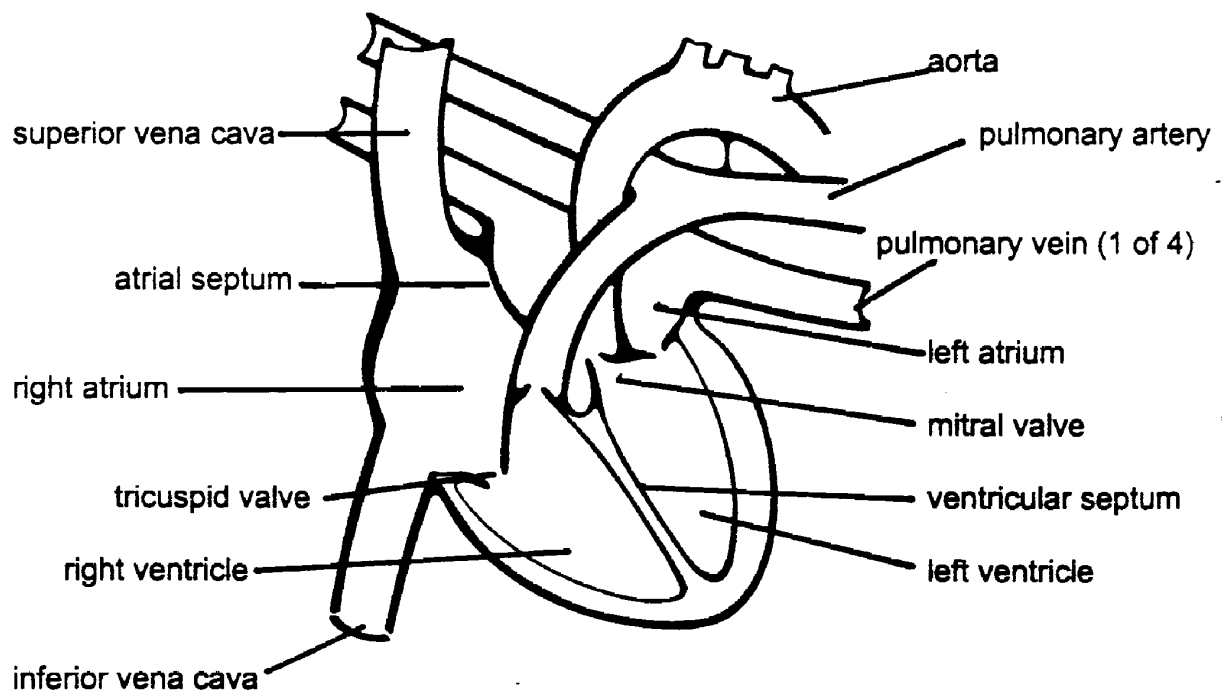
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### NORMAL STRUCTURE AND FUNCTION OF THE HEART

The heart is a muscular pump (myocardium) with inflow & outflow pipes (blood vessels). It can be thought of as a house with:

- ◆ Two upper rooms (right atrium [RA], left atrium [LA])
- ◆ Upstairs dividing wall (atrial septum [AS])
- ◆ Two lower rooms (right ventricle [RV], left ventricle [LV])
- ◆ Downstairs dividing wall (ventricular septum [VS])
- ◆ Two one-way swinging doors between upstairs and downstairs rooms (tricuspid valve, mitral valve)
- ◆ Large pipes entering upper right room from body (superior vena cava, inferior vena cava)
- ◆ Smaller pipes entering upper left room from lungs (pulmonary veins)
- ◆ Two large pipes, one exiting each ventricle (pulmonary artery, aorta)
- ◆ Electrical system (conduction system)
- ◆ Internal plumbing (coronary arteries) which supply blood to the heart muscle itself



## BLOODFLOW IN THE NORMAL HEART:

- 1) Blue blood returns from the body through veins to the **superior and inferior vena cava**, which empty into the **right atrium**. This blood passes through the **tricuspid valve** to the **right ventricle**.
- 2) The right ventricle pumps the blood through the **pulmonary valve** into the **pulmonary arteries**, which carry the blue blood to the lungs. The blood picks up oxygen in the lungs and turns red.
- 3) Red blood returns from the lungs through the **pulmonary veins** to the **left atrium** of the heart. From the left atrium, the blood passes through the **mitral valve** into the **left ventricle**.
- 4) From the left ventricle, the blood is pumped through the **aortic valve** into the **aorta** and from there throughout the body.

## TYPES OF CONGENITAL HEART DEFECTS (CHD)

A. Holes in dividing walls, which can allow blue and red blood to mix

**ASD:** atrial septal defect (hole between upper rooms)

**VSD:** ventricular septal defect (hole between lower rooms)

**AV canal/cushion defect:** atrioventricular septal defect (large hole between upper rooms and lower rooms, including a hole in the floor)

B. Major plumbing problems with pipes, which can transport blood to incorrect chambers or restrict flow of blood through a vessel : \*\* These are very common in CHARGE

**Truncus:** arteriosus (1 pipe instead of 2 leaving the lower rooms)

**Transposition:** transposition of the great vessels (pipe positions switched leaving the lower rooms)

**ToF:** tetralogy of Fallot (1 pipe narrow, 1 pipe overriding, with hole in wall)

**DORV:** double outlet right ventricle (both pipes exiting leaving the right lower room)

**Coarct:** coarctation of the aorta (narrow pipe heading towards the body)

**Interrupted aortic arch** (pipe flow cut off heading towards the body)

**TAPVR:** total anomalous pulmonary venous return (pipe hooked to wrong room)

C. Leaky valves, which can allow backflow of blood through the doorway

**Tricuspid or mitral regurgitation** (backflow between lower and upper rooms)

**Pulmonic or aortic regurgitation** (backflow into lower room from pipe)

D. Tight valves, which can restrict flow

**Tricuspid or mitral stenosis** (narrowing of doorways between upper and lower rooms)

**Pulmonic or aortic stenosis** (narrowing of pipes)

E. Abnormal rooms

**Single ventricle** (one large lower room)

**HLH:** hypoplastic left heart syndrome (very small lower left room)

**HRH:** hypoplastic right heart syndrome (very small lower right room)

## CHDs in CHARGE syndrome

How common are heart defects in CHARGE?

Approximately two-thirds (60-80%) of children with CHARGE have a CHD. Many, but not all, of them are serious.

Is there a typical pattern of heart defects in CHARGE?

Any heart defect is consistent with CHARGE, but ToF, DORV, and VSD are especially common. Some children with CHARGE have extremely complex heart defects that do not easily fit into just one category.

Are the typical heart defects mentioned unique to CHARGE? No, they are also common in:

**VCFS/DiGeorge complex/del22q11**

**Hemifacial microsomia/Goldenhar syndrome**

**Retinoic embryopathy**

## DIAGNOSTIC TESTS WHICH MIGHT BE DONE

Chest x-ray

Electrocardiogram (EKG), Holter monitoring

Echocardiogram (echo, ultrasound)

fetal/prenatal

postnatal

Cardiac catheterization (cath)

Exercise test (stress test)

## MANAGEMENT OF CONGENITAL HEART DEFECTS

### Medication

- ◆ digoxin, to help the heart pump stronger
- ◆ diuretics, to get rid of extra fluid
- ◆ antibiotics, to prevent infection
- ◆ anticoagulants, to thin blood

### Surgery

- ◆ to repair major plumbing problems (truncus, interruption of the aorta, ToF, DORV)
- ◆ to close holes in walls (ASD, VSD, AV canal)
- ◆ to repair loose valves (regurgitation)
- ◆ to repair tight valves (stenosis)
- ◆ to increase aorta blood flow (coarctation)

## OUTCOME OF CHDS IN CHARGE

### Medical outcome

Some heart defects can be totally repaired by surgery, while others can only be improved. Some children will end up with no heart problem at all, some will be much better, and others will continue to have problems with their heart. A few children with CHARGE will remain medically fragile for a long time, in part due to the heart defects.

The outcome and risks associated with heart surgery in CHARGE depend on the type of defect, the type of surgery, and on the presence of other serious health problems, especially choanal atresia tracheo-esophageal atresia, or cleft lip/palate. Many children with CHARGE have unusual reactions to anesthesia.

### Developmental effects of heart defects

Remember that any child who is in the hospital for an extended period of time or who undergoes many procedures is under great stress. One outcome of the stress is delayed development. Some of the early delayed development in children with CHARGE may be attributed to multiple hospital stays and multiple surgical and diagnostic procedures. Some children actually lose milestones while hospitalized, only to regain them later. Do not be discouraged by early developmental delays, however extreme. Many children with CHARGE are truly "delayed" and will catch up over the years to come.

# URINARY TRACT ANOMALIES IN CHARGE: FOR THE UROLOGIST

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## TYPE AND FREQUENCY OF URINARY TRACT ANOMALIES IN CHARGE SYNDROME

- ◆ 20-40% have a urinary tract anomaly (1,4).
- ◆ All types of structural problems (solitary kidney, hydronephrosis, renal hypoplasia, duplex kidney, posterior urethral valves, etc.) have been reported, as well as vesico-ureteral reflux.
- ◆ Facial palsy is significantly associated with renal anomalies, which are usually ipsilateral to the palsy (2)
- ◆ Children with CHARGE are at increased risk for urinary tract infection. Evaluation of a patient who has fever without a source should include a urinalysis and urine culture.

## Diagnostic tests

1. Baseline renal and bladder ultrasound to evaluate anatomy are warranted, as early identification and treatment may reduce long-term morbidity (3,4).
2. Functional studies (such as voiding cystourethrogram) may be indicated as follow-up to anatomic study, or if the patient develops urinary tract infection.

## Medical management and caveats

1. Standard medical therapy is acceptable for treatment of UTIs. Suppressive therapy may be necessary if infections are recurrent.
2. Surgical intervention is indicated for certain abnormalities. Indications for surgery are the same as for children who don't have CHARGE. Caveats for surgery include:
  - a. Anesthetic risk is increased in children with airway involvement such as **choanal atresia** or **laryngotracheomalacia** (both common in CHARGE). Children with

choanal atresia and complex heart defects have the highest rate of serious complications and/or poor outcome.

- b. Swallowing problems with increased secretions (presumably due to involvement of cranial nerves IX and X) may present an additional risk of aspiration.

#### **D. References**

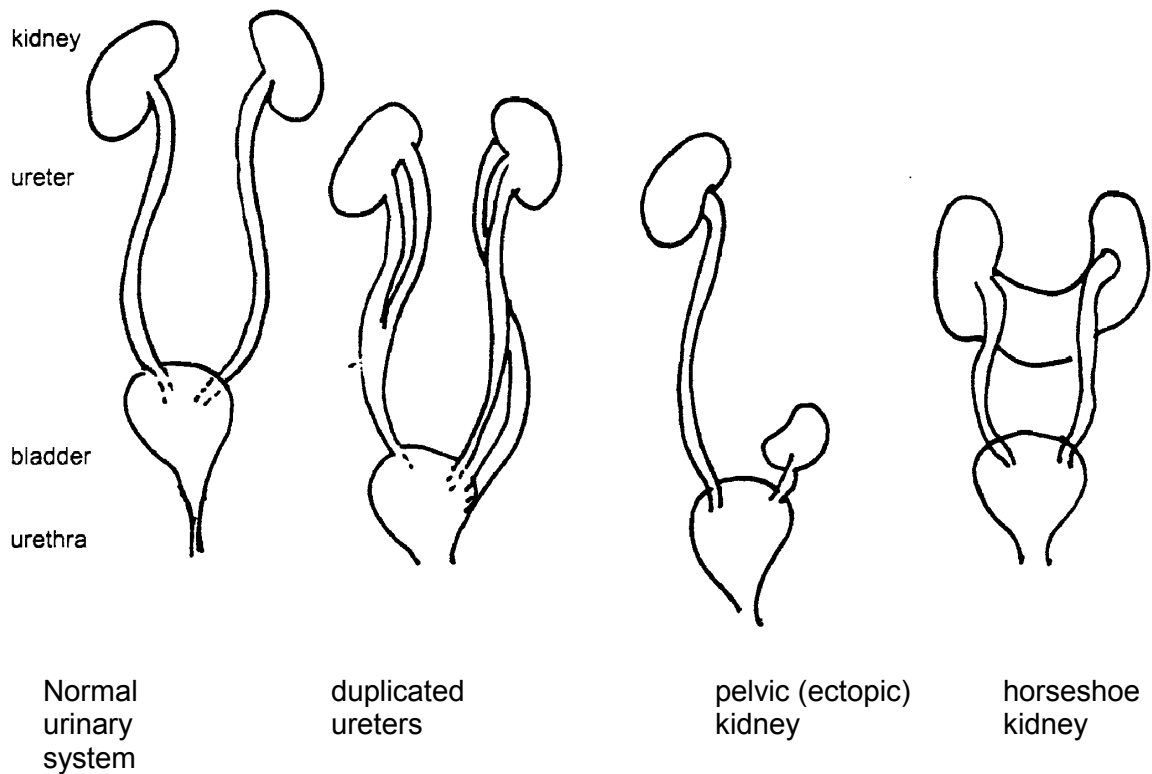
1. Tellier AL, Cormier-Daire V, Abadie V, et al. CHARGE Syndrome: Report of 47 cases and review. *Am J Med Genet* 1998;76:402-409.
2. Blake KD, Russell-Eggitt IM, Morgan DW, et al. Who's in CHARGE? Multidisciplinary management of patients with CHARGE association. *Arch Dis Child* 1990;65:217-233.
3. Blake KD, Davenport SLH, Hall BD, et al. CHARGE Association: An update and review for the primary pediatrician. *Clin Pediatr* 1998;37:159-174.
4. Ragan DC, Casale AJ, Rink RC, Cain MP, Weaver DD. Genitourinary anomalies in the CHARGE association. *J Urology* 1999;161:622-625.



## THE URINARY TRACT IN CHARGE SYNDROME: PARENT INFORMATION

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### STRUCTURE AND FUNCTION OF THE URINARY TRACT



The normal urinary tract consists of two **kidneys**. Urine is produced in the kidney and is drained by the **renal pelvis**. Urine then flows into the **ureters**. These tubes drain into the **urinary bladder**. When we urinate, the urine travels through another tube called the **urethra**. This tube is very short in females and longer in males (as it has to travel through the length of the penis). Urine normally only flows in one direction.

## URINARY TRACT ABNORMALITIES IN CHARGE

Abnormalities of the urinary tract are seen in 20-40% of children with CHARGE syndrome. They can be of two types, structural or functional.

Structural abnormalities (urinary tract isn't formed properly).

1. Absent or hypoplastic kidney - one kidney is not present or small. If a child is missing both kidneys they cannot survive.
2. Ectopic, or pelvic kidney - kidney is in an abnormal location, much lower than it should be.
3. Horseshoe kidney - the two kidneys are joined together to make one kidney shaped like a horseshoe.
4. Duplications (kidney, renal pelvis and/or urethra can be partially or completely duplicated). Obstruction to urinary flow is frequently seen.
5. Hydronephrosis or enlargement of the renal pelvis - excess fluid into the kidneys. This can be caused by a blockage between the renal pelvis (kidney) and the urethra (which is called a ureteropelvic junction, or UPJ obstruction), or by severe vesicoureteral reflux (see below).

Functional abnormalities (urine doesn't flow properly)

1. Vesicoureteral reflux (hereafter referred to as reflux). When the bladder contracts to assist urination, urine normally flows out the urethra. The bladder wall pinches off the ureters to prevent urine flowing backwards towards the kidneys. If the ureters remain open during bladder contraction, urine can flow back towards the kidney. This is reflux. It can be mild to severe. Reflux can predispose to urinary tract infection (UTI). If severe, it can result in hydronephrosis and direct damage to the kidneys, which can ultimately lead to kidney failure.
2. Bladder residual. Bladder does not empty completely. This can predispose to urinary tract infections.

## DIAGNOSTIC TESTS

1. Renal Ultrasound identifies structural abnormalities.
2. Voiding Cystourethrogram (VCUG) This test is indicated if reflux is suspected. It involves placing a small tube (catheter) through the urethra into the bladder. The bladder is filled with a liquid that can be seen on X-ray (contrast). X-rays are taken while the child urinates. This test is necessary to diagnose reflux or bladder residual.
3. Intravenous Pyelogram (IVP) This is another test primarily used to examine kidney structure. Because it involves the injection of contrast into a vein as well as X-ray exposure, this test has for the most part been replaced by the renal ultrasound. It does have the advantage of requiring intact blood supply to the kidneys and provides information regarding the kidney's ability to make urine (which neither #1 or #2 do). IVP can be used to determine if duplicated kidneys are functioning.
4. Radionucleotide renal scan. This test involves the injection of a radioactive material into a vein that concentrates in the kidney. Indications for the test are similar to #3, but this test gives more information about function and less information about structure than the IVP. The amount of radioactive is small and it is rapidly eliminated in the urine so radioactivity dose is small.
5. Computerized tomography (CT) and Magnetic Resonance Imaging (MRI). Both of these tests provide high-resolution images that allow much better definition of structure than the renal ultrasound. One of these would be indicated if the renal ultrasound was unable to resolve a complex structural abnormality of the urinary tract. The CT scan uses X-rays, but usually does not require sedation as it scans very rapidly. The MRI uses magnetic fields (which do not have harmful effects on body tissues) and has better resolution than CT scan. It can also be used to evaluate blood vessel anatomy (Magnetic Resonance Angiography or MRA). The scan time is longer, so young child frequently requires sedation. This is an important issue in CHARGE children due to the increased risk of airway compromise (see section 5f). We recommend full airway precautions, as discussed elsewhere, if sedation is to be used.

## MANAGEMENT OF URINARY TRACT ABNORMALITIES

1. All CHARGE children should have a baseline renal ultrasound done in infancy looking for structural abnormalities.
2. Any child who has fever without an identifiable source on physical examination should have a clean urine specimen obtained looking for UTI. This may involve placing a catheter through the urethra to obtain a specimen or sticking a needle through the abdomen into the bladder (suprapubic or bladder tap). Bagged specimens are inadequate for this purpose. Older children may be able to provide a clean voided specimen. Urine culture must be done to confirm UTI.
3. UTIs can be treated with standard antibiotics. A documented (urine culture positive) UTI is an indication for VCUG.
4. Recurrent UTIs may require use of a daily antibiotic. This is called suppressive or prophylactic therapy. If a VCUG has not been done, it must be done at this point.
5. Surgery is indicated to correct some structural anomalies, and may be indicated in severe cases of reflux. An urologist with pediatric experience should assist with this decision. Follow all surgical and anesthetic precautions as discussed elsewhere.

## OUTCOME

Aggressive medical and surgical intervention should be able to prevent or treat most complications of these abnormalities. Rarely, anomalies may be so severe as to inevitably progress to end stage renal disease (ESRD also known as kidney failure), which may necessitate consideration of dialysis or kidney transplant. There is only a single report in the medical literature of this occurring in CHARGE syndrome. (This was in an adult in whom CHARGE syndrome was not recognized until he presented in renal failure. The authors felt that if the syndrome had been recognized early on, diagnostic test could have been done that may have prevented the renal failure).

## ENDOCRINE PROBLEMS IN CHARGE FOR THE PHYSICIAN

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Although genital anomalies are common in CHARGE and make up part of the diagnostic criteria (minor criteria), there are few data on their management and outcomes, especially long-term. As a result much of the data is anecdotal, and recommendations are therefore based more on experience in groups of children with other similar anomalies. Recent studies have however confirmed delayed or absent puberty (more so in boys than girls) Blake et al., 2005.

### TYPE & FREQUENCY OF GENITAL ANOMALIES IN CHARGE SYNDROME

	<i>Frequency</i>
<b>Males:</b>	
Micropenis	85%
Undescended testes	60%
<b>Females:</b>	
Hypoplastic labia	Very common
Delayed/arrested puberty (males and females)	Very common
Infertility	Unknown

### STUDIES TO CONSIDER

It is thought that the majority of children with CHARGE have isolated hypogonadotrophic hypogonadism (i.e. pituitary gonadotropin deficiency) to account for their genital anomalies. For instance, although a number of different factors are involved in the descent of the testes and the development of the penis, it is clear that gonadotropins (mediated through testosterone) are required. Investigation is therefore aimed at:

- Assessment of gonadotropin function.
- Assessment of gonadal dysfunction arising from:
  - 1) Gonadotropin insufficiency
  - 2) Undescended testes in males

## INFANTS

Postnatally normal infants show a rise in LH, FSH and testosterone in males and estrogen in females which peaks at 8 weeks and subsides by 6 months. This “mini-puberty” offers a window of opportunity to investigate these children using baseline bloods alone in the six months of life. In males a peak serum testosterone concentration over 100 ng/dl can be regarded as normal.

## CHILDHOOD & ADOLESCENCE

Between the ages of 6 months and the onset of puberty the levels of gonadotropins, testosterone and estrogen remain low. At this age the following tests are recommended:

1. LHRH (GnRH) stimulation test, looking at pituitary gonadotropins (LH & FSH).
2. hCG test (in males only). Human chorionic gonadotropin – equivalent to LH, tests Leydig cell function alone. There should be a three-fold rise in testosterone following the injections; the response reflects the amount of functional testicular tissue.
3. For girls pelvic ultrasound should be used to assess the internal genitalia, and the response to therapy.
4. Where appropriate, tests of other pituitary hormones may be performed, such as baseline T4, T3, TSH, 9 am cortisol, prolactin, insulin-like growth factor-1 (IGF-1) and its GH-dependent binding protein (IGF-BP3), electrolytes and plasma/urine osmolality.
5. Formal anterior and/or posterior pituitary function.

## THERAPY

### Micropenis

If the underlying cause is thought to be hypogonadotrophic hypogonadism, then the treatment is testosterone replacement. This can be given by intramuscular injection (testosterone enanthate or propionate 12.5-25 mg 3-4 weekly for 3-4 doses). Topical testosterone cream 2% is also available and is administered once or twice daily for up to 3 months. Absorption may be erratic and (especially if female) the person applying the cream must ensure that they wear gloves. Some pubic hair growth may occur after testosterone administration. The most important single predictor of adult penile size appears to be the initial length of the penis.

It is unlikely that hypoplastic labia need any therapy.

## Cryptorchidism

The optimum timing and mode of therapy to bring down the undescended testis is contentious, even in “normal” boys. Histological changes occur in the cryptorchid testis within 1-2 years, although this must be balanced against the increased technical difficulties of surgery at younger ages. Even if the testes are not felt to have much potential for function, many surgeons would still perform orchidopexy to reduce the chances of detection of malignant change.

Hormonal therapy with hCG (human chorionic gonadotropin) may be appropriate (especially for palpable testes), and is usually given after the age of four years (with a 50% success rate). Traditionally 500-1000 IU is given intramuscularly twice weekly for 5-6 weeks. This may also cause increased penile growth as well as producing testicular descent.

## Delayed/absent puberty

### **MALES**

- Intramuscular testosterone enanthate or propionate, 50-250 mg monthly.
- Oral testosterone undecanoate 20-120 mg daily.
- Patches and long-acting subcutaneous pellets have been used in hypogonadal boys with some success.

Starting with low-dose oral testosterone (eg testosterone undecanoate (Restandol)) 40 mg alt die offers more flexibility initially in titrating/stopping if there are problems such as worsening behaviour.

### **FEMALES**

- Ethinyloestradiol initially 2 mcg/day, increasing over approximately 2-3 years to 10 mcg/day. When full pubertal progression has occurred or if there has been breakthrough bleeding then change either to low dose oral contraceptive pill (OCP) or adult HRT.

### **CAVEATS**

- Testosterone can cause fluid retention and should be used in caution in children with heart failure.
- As hCG testing only investigates the Leydig cell function of the testis, an absent response does not therefore necessarily confirm that there is no testicular function.
- hCG should not be used in girls as it can cause ovarian hyper-stimulation.

- Although relatively rare, hypopituitarism may also occur in CHARGE, and the following may indicate that further investigation of the other pituitary hormones is required:
  - Other midline defects such as clefting.
  - Conjugated hyperbilirubinemia in the neonatal period.
  - Hypoglycemia in the neonatal period.
  - Absence/hypoplasia of pituitary or other midline structures on brain scanning.
- Animal studies have suggested that early exposure to exogenous testosterone may down-regulate the androgen receptor, leading to poor penile size in adulthood. This has not been shown in human disorders where there is early exposure to androgens.
- There are concerns regarding long-term oral testosterone therapy as this has been shown to cause liver dysfunction in some patients.
- If the testes are retained intra-abdominally it is highly unlikely that fertility can be achieved, and even if the testes are brought down then fertility is likely to be compromised. It is also recognized that the risk of malignancy is higher in those with previously cryptorchid testes.

#### **References:**

Burstein S, Grumbach MM, Kaplan SL. Early determination of androgen responsiveness is important in the management of micropallus. *Lancet* 1979;2:983-986.

Goldson E, Smith AC, Stewart JM. The CHARGE Association; how well can they do? *AJDC* 1986;140:918-921.

Blake KD, Russell-Eggitt IM, Morgan DW et al. Who's in CHARGE? Multidisciplinary management of patients with CHARGE association. *Arch Dis Child* 1990;65:217-223.

Ragan DC, Casale AJ, Rink RC et al. Genitourinary anomalies in the CHARGE association. *J Urology* 1999;161:622-625.

Blake KD, Salem-Hartshorne N, Abi Duoud M, Gradstein J. adolescent and adult issues in CHARGE syndrome. *Clinical Pediatrics* 2005;44:51-159.



## **GENITAL/ENDOCRINE PROBLEMS IN CHARGE SYNDROME: PARENT INFORMATION**

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### **GENITAL ABNORMALITIES SEEN IN CHARGE**

Although genital abnormalities (minor diagnostic criterion) are common in CHARGE, there is very little information in the literature describing these problems or treatment. More importantly, as children may have other potentially more serious problems (especially at birth) the genital abnormalities are often not noted or treated.

In both sexes with CHARGE the main problem seems to be an abnormality in production of the hormones (chemical messengers) from the pituitary gland (a small pea-sized gland lying underneath the brain) which control the production of sex hormones from the testicles in boys, and ovaries in girls. This can result in:

- In boys, undescended testicles (one or both) and small penis (micropenis - less than 2.5 cm (1") stretched length) at birth. About three-quarters of boys with CHARGE will have micropenis. About half will have undescended testes. Both problems are due to the fact that descent of the testicles into the scrotum and growth of the penis in the last part of the pregnancy are dependent on the production of the hormone testosterone from the testicles (under the influence of pituitary hormones).
- In girls, the clitoris and labia minora (inner vaginal lips) may be smaller than usual, but this will not be as obvious as in the boys. It is very common in girls with CHARGE.
- In both sexes, failure or slow progress in puberty is very common, more so in boys than girls. This will mean that the boys will not get increase in the size of their testicles and penis, and the girls will not develop breasts or begin their periods without hormone treatments. In both sexes, there will be failure of development of pubic and axillary (armpit) hair.
- Infertility. The outlook for fertility in children with CHARGE is unknown.

## **TESTS WHICH MAY BE RECOMMENDED**

### **BASELINE BLOOD TESTS**

These measure the amount of hormones in the blood, either on a single sample (baseline levels) or after the levels are stimulated (usually by a chemical or hormone injection). The testicles and ovaries are both quite active in the months before and after birth, and baseline levels may be quite helpful. From about six months of age until puberty the production of sex hormones is very low, and therefore it is often necessary to measure hormones after stimulation.

- LH (luteinizing hormone) produced from the pituitary gland is responsible for the production of testosterone in boys, and for the production of estrogen and progesterone in girls.
- FSH (follicle stimulating hormone) produced from the pituitary gland is responsible for assisting in the production of sperm in the male, and ovulation in the female.
- Testosterone, the male sex hormone, produced from the testicles.
- Estrogen, the female sex hormone, produced from the ovaries: this causes breast development.
- Progesterone, another female sex hormone from the ovaries, when present along with estrogen produces menstruation.

### **STIMULATION TESTS**

- LHRH test. In this test the production of LH and FSH from the pituitary gland is measured using luteinizing hormone releasing hormone (LHRH) as the stimulatory hormone. This is usually done over one hour, with samples taken after 0, 30 and 60 minutes after intravenous injection of LHRH. In most patients with CHARGE there is little or no increase in LH & FSH during the test.
- hCG (human chorionic gonadotropin) test. By giving several injections of a substance similar to LH over several days, this measures the ability of the testicle to produce the male hormone testosterone.

Although hCG will also stimulate hormone production in girls it may cause ovarian over-stimulation and is therefore not usually used in girls.

### **IMAGING**

In girls ultrasound can be used to assess the size of the ovaries and uterus (womb).

In boys ultrasound may be used to assess the position of the undescended testicles, although other scans (CT, MRI) may be more accurate.

## **TREATMENT**

### **UNDESCENDED TESTICLES**

If the testicles are undescended they need to be brought down. This is usually done surgically, requiring one or more operations. Ideally this should be done as early as possible, although other medical problems of CHARGE and technical difficulties of performing surgery in young babies usually means that it is performed when they are older. Alternatively, the testicles can sometimes be brought down using hCG injections (usually twice weekly over 3-6 weeks), and there is some evidence that it works best in testicles which are not completely undescended, and after 4 years of age.

### **MICROPENIS**

A penis which at birth is less than 2.5 cm (1") (stretched) is defined as a micropenis. This should be treated with testosterone. This can be given as a cream applied twice daily for up to three months. As it is absorbed through the skin, the person applying it should wear gloves. Alternatively, testosterone injections can be given monthly for 3-4 months. Although there have been theoretical worries that early treatment will affect the growth of the penis during puberty, this does not appear to happen. *Testosterone can cause fluid retention and should be used in caution in children with heart failure.*

The small labia in girls are not usually treated.

### **DELAYED PUBERTY**

Although there may be worries about worsening behavioral problems during teenage years by treating with sex hormones, this must be balanced against the long-term risks of osteoporosis. The timing of treatment will take into account the normal age of puberty in boys and girls (from 11 years of age), but may be delayed to see if puberty occurs spontaneously.

**GIRLS:** Gradually increasing doses of estrogen in tablet form is used. Once full pubertal development has occurred, adding in progesterone either as hormone replacement therapy (HRT) or in the low-dose oral contraceptive pill (OCP) will produce periods if the uterus is of sufficient size.

**BOYS:** Gradually increasing doses of testosterone are given by injection, tablet or patch.

In both sexes it is likely that hormone replacement will need to be given long-term.



## **MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME: FOR THE PHYSICIAN**

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### **TYPE AND FREQUENCY OF MUSCULOSKELETAL ANOMALIES IN CHARGE SYNDROME**

- ◆ Prevalence of musculoskeletal anomalies is between 30 - 50%. This may increase as more cases of scoliosis are noted in older individuals.
- ◆ No consistent pattern of anomalies has been seen, although syndactyly of fingers or toes seems to be more frequent. Some of these patients have an atypical split hand deformity. There is often a specific palmar crease pattern with a so-called “hockey stick” distal palmar crease which is included in the minor diagnostic criteria.
- ◆ Severity of anomalies has ranged from very minor (dermatoglyphic anomalies to moderately severe (congenital hip dysplasia, syndactyly, polydactyly, clubfoot).
- ◆ A single patient is reported with absent muscles of one upper extremity. Muscle abnormalities have otherwise not been reported.
- ◆ Hypotonia, particularly of the upper body is frequent. This may be a neurologically-based problem or possibly a misinterpretation of the presence of ligamentous laxity. There have been no reported cases of a primary myopathy in CHARGE patients. Muscle biopsy would not be indicated, unless a second diagnosis is suspected.
- ◆ Scoliosis is frequent, beginning as young as 6-7 years.
- ◆ Osteoporosis may occur in adults with untreated hypogonadism.
- ◆ Information is derived from literature review of nearly 300 reported patients.

### **DIAGNOSTIC TESTS**

Careful physical examination of the musculoskeletal system is the only “test” indicated in all patients. It is important to screen for scoliosis beginning at school age.

Other diagnostic tests (X-ray, ultrasound) may be indicated based on physical findings.

X-rays obtained for other reasons (i.e. chest x-rays) should be examined carefully for definition of skeletal anatomy.

## **MEDICAL MANAGEMENT**

Treatment is anomaly specific. No differences in therapy are necessary if the patient is diagnosed with CHARGE syndrome. If surgery or sedation is necessary, anesthesia precautions are as discussed elsewhere.

## **REFERENCES**

Williams MS, Rooney BL. Limb anomalies in CHARGE association. Poster presentation 27th Annual March of Dimes Clinical Genetics Conference, 1996.

Tellier AL, Cormier-Daire V, Abadie V, et al. CHARGE Syndrome: Report of 47 cases and review. *Am J Med Genet* 1998;76:402-409.

Davenport SLH, Hefner MA, Mitchell JA. The spectrum of clinical features in CHARGE syndrome. *Clin Genet* 1986;39:298-310.

Harvey AS, Leaper PM, Bankier A. CHARGE association: clinical manifestations and developmental outcome. *Am J Med Genetics* 1991;39:48-55.

Oley CA, Baraitser M, Grant DB. A reappraisal of the CHARGE association. *J Medical Genetics* 1988;25:147-156.

## MUSCLES AND BONES IN CHARGE SYNDROME: PARENT INFORMATION

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### NORMAL STRUCTURE AND FUNCTION

The musculoskeletal (MS) system consists of over 200 bones and 500 muscles. A detailed description is beyond the scope of this manual. (Besides, I can never remember them all.)

Two aspects of the MS system are critical: **structure** and **function**. Normal **structure** means that all of the components of the system (that is the muscles and bones) are present and in the proper relationships to one another. Normal **function** means that the bones are maintaining the normal structural integrity of the skeleton and the muscles (through contraction) are able to move the portions of the skeleton that are supposed to move (the joints). All **structural** abnormalities lead to **functional** abnormalities (although the severity of the abnormality may not be clinically significant), but not all **functional** abnormalities lead to **structural** abnormalities. An example of this would be a person who suffers a spinal cord injury. The muscles and bones are completely normal, but the muscles are unable to function because of the absence of nerve signals.

Two other important concepts are **strength** and **tone**. Most people understand strength, but tone is a harder concept to grasp. **Strength** is the ability of a muscle, or group of muscles to work against a load. It can be objectively measured. **Tone**, however, is a subjective assessment of muscle function at rest. When we are resting (that is not moving a particular muscle or group of muscles) our muscles are not completely at rest. There is a baseline level of activity that can be detected, but is not easily measured. We refer to this as **muscle tone**. Muscle tone helps with posture and maintaining normal skeletal relationships. Tone can be normal, low or high. Low tone is also called **hypotonia**. Individuals with low muscle tone are often described as floppy. This can lead to slumped posture, problems with head control, standing, etc. High tone is called **hypertonia** or **spasticity**. Individuals with spasticity feel like the muscles are constantly contracting. This can lead to abnormal joint positions and result in toe-walking, hip and knee flexion contractures, etc. An experienced physician or developmental therapist can assess Tone, but there is no way to objectively measure muscle tone. Abnormalities in muscle tone can be due to abnormalities of the **nervous system** (brain, spinal cord, spinal nerves, peripheral nerves or connections between the nerve and the muscle), or to abnormalities within the muscle itself (**myopathy**). It may be difficult to distinguish between these two causes on clinical examination.

### TYPES OF MUSCULOSKELETAL ABNORMALITIES IN CHARGE

Between 30 and 50% of patients with CHARGE are born with some type of skeletal abnormality. Severity can range from clinically nonsignificant (minor changes of the creases of the palms due to short hand bones), to quite severe (missing fingers). Several patients have been reported to have fusion of fingers or toes (syndactyly) or clefting of the hand or foot.

Low muscle tone (**hypotonia**) is very common in children with CHARGE, especially in the upper body (trunk). There have not been any patients with CHARGE known to have hypotonia due to a myopathy (abnormality of the muscles themselves). Unless new information becomes available, it is probably safe to assume that the hypotonia is due to a central nervous system abnormality (i.e. brain). Low muscle tone may have an effect on development: if the upper body is floppy, it will be more difficult to sit alone or stand. Combine weak tone with vision loss and balance problems, and you may have a child who does not walk until age 5 or 6.

Scoliosis (curvature of the spine) is common in children with CHARGE. Although scoliosis is generally thought of as a teenage problem, it has been seen in young children with CHARGE. By the teenage years, a majority of individuals with CHARGE may have some scoliosis. This may be due, at least in part, to the low muscle tone in the upper body.

## **DIAGNOSTIC TESTS**

The most important diagnostic test in very young children is a careful physical examination of the musculoskeletal system. Imaging studies (such as X-ray, ultrasound) are indicated if there is suspicion of an anomaly on physical examination. Skeletal survey (to look at all the bones of the skeleton) is not routinely indicated. Tests of muscle (muscle biopsy, electromyogram (EMG)) are generally not indicated, unless a primary muscle problem is also suspected.

In older children, regular physical exam for scoliosis is indicated. If scoliosis is suspected, the doctor may order X-rays to determine the extent of the scoliosis.

## **MANAGEMENT AND OUTCOME OF MS ANOMALIES**

Medical and/or surgical management is based on the type of anomaly. They are not managed differently whether or not the child has CHARGE. Outcome following intervention is generally good, but clearly depends on the severity of the problem and the treatment that is required. Occupational therapy (OT) and physical therapy (PT) may be helpful in dealing with low muscle tone.



# GROWTH IN CHARGE: FOR THE PHYSICIAN

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Growth in children with CHARGE has special considerations in each of the three phases: Infant Phase, Childhood Phase, Pubertal Growth Spurt.

1. **Infant Phase:**

At birth, children with CHARGE Association usually have normal weights and lengths.<sup>1,2</sup> However, within the first 9 months of life there is a decline away from the normal growth curve, down to and often below the 3rd percentile. This pattern of growth may be related to repeat hospitalizations, poor feeding, major acute illnesses such as pneumonias and multiple surgeries. The infant phase of growth is mainly determined by nutrition and those children with CHARGE who maintain their weight in the early months are often the ones who have had major nutritional intervention, usually in the form of enteral feeding.<sup>1</sup>

2. **Childhood Phase:**

Although the growth rate is slower, the childhood phase of growth is the main determinant of final height as it lasts longer than the other phases. Adequate nutrition is important in this phase but also hormones play a role (thyroxine and growth hormone). Growth hormone deficiency is rare but there is an increased incidence in CHARGE.<sup>3,4</sup> Pre-adolescent children may have a more normal rate of growth although poor growth in infancy and lack of catch-up growth during childhood often result in a mean height at or below the 3rd percentile.

3. **Pubertal Growth Spurt:**

Growth deceleration associated with a delay in puberty occurs in more than 70 percent of children with CHARGE and this needs to be monitored closely. Growth in puberty involves the interaction of the sex steroids, especially testosterone and estrogen associated with growth hormone. There is anecdotal evidence that adolescents with CHARGE have fusion of their epiphyses at a later age and therefore often continue to grow in their early 20's.

The majority of children with CHARGE association have normal results on growth hormone stimulation testing. Arbitrary treatment with growth hormone in patients who have normal growth hormone levels has not been adequately studied and is probably not advised.

## **Nutrition    “Growth and Nutrition are Closely Related”**

Children need an adequate quantity and balance of food for optimal growth and development. CHARGE infants who have excessive surgery and a decrease in nutrition intake may require catch-up growth and their energy requirements may be 150 to 200 kcal/kg/d.

Reference Values for Energy and Protein Requirements		
Age	Energy kcal/kg/d	Protein gm/kg/d
0-6 months	115	2.2
6 mts.-6 yrs.	95	1.8
7 yrs-10 yrs.	75	1.2
11-16 yrs.	60	1

## FEEDING ISSUES IN INFANCY AND EARLY CHILDHOOD

### Symptoms and Behaviors of Swallowing Problems (Also see Swallowing Section):

1. The feeding history may include:  
Cough and choking, nasopharyngeal reflux, nasal congestion, food suctioned from the nose or from the tracheostomy.
2. Chronic chest difficulties.  
Recurrent pneumonias, apneas, frequent upper respiratory tract infections.
3. Other behaviors.  
The infant may have stresses on feeding such as sweating, gasping, straining, prolong or shortened sucking pattern. In children, other symptoms may be exhibited i.e. build-up of secretion, spiking temperatures, food remaining in the mouth, choking, eyes tearing or lurching the head forward and facial stress.

More than 90 percent of children with CHARGE have difficulty swallowing foods of different textures. As a consequence parents often discontinue trying to feed their children solid foods.

Children with bilateral posterior choanal atresia often have the most severe feeding problems and may manifest significant failure to thrive.<sup>4</sup> In some instances, there may be obvious physiological reasons for these problems, i.e. cleft lip and palate, facial palsy, choanal atresia, tracheo-esophageal fistula. Occult and hidden anatomical abnormalities of the pharynx and larynx have also been described.<sup>4</sup> These need to be considered in children and infants with feeding problems. One of the most difficult chronic management problems that families have to face is gastro esophageal reflux. There is no quick fix to the problem and different centers offer their expert team.

When gastrostomy tubes are used, children run the risk of oral pharyngeal hypersensitization, meaning that they reject substances and objects in their mouths, especially feeding devices. It is therefore important to try and keep stimulating the oral pharynx even though the child is being fed by gastrostomy or jejunostomy tube. Even with aggressive therapy many children with CHARGE continue to have feeding problems into pre-adolescent and beyond. Parents also report excessive abdominal colic similar of the type commonly found in infancy, which may present to school age children and beyond. The etiology of this colic is unknown but is probably organic in nature.

### **General Management in Feeding a CHARGE Infant**

- Positioning of infant or child
- Use of different textures to find the ones that work
- Aspiration precautions
- Aggressive management during colds
- All of the above can be guided with a feeding team which should include an occupational therapist, physiotherapist, speech therapist, behavioral psychologist, and dietician.

**Breast Feeding** - often difficult in CHARGE infants because of their many surgeries but not impossible especially if expressed breast milk is used.

### **Weaning:**

Solid foods are usually introduced between the ages of 3 and 6 months. After 6 months of age, milk only can lead to anemia and deficiencies in vitamins and iron. Lumpy foods, which are usually introduced at about 6 months, are hard for CHARGE children to manage. Different textures and tastes may also be a problem. Many families use pureed foods until childhood.

### **Failure to Thrive:**

All children with chronic illness can have failure to thrive. Congenital heart disease or respiratory disorders alone can be responsible for failure to thrive. In CHARGE children, malnutrition may result from a combination of decreased intake, malabsorption and increased requirements because of increased work of breathing. Malnutrition from many of the aspects of CHARGE association can respond to enteral feeding.

Malnutrition must be recognized and accurately defined so decisions can be made about feeding. Evaluations are divided into assessment of past and present dietary intake, measurements and lab assessments as in the table above. The consequence of malnutrition is a multi-system disorder. Malnutrition worsens the outcome of illness, respiratory muscle dysfunction and may delay a child being weaned from a mechanical ventilator. Malnourished children are less active, less exploratory and more apathetic. These behavioural abnormalities are rapidly reversed with proper feeding.

Assessment of Nutritional Status
<b>Anthropometry</b> <ul style="list-style-type: none"><li>· weight</li><li>· height</li><li>· mid arm circumference</li><li>· skin fold fitness</li></ul> <b>Lab Data</b> <ul style="list-style-type: none"><li>· low plasma albumin</li><li>· low concentration of specific minerals and vitamins</li></ul> <b>Food Intake</b> <ul style="list-style-type: none"><li>· dietary recall</li><li>· dietary diary</li></ul> <b>Immunodeficiency</b> <ul style="list-style-type: none"><li>· low lymphocyte site count</li><li>· impaired cell mediated immunity</li></ul>

### **Short Stature or Failure to Thrive:**

Short stature and failure to thrive are usually defined as height or weight below the 2nd or 3rd percentile (approximately 2 standard deviations below the mean). Serial measurements are the most helpful as they allow detection of change in the rate of growth. Growth failure can be defined from the child's growth parameters falling across percentile lines plotted on a growth chart. In this way, growth failure may be identified even though the child's height and weight are still above the second or third percentile. As most children with CHARGE have a normal birth weight and length, then it is often obvious within the first 9 months when they fall away from the middle percentiles. CHARGE children who suffer from malnutrition and chronic illness are usually moderately short but significantly underweight. In contrast, if growth hormone deficiency is the cause for growth delay, children are usually somewhat overweight for their height.

Short stature may cause psychological problems. Adults assume that the children are younger than their true age and so treat them inappropriately.

### **Growth Hormone**

Diagnosis of growth hormone deficiency is difficult because of the pulsator nature of growth hormone secretion. In pituitary provocation tests, a variety of stimuli are used to provoke growth hormone release. The most common is clonidine, glucogone and insulin induced hypoglycemia. The hypoglycemia induced by insulin is potentially dangerous. To avoid these complications, the test should only be performed in specialist centres. Growth hormone deficiency is treated with biosynthetic growth hormone, which is given by subcutaneous injection usually daily. There are other potential applications of growth hormone therapy, which is under study including children with different syndromes and those with chronic illness. CHARGE would fall within this realm, however, growth hormone is not being used at present for children who have not been diagnosed with growth hormone deficiency.

### **REFERENCES:**

1. Blake K, Kirk JM, Ur E. Growth in CHARGE association, Arch. Dis. Child. 1993;68:508-509.
2. Harvey AS, Leaper PM, Bankier A. CHARGE Association: Clinical Manifestations and Developmental Outcome. 1991;39:48-55.
3. August, PA, Rosebaum KN, Friendly, D. Hypopituitarism and the CHARGE association. J. Pediatr. 1983;103:424-425.
4. Blake KD, Russell-Eggitt IM, Morgan DW et.al. Who's in CHARGE? Multi-disciplinary management of patients with CHARGE association. Arch. Dis. Child. 1990;65:217-223.
5. Davenport SLH. Understanding neurologic impairment is key to improved prognosis in CHARGE syndrome. Topics in Pediatrics. 1993;11:11-14.

## FEEDING AND GROWTH IN CHARGE: PARENT INFORMATION

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Although some children with CHARGE have growth hormone deficiency, much of the slow growth in CHARGE is likely due to nutritional status and general medical problems.

Difficulty in feeding is one of the most common and prolonged problems in CHARGE. As a consequence of poor feeding, the long term poor nutrition results in growth failure and can contribute to the developmental delay. Assessment of growth is important and below are the growth parameters that should be measured routinely in your children (3-4 times per year):

1. Weight - If possible, measurements should be on the same weight scales.
2. Length or Height - In children under 2 years old, length is measured lying horizontally. Because of balance problems and delayed mobility, children who are not comfortable standing to be measured should also be measured horizontally.
3. Head circumferences should be measured in the first 2 years of life. This is the widest circumference around the head called the occipital frontal circumference and is an indication of brain growth and overall size.
4. The measurements of growth parameters should be plotted on percentile charts. It is wise to keep a copy in your binder of your own child's growth progression and ask to be shown how to plot and read the charts.
5. Clinic - It is important to attend a growth and/or endocrine clinic. If growth hormone deficiency is suspected, your doctor may order special testing.

### **Feeding and Swallowing In Children With CHARGE (Also see Swallowing Section):**

Feeding problems are frequent and represent a permanent concern for most parents. Feeding studies may show that the infant can suck normally but liquid pools in the back of the throat without passing smoothly into the esophagus. Swallowing can be uncoordinated and also lead to gastroesophageal reflux. Children are often uncomfortable, show signs of restlessness when they are being fed, they may gag and aspirate (the food or liquid goes down into the lungs). These children have a hard time feeding orally. Some can handle pureed foods but cannot tolerate liquids or solids. It is important to find the consistency and type of food that your child will tolerate. Positioning and behavior modification when feeding are also important. A team of therapists should be involved with your child who has feeding problems. A therapy team includes a speech-language pathologist, an occupational therapist, a psychologist and a physician with some interest in feeding problems. If there is a center where there is a feeding team, this center should be consulted.

### **ANATOMICAL ISSUES WHICH HAVE AN IMPACT ON SWALLOWING:**

- ◆ Retrognathia - posteriorly placed mandible
- ◆ Micrognathia - underdeveloped mandible (jaw bone)
- ◆ Tracheoesophageal fistula (TEF) - presence of a fistula (a joining) between the tracheal and esophageal walls.
- ◆ Cleft lip and Palate - refers to a "split" or "separation" in the lip and/or palate
- ◆ Facial palsy - usually on one side but can be both sides. The side of the paralysis is the side where the eye has incomplete closure - more noticeable when the infant is crying.
- ◆ Choanal Atresia or stenosis

- ◆ Lower Cranial Nerves Involved in Swallowing and Sucking (IX X XI) - affects sensory (feel) and motor function (activity) involving sucking and swallowing.

## TESTS USED TO EVALUATE SWALLOWING:

### ***Barium Swallow:***

Assessment of anatomical structure function  
Examines the esophagus pharynx, and larynx  
Uses large amounts of liquid purees, and solids  
Positions the patient in the supine (lying down)  
Follows the bolus (watch as food goes down)

### ***Videofluoroscopic Swallow Study (VFSS):***

Assessment of swallowing  
Examines the oral cavity  
Uses small amounts of liquid  
Positions patient in the upright position  
Does not follow the bolus

## ALTERNATIVE TYPES OF FEEDING:

**Nasogastric Tube** - For short periods and for supplementation. This may not be a safe method for a child with a compromised swallow because of increased secretions produced by nasogastric tube, which can increase risk of aspiration.

**Gastrostomy Tube or Button** - Gastrostomy tube or button is a preferred choice for long-term supplementation if stomach function is intact.

**Jejunostomy Tube** - Preferred choice if severe gastroesophageal reflux is present.

### Effects of Tube Feeding

1. Hypersensitivity of the oral cavity - as the mouth is not being stimulated, child gets sensitive to anything that is put in the mouth. Desensitizing the oral cavity is important. Always encouraging textures, tastes and sensitizing procedures even if your child is not feeding orally. (Seek the expertise of an occupational or speech therapist.)
2. After tube feeding is started even for a short period of time it is difficult to get the child back on to oral feeds.
3. Lumps and different textures may remain a problem into later childhood.
4. The tube may not work, dislodge, and need replacing. Granulation tissue (healthy scar tissue) can collect around the tube and so can infection.
5. Parents like the button as it gives their child better mobility and freedom to move.

## THE CHARGE ADOLESCENT: FOR THE PHYSICIAN

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The adolescent period is a stage of change and offers its own challenges for all families but especially to those with CHARGE teens.

Behavioral problems can be a big issue. These may be outbursts, tantrums, self-abuse, and defiance.<sup>1</sup> For a non-CHARGE adolescent unruly behavior is more likely due to psychological concerns than from medical ones. With the CHARGE adolescent a medical cause needs to be considered first especially if these outbursts occur without any provocation or warning and are waking the adolescent from sleep<sup>3</sup>.

There are certain co-morbid psychological conditions that can occur with CHARGE. These may be detected before adolescents and these include:

- Attention deficit disorder (ADHD)
- Pervasive developmental disorder (PDD)
- Obsessive compulsive disorder (OCD)
- Anxiety
- Depression
- Learning difficulties

If it is suspected that the CHARGE adolescent has one of these co-morbid conditions secondary to the CHARGE diagnosis, it is important to have a full psychological evaluation including a detailed home and school profile. The psychologist should have understanding of adolescents with sensory deficits. It is important to take care of the above labeled diagnosis as it is easy to overlook sensory deficits i.e. hearing and visual loss.

Drugs used to treat these co-morbid conditions may be required in smaller doses than for non-CHARGE adolescents. Before starting medication, it is best to treat the adolescent not the label. Any of the listed diagnoses often gives rise to adverse behaviors but so do frustrations in not being able to communicate or frustration in this non-CHARGE world. It is important to observe when these types of behavior are occurring and to try behavior modification techniques along with a very structured day.<sup>4</sup> Planning a daily routine and preparing the adolescent for any change may be a good strategy. Positive rewards for good behavior and a routine are a very good start to a behavioral modification program. The adolescent with CHARGE responds like many other non-CHARGE adolescents and would benefit from having a set of rules about behavior that is and is not acceptable,<sup>1</sup> e.g. The parent may tell their teen they can touch a person's hand, shoulder and arm, but that they are not allowed to touch peoples breasts.<sup>1</sup>

Observations about behavior in CHARGE adolescents directly from a handout from Veronika Bernstein<sup>1</sup> are as follows:

- ✓ *Denotes behaviors which can occur in any adolescent.*
  - ✓ mood disturbances
  - ✓ marked irritability
  - ✓ emotional lability
  - ✓ anxious
  - ✓ depressed

- ✓ insomnia
- ✓ excessive sleepiness
- ✓ eating problems
- diminished taste
- changes in cognition
- ✓ changes in concentration
- deterioration in vision and hearing related to not paying attention
- slowing of mental processing
- ✓ diminished initiative
- impaired memory
- ✓ increased impulsivity
- repeating same sign or phrase over and over
- obsessive-compulsive rituals
- tics starting with facial grimaces to major muscle groups
- ✓ increased aggression
- increase in self-injurious behaviors
- increase in sensation seeking
  - ✓ novel experiences
  - ✓ intense experiences
  - ✓ increase in risk taking behaviors
  - ✓ violation of well learned rules
- ✓ increase in thoughts of invincibility
  - and telepathic powers
- interest in sexually related behaviors:
  - increase in masturbation
  - increase in staring at others (inappropriate)
  - increase in touching others (inappropriate)
- ✓ hate to be corrected
- ✓ hate to be told what to do
- ✓ hate to be bored
- ✓ hate to be bothered
- ✓

*The above could describe any typical adolescent - with or without CHARGE.*

If the adolescent is experiencing regression in behavior or cognition (mental processes - memory), it is important to rule out an underlying medical deterioration i.e. hypothyroidism, heart disease, regression in hearing and vision.

## REFERENCES.

1. Bernstein, Veronika, 1995, *CHARGE Adolescence Is it Behavior? Is it Communication? Handout.*
2. Blake, K., et al., 1998, *CHARGE Association: An Update and Review for the Primary Pediatrician*, Clinical Pediatrics, pp. 1-16
3. Blake, K., *CHARGE into the year 2000*, presentation handout.
4. Blake, K., 1997, *CHARGE Association Workshop "Growing Up with CHARGE"*.
5. Oley, C. A., M. Baraitser, and D. B. Grant, 1987, *A reappraisal of the CHARGE association.*



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### **Two key things to remember about your adolescent:**

Their success and performance will vary from day to day. Just because they accomplished something yesterday, don't look down upon them because they can't do it today.<sup>1</sup>

There is no miracle strategy for dealing with your adolescent. Just keep trying new things until you find something that works with your teen.

### **MEDICAL ISSUES:**

An important aspect of your child's adolescents is the onset of puberty. Both males and females with CHARGE may require hormone replacement therapy. If a deficiency in sexual hormones is suspected, an appointment with an endocrinologist is suggested.<sup>5</sup> Many families forget the "sex hormone" issues and are either never referred to an endocrinologist or go too late.

In males, androgen replacement therapy (male sex hormones) may cause significant growth in the penis. The psychological boost derived from this treatment is very significant and sufficient reason for considering this therapy. Males also tend to have poor growth of facial hair, which may be alleviated by this therapy<sup>5</sup>.

Females without hormone replacement therapy tend to enter puberty late and have irregular periods. They may also have no breast development without estrogen replacement therapy.<sup>2</sup>

As more CHARGE children enter adolescents there is an increased awareness of problems and how to approach these. Skeletal curvature (scoliosis) is of concern and was brought to our attention when conducting a survey at the 997 Family College Association Conference.<sup>4</sup> If there is concern, ask to see an orthopaedic surgeon. This scoliosis may increase during the growth spurt of the adolescent (13-20 years). The growth spurts in CHARGE adolescents occur later than the documented norms for growth. This puberty pattern reflects the potential for later growth and for a normal adult stature.

As many CHARGE children obtain milestones late (walking, running, communicating), it is not unreasonable to suspect that many of them crave for more learning opportunities later in life. These need to be respected and as some of our adolescent case studies show it is not reasonable to expect the adolescent to be happy doing "basket weaving" or other menial tasks.

### **REFERENCES.**

1. Bernstein, Veronika, 1995, *CHARGE Adolescence Is it Behavior? Is it Communication? Handout*.
2. Blake, K., et al., 1998, *CHARGE Association: An Update and Review for the Primary Pediatrician*, Clinical Pediatrics, pp. 1-16
3. Blake, K., *CHARGE into the year 2000*, presentation handout.
4. Blake, K., 1997, *CHARGE Association Workshop "Growing Up with CHARGE"*.
5. Oley, C. A., M. Baraitser, and D. B. Grant, 1987, *A reappraisal of the CHARGE association*.



## LIFE EXPECTANCY IN CHARGE

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Children with CHARGE have been shown to have a 70% survival rate to five years of age (from actuarial survival analysis).<sup>1,2</sup> The death rate is the highest in the first year of life.

The highest mortality is seen with a combination of CHARGE features of bilateral posterior choral atresia with either congenital heart defects or tracheo-esophageal.<sup>1</sup> If all three of the above are present, they offer the greatest risk of mortality or morbidity to the infant.

Patients with CHARGE have a high postoperative mortality.<sup>3</sup> The reasons for this are postulated as hidden structural abnormalities of the larynx and/or pharynx with associated motor incoordination. This results in difficulty in intubation and problems after extubation. It is therefore important that the surgeons and anesthesiologists be aware of how complex these children are. When contemplating surgery in these children an experienced pediatric anesthesiologist should be involved, even if the surgery is rated as a minor procedure. It is advised that, if possible, a number of procedures be conducted under one anesthetic, thus limiting the number of anesthetics. A recent CHARGE girl at our hospital (IWK Grace Health Center, Halifax, Nova Scotia) undertook seven operations at the same time under one anaesthetic so it can be done!

### References:

1. Blake, K., et al., 1998, *CHARGE Association: An Update and Review for the Primary Pediatrician*, Clinical Pediatrics, pp. 1-16
2. Blake, K., et al., 1990, *Who's in CHARGE a multidisciplinary approach*, Archives of Disease in Childhood, February, vol 65, no. 2, pp. 217-223.
3. Blake, K., et al., 1993, *CHARGE association looking at the future - the voice of a family support group*, 1993;19,395-409.



# **CHARGE SYNDROME MANAGEMENT MANUAL FOR PARENTS**

## **Introduction to the Developmental Sections**

Meg Hefner, M.S.

Every individual with CHARGE is unique. Each has his or her own unique collection of medical problems. Each individual has his or her own personality and learning style. On top of that, each individual is in a different family setting and different medical and educational environments. We would love to be able to provide every family with a complete set of resources tailored to their needs. Obviously we cannot do that. As a consequence, every family will find different parts of this Manual helpful to them. Sometimes we have presented the same or similar information in a couple of different formats. Look it all over and choose the parts that are helpful for you. Use the loose-leaf format to reorganize, add, and delete information that is appropriate for your situation.

GET SOME TABS. There is a lot of information here. It will be much easier for you to find what you are looking for if you break the information up in to sections with tab dividers.

A note on “**DeafBlindness**.” We recognize that not all individuals with CHARGE have both hearing loss and vision loss. Nevertheless, much of the information is labeled “deafblind” or aimed mostly at that population. We did this for two reasons. First, families dealing with individuals who DO have dual sensory loss will have the most difficulty with development and communication and will have the hardest time finding appropriate resources. Second, many of these resources are just plain good resources regardless of the title.

Please don’t skip a section just because it is labeled “DeafBlind” – take a look and see if it has something to offer you as well.



## MEDICAL INFLUENCES ON DEVELOPMENT IN CHARGE

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Medical problems and overall health have a tremendous impact on development. One of the things which makes CHARGE syndrome so complex is that each medical feature can be 0% (absent) to 100% (severe involvement) in any given child. The purpose of this table is to provide a quick visual reference for most of the medical issues the child and parents have to deal with. Features on the severe end of the spectrum should be viewed as priorities. However, mild problems should not be ignored, as they may significantly complicate the overall picture.

On the back of this page is a summary table listing functional problems that have an impact on learning. You may wish to check off those problems which are present (left most column) and make a mark on the scale on the right indicating mild to severe involvement for each feature in your child. Don't get hung up on the percentage. This is only a gross estimate, the purpose of which is to know the relative impact of each when compared to the whole.

Y N	System Affected	Condition	0%-----50%-----100%
			Mild Moderate Severe
	Eyes	Vision loss (coloboma)	
	Ears	Hearing loss	
	CN* 1	Lack of smell	
	Choanae	Nose breathing difficult, stents	
	Inner ear	Balance problems	
	Receptors	High pain tolerance	
	Receptors	Tactile defensiveness	
	CN 7	Facial palsy Rt, Lft, Both	
	CN 9&10	Swallowing problems	
	CN 9&10	Recurrent aspiration	
	CN 9&10	Tube feeding	
	CN 9&10	Tracheostomy care	
	CN 9&10	Copious secretions	
	Heart	Heart problems	
	Kidney	Reflux, kidney probs	
	Growth^	Small for age	
	?Immune	Ear infections	
	?Immune	Sinus infections	
	?Muscles	Hypotonia	

\*CN = cranial nerves which supply the head and neck. All 12 may be affected but the most common are listed.

^Some children have problems with growth hormones but more are small because their bodies use up so much energy from illness or because they do not get enough nutrition due to the swallowing problems.



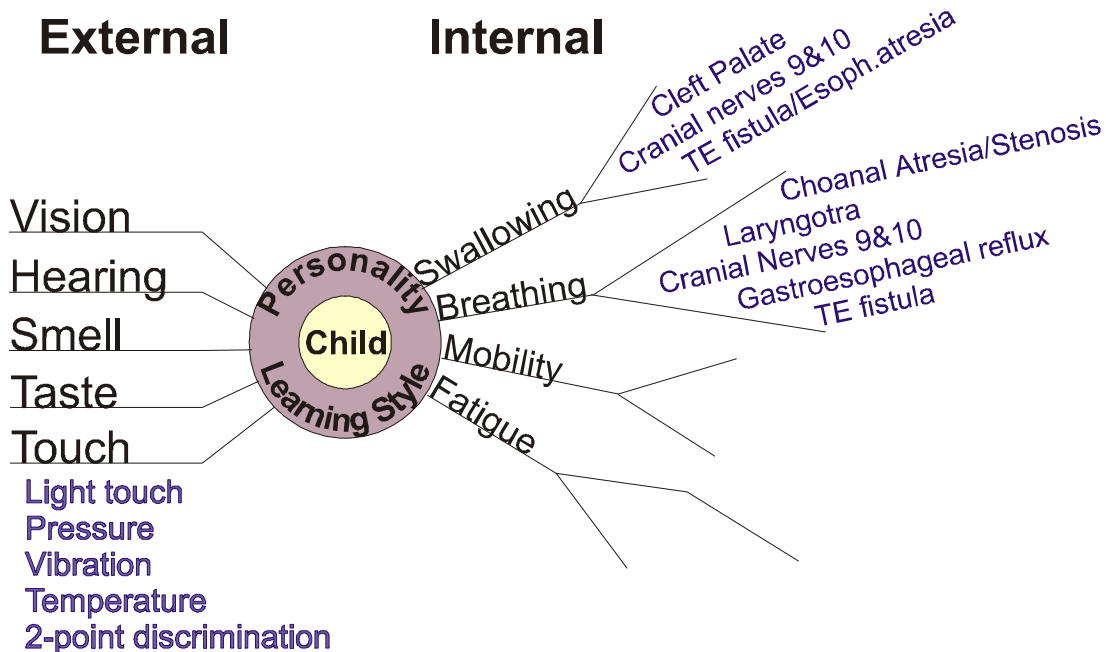
# PHYSICAL INFLUENCES ON DEVELOPMENT IN CHARGE

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By Sandra L.H. Davenport, M.D.

CHARGE is such a complex disorder that many of the physical components have a profound effect on the child's development and understanding of the world. The above diagram breaks these influences down into those that are External and those that are Internal. In one way all are internal since the five senses are all part of the body; however, these senses require outside stimuli in order to perform. Pain is placed between the two because it can occur because of both external and internal events.

The Internal influences that have the most profound effect are problems with swallowing, problems with breathing, delayed mobility and problems with fatigued and illness. Each of these problems has multiple causes, which are listed and further explained in the medical section of the manual.

The "External" senses are the source of contact with the outside world. Decreased vision impairs understanding of the environment. The vision loss in CHARGE is somewhat unique because most children have colobomas, which cause upper visual field losses. This means that they may have blind spots above the direct line of gaze. More importantly, they may involve the macula or the optic nerve, which will cause blurred vision.

Decreased hearing results from malformations of the middle and inner ears and/or middle ear infections. Hearing loss is common in CHARGE and causes major problems with understanding and imitating speech as well as not being able to detect environmental sounds. Most children that have hearing loss due to malformations of the inner ear also have problems with balance because the vestibular system is malformed.

Smell may be decreased or absent in CHARGE. This results in lesser appreciation of food and in social mishaps because they don't know when they stink. Smell is also important because it is the third distance sense. They can often tell who is coming by perfume or body odor. Rooms of the house in different places outdoors may also have distinctive smells. These may help them figure out where they are.

Taste is almost always normal in children with CHARGE. It can be important for exploration when the other three senses are not working. Older children who have profound vision and hearing loss and have not had adequate communication training may spend a lot of time mouthing objects. Putting objects in the mouth gives more information than simply handling them.

Touch is actually much more complex than people realize. The sensors under the skin can distinguish between sharp and dull, soft touch and pressure, vibration, as well as differences in temperature. Two-point discrimination means being able to tell the difference between one pin and two pins. The pins can be very close together on fingertips and still recognized as two objects. Most of these aspects of touch seem to be normal. However, a lot of children prefer deep pressure to light touch.

Pain may be different in many children with CHARGE compared to their peers. Parents report that their children have very high pain thresholds. One consequence of this may be that they cannot understand why other people react to painful pinches or kicks etc. Because they don't feel the pain themselves they may not be able to empathize with other people's pain.

## INFLUENCE OF SENSORY LOSS ON DEVELOPMENT: The Communication Bubble

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### Are all five major senses working?

The chart below is not based on any reliable data. Estimates are quoted in the literature that the percentage of information that we take in via our eyes is anywhere from 60 to 90%. Of course, if you are listening to an audio tape, vision doesn't matter at all. If you are watching a film strip, hearing is of no consequence. In addition, some people are visual learners and some people are auditory learners. It is well-known that two people with identical audiograms may have strikingly different abilities to understand speech and other sounds. Perhaps the one who understands speech better is actually an auditory learner. But even that person misses information. If he/she uses speech-reading extensively, then a vision impairment on top of a hearing impairment will cut down on understanding as well.

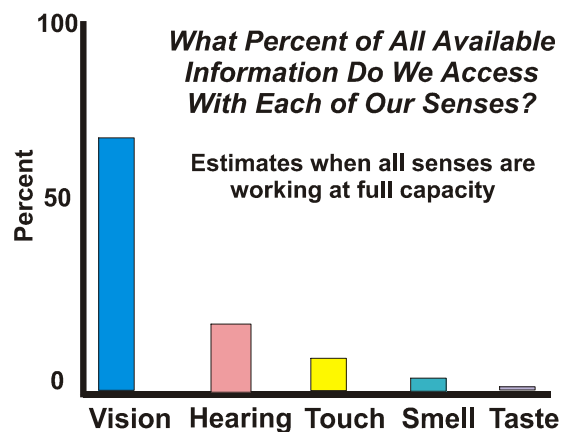
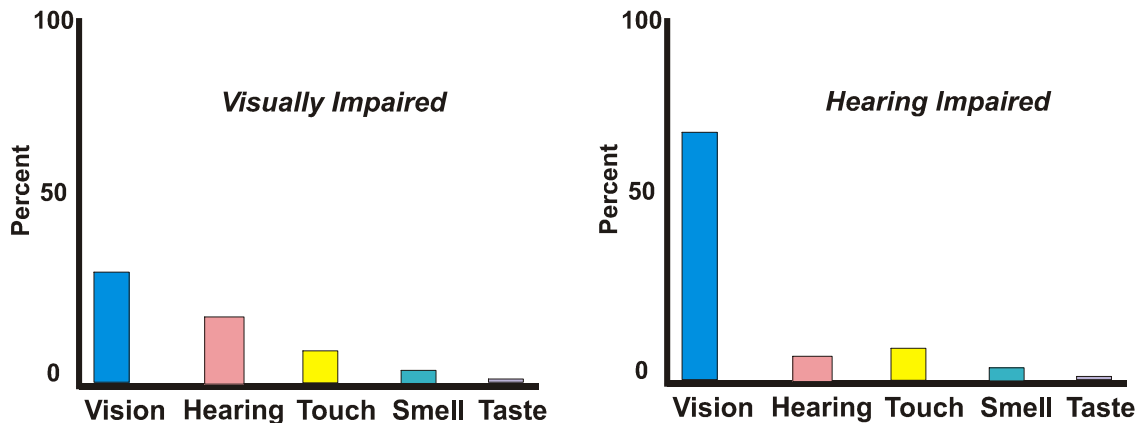


Figure 1: Full access to information from all senses.

However, consider what happens if a child is visually impaired AND hearing impaired. If a child has moderate visual impairment, the Vision bar might be half as high. If the child is moderately hard of hearing, the Hearing bar would be shortened by half. Relatively speaking, then, the other senses become more important.



Figures 2 & 3: Compare the differences when half of either vision or hearing is gone.

Now consider how less information is available when half of BOTH the vision and the hearing are missing. Notice how much more important the senses of touch and smell become.

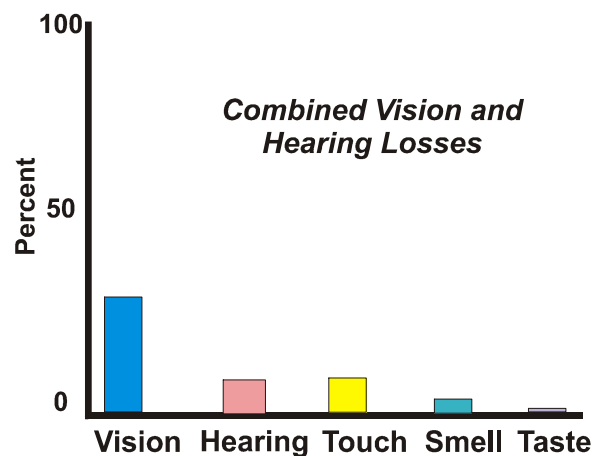


Figure 4: Combined vision/hearing loss with half of each gone.

## **What is Deaf-Blind?**

**Definition:** Any combination of hearing and vision loss that interferes with access to communication and the environment and requires interventions beyond those necessary for hearing or vision loss alone. For educational purposes in most states, the child needs to meet the criteria for deaf/hard-of-hearing as well as for blind/visually impaired (check with your own DeafBlind Project director)

Deaf-Blind rarely means totally deaf and totally blind. No other succinct term has been found that fits all of the conditions listed in the table below. Dual sensory loss, Hard of Hearing/Visually Impaired, etc. are cumbersome. Most parents and educators have finally settled on DeafBlind even though that term is a somewhat inaccurate and, often, a scary term.

The importance of having a DeafBlind label, however, cannot be underestimated. The educational needs are truly different when both senses are affected. It is not enough to have the consultants come in and give their input without considering the effect of the other sensory loss. The reason is that the techniques used to compensate for vision loss often involve hearing and those used for hearing loss frequently involve vision. Note in the table below that the term DeafBlind is used even if the child is found to be just visually impaired and hearing impaired. The combination makes a HUGE difference in the way information should be presented.

## **What combination of vision & hearing is present?**

When one or more senses are impaired, additional educational consultants are needed, even (or perhaps especially) during the Early Childhood phase of development. Research out of Denver shows that, if a communication system is established BEFORE six months of age, a deaf child can develop completely normal language. Even if they learn to read sign language, they do not fall behind their hearing peers in expressive language and reading. The implications are staggering. Even though medical issues in CHARGE occupy the thoughts and minds of parents and caretakers during the first 2 years of life, it would appear to be very important to start some form of communication as early as possible.

The table below introduces the kinds of special educational consultants needed to address the vision and hearing issues. However, physical therapists, occupational therapists and a variety of other consultants may also be needed (just like in the hospital and clinic). Note that the kinds of modifications given depend on the nature of sensory input.

<b>Sensory Status</b>	<b>Consultant Needed</b>	<b>Modifications</b>	<b>Comments</b>
Hearing Sighted	None		
Hearing Visually impaired	B/VI teacher +/- O& M	Large print, contrast, placement of student	
Hearing Blind	B/VI teacher +O& M	Braille, O&M, audio tapes & instruction	
Hard of Hearing Sighted	D/HH teacher +/-SPL	Hearing aids, +/- FM or other amplification, noise reduction, classroom placement	
Deaf Sighted	D/HH teacher +/- Interpreter	Deaf classroom or program, interpreters	
Hard of Hearing Visually Impaired	B/VI, O&M, D/HH, SPL, DBP consultant	Amplification, large print, be within the "Communication Bubble"	Deaf-Blind
Hard of Hearing Blind	B/VI, O&M, D/HH, SPL, DBP consultant	Amplification, Braille, O&M, +/- sign language	Deaf-Blind
Deaf Visually Impaired	B/VI, O&M, D/HH, DBP consultant	Modified sign language, O&M, large print, be within the "Communication Bubble"	Deaf-Blind
Deaf Blind	B/VI, O&M, D/HH, DBP consultant	Modified sign language (possibly tactile), Braille, O&M	Deaf-Blind,

Table modified from Rob Last

B/VI = Blind/Visually Impaired

D/HH = Deaf/Hard of Hearing

SPL = Speech/Language

O&M = Orientation & Mobility (learning how to know where you are and move safely with low or no vision)

DBP = DeafBlind Project

## The Need to Establish A Communication Bubble

Establishing what Susan Smith, the parent of two children with CHARGE, calls the “Communication Bubble” is essential if you want to make sure the child knows you are there and are trying to communicate. The concept of a “bubble” is a good one because you can imagine the space within which you need to be. As Eric Kloos says, you need to be “on the child’s radar.” Get outside the bubble and you might as well not exist. Too many times, we think the child is tuned out, not paying attention, or is too “retarded” to answer when, in fact, he/she may not even know that you are trying to say something.

Each parent should know how far a child can see what kinds and colors of objects, with or without movement, in different lighting conditions and against different shaded backgrounds. The eye doctor will usually not be able to tell you this except by guessing though some do this kind of testing. A vision teacher, however, can do a Functional Vision Evaluation, to help establish these visual abilities. Figuring out what the child hears clearly, at what distance and on which side is also important. Neither of these is easy to determine so parents and team members will likely have to engage in a continuing evaluation process.

## What Types Of Communication Systems Are Available?

Emerging language	Touch cues	
	Object cues	
	Gestures	
	Pictures	
Formal language	Oral Language	Auditory/Verbal
	Oral Language supported manually	Cued Speech
	Sign Language	Signed English
		ASL, Auslan, etc. (the native sign language of a given country)
	Total Communication	Combination of oral and sign languages

Table modified from Rob Last

## How Do You Choose the Right Method?

There is no one right answer to this question. Too often, however, the confusion results from passionate arguments for one method of communication, like spoken English or American Sign Language, without fully understanding the effects of dual sensory loss. **The most important thing is to get language into that developing brain in a form that can be consistent and effective.** What that form takes will likely be determined a team meeting of the parents and professionals. We learn any language by receiving it and then repeating it. We learn it fastest and best by total immersion. Learning it for an hour in the classroom twice a week or in bits and snatches with a word or sign here or there is simply not enough to become fluent. This means that whatever method is chosen needs to be woven into everything the child does, from getting up in the morning to going to bed at night. Keep in mind that perfectly good language by whatever method is useless to the child unless it is delivered within the communication bubble. The other very important point is that a lot of communication precedes formal language. Every mother can “read” her child’s communication regardless of whether that child uses any formal

language. Sometimes it is guessing, but the child will let the mother know when she's got it right. Therefore, it is important to lead up to formal communication step by step. We use sight cues and verbal cues like outstretched arms with some encouraging words to let a hearing/sighted child know what we want to do next. For a child without good vision or hearing, concrete object cues and touch cues are used instead. DeafBlind project staff can help the regular and special education staff learn how to use these methods. As parents and educators we may get hung up on wanting our children to speak our own language using the method we use. That is as natural as immigrants wanting their children to continue speaking their language and remembering their culture. However, the issue is not the form but the substance. The child needs to learn that an object, touch, picture, word or sign is a symbol that stands for a thought. Many of them strung together convey complex ideas or stories. They include naming of objects, actions, remembering the past, anticipating the future and, later, getting into abstract thoughts and discussions.

So, above all, just get to it! COMMUNICATE, COMMUNICATE, COMMUNICATE.



# BEHAVIOR AS COMMUNICATION

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All children behave. Much of this behavior is a matter of concern to parents, teachers, and others. Bookstores are full of information on how to cope with the misbehavior of children. No one should be surprised that children with CHARGE also have behavior that is a matter of concern. While the misbehavior of any child can be challenging, the misbehavior of children with CHARGE can be overwhelming for several reasons. First, there are frequently communication problems that make it difficult to discuss behavior problems and expectations with the child. Second, medical complications may limit the kinds of responses to misbehavior that parents and others are willing to utilize. Third, some of the behavior demonstrated is quite unusual when compared with the behaviors of the typical child.

When considering behavior management, one needs to be very clear about its goals. Eliminating misbehavior is not achievable, and by itself is most likely not desirable. To understand this, two important principles need to be described. First: all behavior has a purpose. Behavior is not random. Sometimes the purpose may be very simplistic, like scratching an itch on my head, but it is goal oriented. This means that the behavior demonstrated by our children with CHARGE is not random. They engage in it for a reason. The second principle is equally important: behavior serves a communicative function. This means that we use behavior to communicate something to other people. If we understand the communication, and if we understand the purpose of the behavior, then any behavior can make sense.

Now, to return to the issue of eliminating misbehavior. If we focus our efforts on stopping a behavior, we have taken away a means to a goal, and a means to communicate. That may suggest to the child that we are not interested in their goals or in communication with them. This may lead to withdrawal on the part of the child as they give up trying to engage us in communication, and it may lead to different, perhaps even more difficult, behaviors to try to achieve what the child experiences as important goals.

Thus the primary objective of behavior management is to understand the purpose and communication underlying misbehavior, and to use that to help the child find more appropriate communication methods to achieve their goals, or to help them find more appropriate goals. A temper tantrum may be a means to communicate the desire to receive attention, but children also have to learn that they need not be the center of attention all of the time.

When considering the goals of behavior management, six major principles need to be considered. **First**, as already stated, problem behavior serves a purpose for the person displaying it, part of which is communicative. Communication in general is fraught with difficulties. That is because all communication is subject to encoding of messages, and then decoding and interpretation. If I choose to communicate my frustration about a situation, I must first take my message of frustration and put it into some kind of communicative format, usually language. This is encoding. The listener must then hear my message and interpret it. This is decoding. Of course the listener interprets based on their frame of reference at the moment.

People with outstanding communication skills are articulate in encoding their messages, and accurate in decoding. However, when dealing with a person who lacks communication skills, and who may in fact be nonverbal, communication becomes extremely difficult. That is why communication with children can be a problem, and certainly why communication with children who have CHARGE is difficult.

**Second**, functional assessment is used to identify the purpose of problem behavior. If you know the purpose, the behavior is understandable. Our first task in working with a child with behavior problems is to understand the message behind their communication. We must decode their encoded message. To do this we try to determine the function or purpose of the behavior. Much has and is being written about methods for functional assessment. A great deal of training is needed to utilize some of them. IDEA requires functional assessment in the schools, and school psychologists should be able to conduct such an assessment.

**Third**, the goal of intervention is education, not simply behavior reduction. Once you believe you understand the purpose of the behavior, how do you intervene? The difficulty here is to modify the misbehavior without shutting down the child's attempts to communicate. Here are several general considerations. First, do not become annoyed, threatened, hurt, or give up. Second, help the child find a more appropriate method of communicating the message to you. A third consideration is to avoid crisis management. This involves ignoring the behavior, protecting the individual and others, restraining the individual if necessary, removing anyone who is in danger, and engaging in behaviors that generally reduce the misbehavior. The problem with crisis management, which of course must be used at times, is that it does not teach the child new ways of communicating and reaching their goals. A fourth consideration is to be patient. It takes time to modify behavior in a way that teaches the child the benefits of clearer communication.

**Fourth**, problem behavior may serve many purposes and therefore requires many interventions. The same behavior, for example having a temper tantrum, may be used when the child wants something, and when they want to avoid something, and when they simply want attention. Intervening for situations where they child wants attention, may not eliminate its use when the child wants to avoid going to see the doctor.

**Fifth**, intervention involves changing social systems, not simply individuals. In considering intervention strategies, there are approaches that are directly applied to the child, those that are indirectly applied by changing the behavior of other people in the child's environment (such as changing the behavior of a teacher or a parent), and those that modify the environment itself. A question that might be asked is whether the problem is that the child is communicating inappropriately, or whether the social system is not listening to the child. Very often the approach is to make the child change to meet the demands of the system, as opposed to looking at how systems might better meet the needs of children.

Finally, lifestyle change is the ultimate goal of intervention. Dealing and coping with our children's problem behaviors is enormously stressful, due to worry and a sense of helplessness. Our goal should be children and families who are able to meet their needs and find satisfaction with their lives. Here are some ideas to help you cope:

First, be glad your child is trying to communicate. You may not like the method, but at least there can be some connection.

Second, choose your battles wisely. Not every misbehavior has to be deal with. Not every troubling behavior needs to be labeled as misbehavior. Go after those that cause your child the most difficulty.

Third, don't feel like you have to go it alone. In fact, everyone in your child's environment needs to be on the same page in terms of responding to the problem. So mobilize as much help as you can from those other people.

Finally, take care of yourself. You are not simply the parent of a child with CHARGE. You are a person in your own right, with your own needs and goals. If you never have a chance to work on your own goals, you will become less and less useful to your child.



# MINNESOTA DEVELOPMENTAL TIMELINE

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Minnesota DeafBlind Project

## PURPOSE

Most developmental tables and charts show developmental milestones without taking into account any sensory losses, hospitalizations, illnesses, educational interventions or major family events. The Minnesota Developmental Timeline shows the relationship of all these factors over time. The purpose is to understand more clearly the unique circumstances which affect a child's learning. This chart should be kept by the family with copies becoming part of a child's permanent educational and/or medical records. It should also be updated periodically on the parent's original with the chart copies being replaced as necessary. Some parents may decide they do not want to keep the original.

The chart is both succinct and approximate, i.e. exact dates and lengths of hospitalization, great detail on milestones are not necessary. The focus should be on the overall picture and the relationship of events. Details can be written on the back of the page or on separate sheets.

## WHO SHOULD PROVIDE THE DATA:

Parents or other caretakers usually can provide the most of the information. Medical and educational professionals can help make the descriptions more accurate and precise and help fill in the informational gaps.

## HOW TO CONSTRUCT THE CHART:

1. While a form is often provided and you can certainly use a handwritten form, you may also wish to develop your own chart using a drawing program such as Corel Draw or by using as a spreadsheet like Excel. The drawing programs produce pretty results but are more time-consuming to construct. The spreadsheet is more readily available and will make it easy to update the information yourself while keeping the information easy to read. Two drawbacks are that the age/date columns become uneven and the hospitalization lines will have to be drawn in by hand.
2. Enter child's name, birthdate and the date the chart was filled in at the top of each page used. Decide whether you want the hatch marks across the top of the chart to represent days, weeks, months or years. Sometimes having a whole year on one page will not give enough space to fill in all the important events. A young child might need one page to represent 6 months while an older child might need one page to depict the first 3 years of life. Write in the age at the larger marks and also put the date plus age at least twice on each page, e.g. 6 mo. (8/96). Having both makes it easier to fill in information. Sometimes parents remember an event occurred just after New Year's but would have to figure out how old the child was at that time.

3. Mark in all hospitalizations (see Figure 1). These are bars which span the entire page and are shaded or colored in. The reason for doing this is that children typically show delay or even regression in development during and sometimes after a hospitalization depending on the severity of the illness and the degree of perceived physical or emotional trauma experienced. Hospitalizations therefore have a major impact on all areas of development.
4. Write in precise terms the reason for hospitalization or major illnesses (see Figure 2). For instance, PE tubes, Nissen, pneumonia, EUA (exam under anesthesia), etc. Explain procedures or abbreviations on the reverse side. Under Other Illness write in medical problems like ear infection, swallowing problem, etc. A line can be drawn from the description across the page to the date or age at which the problem resolved. For instance, swallowing problems might last from birth to 3 ½ yr. so the words are written at the birth line and a line with an arrow is drawn across the entire first page and subsequent page(s) ending at 3 ½ as shown in Figure 2.
5. Fill in developmental milestones (see Figure 3). Motor and Mobility includes both gross motor milestones and O&M (orientation and mobility) milestones if applicable. Under Vision, Hearing, Taste and Smell note what sensory stimuli the child responded to at different ages and give numbers for visual acuity, degree of hearing loss, etc. when tests were given. Also note when glasses, hearing aids, FM system at home or school, or any other device was introduced. Use a line across the page to note how long the child used the device if it was discontinued. Use an asterisk or number to indicate an explanation on the reverse side as to how long the child used the device. For instance, a hearing aid might have been fitted at age 3 yr. but was only used during preschool Mon, Wed, Fri, for 2 ½ hr during the school year. A dotted or dashed line might indicate this kind of non-continuous use.
6. Under Educational Services note when teachers and therapists began and stopped working with a child. (See Figure 4) Use abbreviations like ECSE (early childhood special education), PT (physical therapy), etc. Note whether this is at home or school and duration of service, i.e. 2x/wk for 30 min. This kind of detail can be put in an explanatory note on the reverse side.
7. Major Family Events are entered next (see Figure 5). These should include anything that can affect development like the birth of another sibling, death of a grandmother, a move, foster placement, frequent changes in home health care aides, etc.
8. Color code major areas that need emphasis (see Figure 6). For instance, hospitalizations might be colored with yellow highlighter. Anything that affects hearing can be highlighted in pink such as hearing milestones, ear infections, PE tubes, deaf/hard of hearing teacher, deaf uncle moved into town. Please note that color coding does not show up on black and white copies so use this judiciously.

#### **POSSIBLE USES FOR THIS CHART:**

1. Summary for parents/guardians.
2. Summary of past history for child's education file. The chart can be amended periodically and available for all new staff to review.
3. Summary for medical file.
4. Summary for case managers in settings outside education or clinics.

Name: \_\_\_\_\_ B.D. \_\_\_\_\_ Date: \_\_\_\_\_

Age (yrs) (mo.)												
Ill or in Hospital												
Other Medical												
Motor & Mobility												
Fine Motor												
Tactile												
Vision												
Smell												
Taste												
Hearing												
Communi- cation												
Education/ Services												
Family Events												

Name \_\_\_\_\_

B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	Birth	Ill or in Hospital	Other Medical	Motor & Mobility	Fine Motor	Tactile	Vision	Smell	Taste	Hearing	Communi- cation	Education/ Services	Family Events
3-4-94	Birth	G-tube/Nissen fundoplication VSD, PDA, hypo- plastic Rt. vent. artery, mild hydronephrosis started w/ unilateral facial palsy	Swallowing problems Oral stim. program started w/ unit at home				Colobomas of both optic nerves Lt > Rt			Turn to voice. Rt hearing aid used thru hospital		OT to home	No home nursing help
5/94		Pale, limp, xanth to GERLUX	OT at home			Markedly defensive to all touch. Started on- going pro- gram of brushing	Tracked w/ both eyes when well						
7/94		Heart surgery Hemi-Fontan	Recurrent ear infections								Parents started learning sign lang		
9/94													
11/94		Respiratory infection ICU on vent.										SKI-HI lang teacher 1x/wk PT	
1/95		Respiratory infection	On steroids stopped oral feed, "nasty" disposition					No definite reaction to any smell	Wrinkles face to piquante sauce & sour pickles				
3/95	7 yr	Bowel obstruction	Started oral stim again	Rollled, did not stay on front							1st word = "more in sign"		
5/95		Scooped airway Decannu- lations x2								Add Lt hear- ing aid but hard to keep on floppy ears so use still inconsistent			
7/95							Reached for something on floor but could not round for it						
9/95					Pick up lint Stack 2 blks							ECSE x 6 mo. PT, OT, DHH continued at home during school year until preschool	
11/95		PE Tubes #1 Trach closure											
1/96		Aspir. Barium Resp. arrest					Glasses for farsighted- ness helped w/ accuracy						
3/96	(2 yr)												



Name \_\_\_\_\_

B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	3/96 (2 yr)	5/96	7/96	9/96	11/96	1/97	3/97 (4 yr)	5/97	7/97	9/97	11/97	1/98	3/98 (5 yr)
Ill or in Hospital		Heart cath		Heart surgery: Fontan				Otoplasty Eye muscle surgery					
Other Medical	Swallowing problems	Recurrent ear infections								oral stim program at school			
Motor & Mobility	5-pt crawl Few steps holding on												Walked independently
Fine Motor				Held pencil fairly well w/ little drawing						Made circles w/ open bottoms			
Tactile										Refused to touch anything, e.g. fingerprints			
Vision													
Smell	No definite reaction to any smell									Initiated smelling flowers but never discriminated among smells			
Taste													
Hearing									L hearing aid	FM unit at school			
Communication	2-word sentences Increasing vocabulary & understanding	"Funny Daddy" on Father's Day											
Education/ Services								Summer school July		Started preschool w/ interpreter			
Family Events													

Name \_\_\_\_\_ B.D. 3-8-94 Date: 10/5/98

Age (yrs) (mo.)	3/98 (5 yr)	5/98	7/98	9/98	11/98	1/99	3/99 (6 yr)	5/99	7/99	9/99	11/99	1/00	3/00 (7 yr)
Ill or in Hospital	PE Tubes #2												
Other Medical				G-tube feeds but not aspiric ating									
Motor & Mobility	Falling - bruise on head			Can stand up in mid room Less falling									
Fine Motor			Makes triangle w/ round bottom	Letters-not interested. Draws face not body Closing circles									
Tactile				Now touches anything but the head to initiate touch									
Vision			Rt: 20/50 Lt: 20/60 Upper fields partly missing	Can ID each kind of flower by color									
Smell	No definite reaction to any smell												
Taste													
Hearing		FM unit purchased for home use	Began responding more to voice										
Communi- cation			Using both sign and voice.	Asking others to fingerspell everything. Uses 7-8 word sentences									
Education/ Services		Summer school July PT all summer for balance											
Family Events													

# INTRODUCTION TO TEAMS

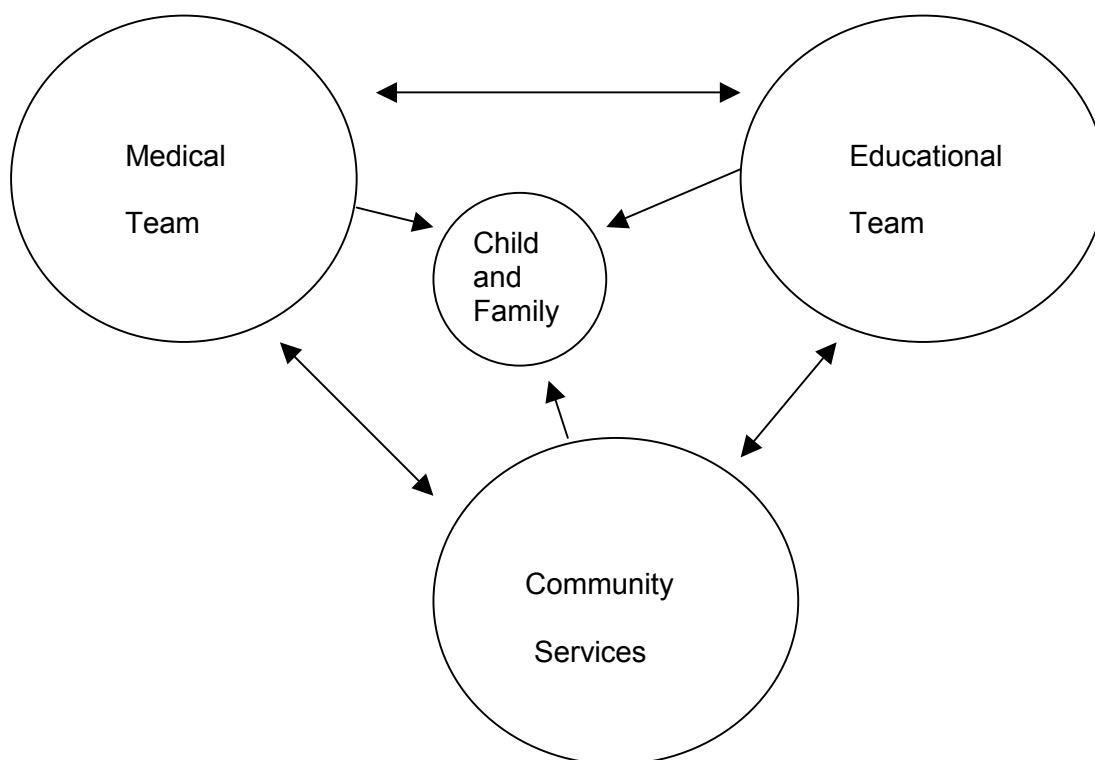
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All children with CHARGE are unique and often absolutely delightful. Underneath all the possible medical and educational challenges is a terrific human being who wants to be a part of the family and community just like every other child. Regardless of the number of medical and sensory issues, every single one of these children has the potential to learn, to communicate and to lead a satisfying life.

This is not to say that no challenges exist. They do, to a greater or lesser extent. It would be wise to keep in mind, however, that the way the children deal with their own challenges may reflect the attitudes of the people around them. As in all relationships, the more positive we are, the more positive they are.

Because each of the many possible medical findings in CHARGE can vary from being absent to profoundly affected, no two children have exactly the same findings. As a result, a variety of specialists enter the lives of families to deal with the different individual issues. The parents themselves are the only ones that stay constant in the life of the child as professionals come and go. They may need to interact with a variety of specialists, many of who have never heard of CHARGE. By default, the parents become the tireless teachers, team leaders and case managers.

Families will interact with three major systems: medical, educational and community services as shown in Figure 1:



### **Coordination of services and communication between systems (teams)**

When many different specialists are needed in any of the systems, it is usually necessary to have a **coordinator** within each system. If one coordinator is not automatically assigned, the family would be wise to ask for one. In reality, the parents (most often the mother) are the ultimate coordinators.

Somehow information needs to flow within and between the systems, each of which has its own way of doing things. The ideal would be for the coordinators from each system to talk to each other, but this may be difficult. It is often difficult to get member of the *same* system to communicate with one another.

Typically, *medical* people deal with other medical people in writing, both because of the legal requirements and because they may not be able to talk to each other because of time or distance. They will write a summary letter when requested to do so. You can request copies of these letters as well as other medical records. You can request copies for yourself and/or copies to be sent to others.

Typically, *educational* people talk to the classroom teacher (not always to the parent) and put their findings in writing only when requested. Again, you can request written summaries be done and have copies made for you and others. Certain summaries that deal directly with the assessment of medical issues (e.g. Functional Vision Evaluations and Audiograms) should also be sent to the medical coordinator.

Each of the *community agencies* has a different way of dealing with issues, so some keep written records and many do not. This means it may be hard for the family to keep track of what is going on. You should ask for written reports at every opportunity. These records will come in handy whenever the child has to move from one system to the other but especially going from one town to another.

## TEAMS AND RESOURCES

Jeanne McMullen, mom to Caitlyn (age 6) CHARGE  
Pittsburgh, PA

Effective management of the child with CHARGE Syndrome has always been a work in progress for us. The key is to develop a good team of professionals around you. Consider yourself the team captain and your service providers the players. Continually evaluate which team members are playing their positions optimally and which ones are not, for if one position is slacking, it affects the child's ability to function as a whole. Often our mistake has been to stay too long with a doctor or program, yet if someone is not pulling his weight now, he never will. Remember, "Keep the best; replace the rest."

### The Medical Team

When attending appointments whether at school or the **doctors**, go prepared to do business. Dress professionally, and come prepared with any documentation to back up your concerns. This may include physician reports (remind each physician to "cc" every report to you and request a copy of all evaluation and lab results for coordination of care), diary of ongoing symptoms as they occur, or communication book from school. Write down your concerns ahead of time (use the Medical pad that comes with the Manual). Remember to prioritize your concerns. Seeing a physician with more than three concerns in one appointment may confuse the doctor and allow you both to be sidetracked. Marathon IEP meetings leave you not thinking clearly, and end unresolved. Remember you can always schedule follow up appointments.

Upon return home, jot down a detailed minutes of the appointment including what concerns discussed were left unresolved. Using a spiral bound notebook for each (medical, educational, and community), will give you something admissible in court should the need arise. Spiral bound, not loose leaf, cannot be added to at a later date and carry more weight with a judge.

Personality conflicts and differences of opinion will be inevitable, but if you keep your cool, your opinion will be respected and heard. Many professionals have experience dealing with irate parents and will often just respond by ignoring them. Keeping the lines of communication professional and amicable will alleviate stress and in the long run work in your favor. Being pleasant doesn't have to mean being a pushover. Be firm.

My medical team is constantly changing. A good doctor must be a good listener and must address concerns adequately. I have learned that the "best in the city" is often not the best for us. Doctors with overblown reputations are often overscheduled and have students or new technicians doing much of the examining. They allow little time for parent concerns, and mistakes can be made in their rush. Include **nurses** in your team. They are easier to reach, are often better listeners, and can handle many problems themselves.

Learn the system at the hospital. Most hospitals have a **patient advocate** who you should get to know before a problem arises. Ours has been instrumental in cutting through "red tape" when scheduling dual procedures, searching for lost records and handling complaints. During long recoveries following surgeries, having her assistance meant we could grab a bite to eat or

use the facilities--something necessary to get us through the day. Take advantage of their **home health care coordinator** when faced with problems regarding your home care providers.

## The Community Team

Advocating for services in your community takes time and will never end. Start with your **blue pages** in the phone book. Many large cities maintain help lines that parents can call like “The Special Kids Network” in Pennsylvania. **Parent-to-Parent** is a good way to start too, for most parents know far more about the system than any single professional can. Most states have programs under “**Family Support Services**” which provide services and funding for therapeutic programs not covered by insurance such as therapeutic horseback riding or assertive technology. Eligibility requirements and services vary from state to state. In Pa., like many other states across the U.S., the transition from institutionalized care to family centered care has become a priority. Here, eligible children are granted intensive therapeutic services to teach them more independent and appropriate behavior at home and in the community. These services, which can alleviate some of the burden and stress at home, are often only available to those families that hear about them from other families and then advocate effectively for them.

Your local schools serving deaf and blind children often have catalogues for recommended toys and other resourceful information that can help even when your child is still too young to attend. The **library** often can do funding searches for you when looking for grants. Organizations such as **Easter Seals** often have loaner assistive technology you can try before making a major purchase.

Only by chance have we learned about programs in our state which not only allow my child **Medical Assistance** but also pay her monthly premiums to keep her on private insurance as well. Through trial and error, we learned how to effectively advocate for reimbursement for traditionally noncovered services such as transportation expenses (MA transportation program), nutritional supplements (**WIC**), and even adapting her tricycle (investigating the MA system and appealing). We have been fortunate to have a **toy-lending library** in our city whose primary goal is to provide an accessible and inclusive play-space for children of ALL abilities to play together. They have been instrumental in helping families like me secure state funding to hold workshops, camps, and build more programs that serve our needs.

## The Educational Team

Every state in the U.S. must maintain a help line for concerns regarding educational rights like the “**Parent Education Network**” in our state. They can answer questions over the phone as well as offer workshops on educational advocacy. Ironically, good advocacy skills may be necessary to find a good educational advocate to help you, as the system is overworked and underfunded. Most **educational advocates** will often only support you in your role as advocate, as opposed to advocating for you. We have been fortunate to find one at our county. His role has been carefully defined as to not “get parents what they want” but to insure legal compliancy within the school district, which is basically all you really want or need in an advocate. As you build your support team in the community, friends, social workers, etc., remember many of them can be utilized at IEP meetings to avoid being outnumbered. Having support at these meetings can alleviate stress as well as having witnesses for possible due process.

They say it “Takes a Village.....”. It cannot be more true when you have a special needs child. When my daughter was an infant, her medical needs were far too intense, and we as parents far too naive that we were unable to handle anything more than our grief. *Yet, a few well-placed phone calls in the beginning would have saved us thousands of dollars, would have made a difference in our daughter’s access to care and services, and resulted in a better quality of life then and now.* Now, I have become a parent many turn to for information. As I help them, they help me, for there’s always something they’ve learned that I haven’t (like using adult socks to teach my daughter initial dressing skills). Learn the system. Develop relationships. And NETWORK, NETWORK, NETWORK!





## EDUCATIONAL TEAMS

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The educational team is a group of people who join together to collaboratively design and carry out plans for a student in special education. The team approach allows many people with different perspectives to come together and work to enhance the education of the student. Each team member may have specific knowledge about certain areas of a child's education (for example, a physical therapist has knowledge of a child's gross motor abilities). However, when all members come together and share knowledge, everyone's expertise on the child is enhanced. The team may address discipline-specific issues or general issues. For example, an issue of communication and behavior may lead to ideas from the speech and language pathologist. However, all team members working with the student may contribute to this discussion by talking about how the child communicates and behaves with each of them. In that way, a bigger picture is seen, and solutions come more easily. When members share their knowledge, it can lead to sharing roles. Each member becomes aware of all aspects of the student's learning. Here is a list of common team members and their typical roles with students:

Team Member	Knowledge Area
Parent	General
General Education Teacher	General Curriculum
Special Education Teacher	Social/Self-Help/Cognitive/Curricular Adaptations
Instructional Aide	Carry out instructional plans with student
Speech and Language Pathologist	Communication/Language/Speech
Occupational Therapist	Fine Motor, Oral-Motor, Sensory, Functional Adaptations
Physical Therapist	Gross Motor, Sensory, Posture, Endurance, Mobility
Audiologist/Hearing Specialist	Hearing Aids, Hearing Ability, Auditory Training
Vision Teacher	Vision Ability/Visual Adaptations
Orientation and Mobility Instructor	Cane Skills, Routes, Mobility
School Psychologist	Assessment, Behavior, Educational Recommendations
School Social Worker	Counseling, Social Skills, Community Connections and Resources
School Counselor	Counseling/Therapy/Social Skills
Building Principal	Resources/Schedules
Special Education Administrator	Resources/Special Education Rules

Teams **evolve**. Not everyone comes to the table with an idea about what will happen. Rainforth & York-Barr (1997) explain:

The primary function of collaborative educational teams is to support students with disabilities during their public school years to acquire the knowledge, skills, and dispositions to lead meaningful, contributive lives. This work is given specific focus through each student's IEP. . . Not all team members come to a team with clarity of purpose regarding educational process and outcomes or their respective roles and contributions; certainly, not all team members come with *shared* clarity of purpose. As members work together, clarity emerges (p. 250).

**Team Leadership.** A team will need a leader. This is often the case manager, or special education teacher, although anyone else on the team, including the parent, could take on this role. This person will need to organize agendas, check schedules, send out notices and minutes, and facilitate the meetings. The team may choose another member to record the minutes. The team facilitator's role is one of cheerleader. This person must support and encourage other members of the team.

**The parent's role.** Parents should be welcomed as equals in any team that forms around their child. A parent's role on the team is to provide information about the child, make suggestions about strategies that may work with the child, and advocate for the needs of their children, among other things. Although advocacy means speaking up for what you believe is best for your child, it is best to do this in an assertive, but not aggressive way. The team process may seem as if is stalling at times, but it does take time for everyone to come around to one person's way of thinking. If you can, find a way to advocate without being adversarial. And always remember that you are the expert when it comes to your child. You know your child better than anyone!

## **TWO SCULPTORS**

*I dreamed I stood in a studio  
And watched two sculptors there,  
The clay they used was a young child's mind  
And they fashioned it with care.  
One was a teacher; the tools she used  
Were books, music and art.  
One, a parent who worked with a guiding hand,  
And a gentle, loving heart.  
Day after day the teacher toiled  
With touch that was deft and sure,*

*While the parent labored by her side  
And polished and smoothed it o'er.  
And when at last their task was done,  
They were proud of what they had wrought;  
For the things they had molded into the child  
Could neither be sold nor bought.  
And each agreed he would have failed  
If he had worked alone,  
The parent and the school,  
The teacher and the home.*

*Author Unknown*

For more information about educational teams, the following resource, quoted in this document, is very helpful:

Rainforth, B. & York-Barr, J. (1997). Collaborative Teams for Students with Severe Disabilities: Integrating Therapy and Educational Services, Second Edition. Baltimore, MD: Paul Brookes Publishing.

## ROLES OF PEOPLE ON YOUR EDUCATIONAL TEAM

EDUCATOR	ROLE
Teacher – Classroom	
Teacher – Special Ed	
Teacher – MSMI	
Teacher – DHH	
Teacher – Vision	
Teacher – O&M	
Teacher – DB Specialist	
Teacher – DAPE	
Teacher –	
Intervener	
Paraprofessional	
Interpreter	
OT	
PT	
Speech	
Audiologist	
Psychologist	
Principal	
Guidance counselor	
Director of Special Ed	
RLIF	
Vocational specialist	
Parent advocate	
SSB counselor	



## A CIRCLE OF FRIENDS

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A Circle of Friends is a circle of support, which forms around a person with a disability. It is meant to be a support to the person's inclusion into the school, community, and workplace, and thus, is considered an "inclusion tool". The person with the disability invites who he or she wishes to be involved in the circle, based upon who they feel supports them in their lives. For a person with limited communication skills, those closest to the person (parents, for example) would decide who would be invited.

Circles of Friends often start by using a person-centered-planning process: Making Action PlanS (**MAPS**), Personal Futures Planning, or Planning Alternative Tomorrows With Hope (**PATH**). Then the action plan is carried out by the members of the circle. Here are my experiences with it:

I am a member of a circle for a 35-year-old woman who has significant disabilities. After carrying out MAPS and action plans, we successfully assisted her in finding the needed supports to move from a sheltered workshop where she was doing meaningless piecework and hating her life, to a job in the community. The people involved in her circle were able to advocate with the agencies serving her to free up the money used for the segregated program to be used for the integrated program. She now has an in-home assistant who facilitates her inclusion into the community. Our next step as a circle will be to attempt to help her move from her parents' home to a home of her own, with the needed support. She is so much happier.

Circles of friends may or may not involve professionals, depending upon whether the person feels these folks are part of her/his support network. This person included her Developmental Disabilities (**DD**) caseworkers in her circle. Community Mental Health, the agency responsible for services to the DD population in Michigan, also supplied the circle facilitators.

The MAPS process is facilitated by two people: a verbal facilitator and a graphic facilitator (who records the ideas of the others through the use of words and imaginative graphics depicting the ideas so that everyone can access them).

My son Jacob's circle of friends includes his school friends, for the most part, although the deaf-blind consultant attends and helps facilitate. She has provided deaf-blind simulations, helped the kids come up with name signs for themselves to use with Jacob, and answered questions for the group. This year the general education teacher has been attending. This has never happened before, and really helps her to understand what the circle can provide to the school in terms of ideas for supporting Jacob. As much as we would like for the school personnel to attend, it happens seldom. We meet after school once a month, and the teachers just can't or don't want to stay that long, for something they don't understand. What is difficult is that I attend team meetings and circle meetings, and it is very clear to me that these two groups should be consulting with one another to facilitate Jacob's inclusion, but they just don't.

Jacob's circle has about 18 student members, about half boys/half girls. They have identified themselves over the years as the kids who really seem to connect with and care about Jacob in his inclusive setting (he is in sixth grade, this year). Although Jacob doesn't give these kids

much social feedback, they really care about him, and have hung in there with him. Most of them are charter members, since 1st grade. During each meeting we spend some time planning, snacking, interacting, and having fun. Until now, the circle has mostly focused on social interaction with Jacob. This year we are focusing more on transition to the middle school (there is only one middle school in town.) The kids have decided to go to all of the sixth grades in the city (six schools) to speak about Jacob and his circle of friends. Then when Jacob starts middle school inclusion, there will be (hopefully) much less "pointing at the freak" than there would otherwise be, because of understanding having been facilitated. Activities we have undertaken in the past couple of years have included the following:

Halloween party: the kids each bring a snack, decoration, and game that Jacob can access (usually they make something really neat), and something for the haunted house. (I don't have to bring anything!) They come in costume and have a great time. This year, after the db simulation, they decided to do the haunted house in the dark, to see what it would be like for a person who is blind.

Birthday Party: each year they decide to hold this in the same place, the local pool. They tried the gymnastic center one year, but Jacob hated it, and they learned that because swimming is his favorite activity, and because it is his birthday, the pool is a better place. One year in thinking about a party they were talking about what they would enjoy, and began to talk about going to a movie. One of the members spoke up suddenly and said, "We'd like that, but what would Jacob like?" The party was held at the pool.

Last year for his birthday they made him a quilt with photos of themselves scanned onto it. They decorated it with tactile paint, etc. We keep it on his bed, so he can see all of them. It is really beautiful. I will be bringing it to Indianapolis for my presentation on Circles, so all can see it.

Canoeing last summer: Jacob loved this. He sat in the middle at the bottom with another student sitting behind him, and trailed his fingers in the water. I didn't even have to be in his canoe, which made it much more fun for me!

School talent show: One year the circle did a "men in black" "Men in Black" number, really cute, all dressed up in ties and sunglasses, and Jacob was "the alien", because he was still using a wheelchair. They made (Jacob helped) a paper mache alien head to fit over his wheelchair, and we rigged up a jellybean switch for him to operate the lights in the eyes. It was a cool dance. You will see the video of that at the conference, too. Last year they did a number from Tarzan, Trashing the Camp. They dressed up as gorillas, and danced. Jacob and a few others stood in the back and were percussion, hitting pie plates with wooden spoons during the number.

One girl was running for student council president, and on her poster, she wrote:

"Activities I am Involved In:

Jacob's Circle of Friends

Junior Choir

Girl Scouts"

. . . . in that order!!! I was thrilled by this!

A volunteer special education student facilitates the circle each year from our university. This has worked out great, as I still have input, but much less work. She brings the snacks, supplies, etc.; I just reimburse her. Ideally, the circle would be facilitated during school by a teacher, and could include the whole class that Jacob is in. This is a different model, but would lend itself to more ideas being generated for inclusion in school lessons. This might be a good model for

Junior High.

Membership in the circle evolves, and will continue to. Some drop out, some join, but most stay the same. As they move away to college, etc., we hope people will continue to identify themselves as they get to know Jacob. We hope the circle continues throughout his life span, although this is difficult, as kids get busier with sports, adults get busy with studies, jobs, families. It will change, but hopefully, will always be there to fall back on in a time of crisis, even if through an email listserv!! Our ultimate goal is that Jacob will have a circle of support in place to take over decision making for him in case something happens to his father and me. He would have a guardian, of course, but also a "steering committee", with his best interests, dreams, and wishes at heart!

Another focus for us this year is fundraising. The kids (hopefully about 8 of them) are going to try to raise enough money to support their attending the CHARGE conference in Indy. I have invited them to help me present on Circles of Friends. I have had them do this in several forums before, and they do a GREAT job! I hope they can raise enough funds, because I really want all of you to meet these great kids. And their parents always extol the virtues of the circle, how valuable it has been to their character building, sensitivities to differences, compassion, and ability to think and plan for things. It also becomes a general circle of social support to all of them, not just for Jacob, but more and more for the group as a whole--they ALL support each other!

In a recent video interview, one charter circle member was asked why she thought the circle was important. I kind of held my breath, wondering what she would say. She blew me away. She said, "the circle is important because we help make sure that Jacob participates and is included in everything. That's important, because if he wasn't included in everything, he'd just be with teachers all day, and who would want to just be with teachers all day? That's no fun!" I loved that response, because it shows that she sees Jacob as a KID, first!

Lastly, these kids are the kids who will grow up to be leaders, workers, legislators, educators, etc. in our and other communities. They will use this experience to further the lives of people with disabilities in whatever career they pursue. They will grow up to be Jacob's employers, support persons, and friends, and employers, support persons and friends to others with disabilities. Jacob will have a group of people committed to ensuring he has a rich, life. Not just an integrated life, not just a life free of pain, abuse, neglect, segregation. An "enviable" life.

For more information on MAPs, PATH, PFP, or Circles of Friends, consult the following references:

Falvey, M.A. (1998). Inclusive and heterogeneous schooling: Assessment, curriculum, and instruction. Baltimore, MD: Paul Brookes.

Koegel, L.K., Koegel, R.L., & Dunlap, G. (1996). Positive behavioral support. Baltimore, MD: Paul Brookes

Pearpoint, J., Forest, M., & O'Brien, J. MAPs, Circles of Friends, and PATH: Powerful tools to help build caring communities. In Stainback, S., and Stainback, W. (Eds.), Inclusion: A Guide for Educators. Baltimore, MD: Paul Brookes





## PERSON-CENTERED PLANNING

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Person-Centered Planning is a process that facilitates inclusion of individuals with disabilities into natural environments, including general education classrooms in neighborhood schools, work environments, and other aspects of their natural communities. These techniques employ the use of large group graphics (large paper and colored markers which “graphically” illustrate the points being expressed), and facilitation techniques that enable groups of people to learn more about and plan a more positive future for a person with a disability and their family (Koegel, Koegel, & Dunlap, 1996).

The outcomes gained through Person-Centered Planning techniques include:

- Being present and participating in community life;
- Gaining and maintaining satisfying relationships;
- Expressing preferences and making choices in everyday life;
- Having opportunities to fulfill respected roles and to live with dignity; and
- Continuing to develop personal competencies (Koegel, Koegel, & Dunlap, 1996).

All person-centered planning approaches begin with a focus on the wants and needs of an individual and recognize the importance of both formal and informal supports in assisting the person to achieve his or her dreams (Mount, 1994).

### MAPs: Making Action Plans:

MAPs is a person-centered planning tool used with people with of all ages, and is frequently carried out by the person and their Circle of Support. A MAP has two facilitators. One who records what happens on large paper with colored markers, and the other facilitates the process. A personal and informal atmosphere is used (snacks, beverages, etc.) Everyone who is important in the child's life is present and participates. The focus during the process is on what the person and/or the family want. There is commitment to a plan of action (what can be done right away) and a date is set to meet again (Pearpoint, Forest, & O'Brien, 1996).

The following questions are asked of the person and the Circle:

- What is this person's history? What is your history with this person?
- What are your dreams for this person? What are this person's dreams?
- What are your nightmares for this person? What are this person's nightmares?
- Who is this person? (One or two word phrases)
- What are this person's strengths?
- What are this person's needs?
- What would a perfect school day look like for this person?/Action plan

From this person-centered approach, planning activities can then be carried out. These may include IEPs, team meetings, Circle of Friends activities, or anything else.

Pearpoint, Forest, and O'Brien (1996) write:

A MAP is not an academic exercise. A MAP is a genuine personal approach to problem solving. A MAP is for people who are vulnerable, and the outcome decisions of a MAP session have life and death implications for how the person will live his or her life. It is not a professionally controlled, expert-model, top-down management tool. A MAP is a group, problem-solving, cooperative, collaborative team approach to planning (p.74).

### **Circles of Friends: (or Circles of Support)**

A Circle of Support is a group of students who come together regularly to share and problem solve with and around the life of a person with a disability. These can be formal (i.e., with a facilitator and scheduled meetings), or informal (i.e., a group of friends that meets at lunchtime at school). Formal Circles of Friends may start with a MAPs process, and move on from there to help the person with a disability realize his or her dreams.

For students, a Circle is typically facilitated by school personnel, such as the classroom teacher, with either a group of children or the whole class, on a regular basis, to assist with person-centered planning for the child who has a disability in an inclusive setting. However, a Circle may be facilitated by a family or community member, outside of the school setting. For adults, these may be facilitated by case workers, or friends. The goal is to eventually withdraw the formal facilitator and let the naturally occurring peer relationships take over. The circle may evolve into a support circle for each member, and not just for the person with a disability.

Pearpoint, Forest, and O'Brien (1996) write:

A circle of friends is not a trick or a gimmick; it is a powerful tool. Like a chisel, it can pry open one's heart, soul, and thoughts or create a work of art. A work of art does not happen overnight; neither does building circles or communities. Circles and community building are commitments. Circles and community building are as important as math, physics, or history and are part of a curriculum of caring (p. 77).

### **Personal Futures Planning (PFP) and Planning Alternative Tomorrows with Hope (PATH)**

Personal Futures Planning and Planning Alternative Tomorrows with Hope are other person-centered planning tools, which expand on the MAPs process. All of these tools expand and alter the typical "system-centered" planning done for these individuals, and take into account the preferences, strengths, and individuality of an individual with a disability.

To implement a person-centered-plan or start a Circle of Friends for your child, contact your local agency serving people with developmental disabilities (i.e., in Michigan, Community Mental Health serves that purpose), or your child's school and ask them to research and carry out this process with your child.

# ASSESSMENT OF CHILDREN WITH CHARGE

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Editorial Comments by Sandra L.H. Danvenport, M.D.

Assessment should be driven by what questions the educational team and the parents have about the child. It should be used to guide the I.E.P. process. It should be a holistic process and not a tool that condemns the child to a perceived inability to learn.

Assessment does not require standardized measurements. Often, a "Portfolio Assessment" is far more helpful. The portfolio may include review of records, review of current developmental milestones, review of progress toward goals, and functional assessments of vision, hearing, etc. No standardized tests exist which can accurately assess children who have multiple sensory impairments. *A. standardized test score is like a drive-by shooting.*

Because in assessment drives the I.E.P. process, facts should be stated in the organized manner. In addition, it is extremely important that useful and valid recommendations be made as well.

*"Is it possible to accurately assess a child who does not see or hear, who has had so little exposure to intensive language stimulation that his or her primary means of communication is nonspeech vocalizations, body expression, and approximately 20 manual signs? Are there clues that would tell you he could learn given the right approach? Is it possible that the use of totally inappropriate assessment tools has added significantly to the attitude that this is not only an untestable child, but an unteachable one?" (Wolf-Schein, 1998).*

## What is important to know about assessment of children with CHARGE?

The first thing to note is that children with CHARGE are more different than they are alike. There is no "normal level of functioning" for this group of kids. There are adults with CHARGE who attend college and graduate school with minimal or no support, and there are adults with CHARGE who use one-on-one support personnel for all activities of daily living and work. It seems that some of the variation depends upon how much vision and hearing the child with CHARGE has. In a study on CHARGE and developmental delay (Hartshorne, 1999), I found that children with CHARGE who had better hearing and vision scored higher, overall, on a scale of adaptive behavior skills. I also concluded,

At present, there is no valid and reliable method for estimating the range of cognitive abilities for children with CHARGE, or for any subgroup within the deaf-blind population. However, because of these results, we can be relatively confident that the intellectual abilities of these children are routinely underestimated, due to other disabling factors.

We are able to accurately assess the intelligence levels of some children with CHARGE in traditional ways because they have little or no sensory impairment. However, there are many children with CHARGE for whom the use of traditional assessment procedures is grossly inappropriate. A child with limited hearing and vision will not do well on a traditional IQ test. If this method is used, it is likely to result in an underestimate of the child's true abilities, and inaccurate labeling. (See drive-by shooting quote, above.)

## **So, what should assessments look like for these kids?**

First and foremost, the primary purpose of assessment should not be to place a label on a child based on test performance. The primary purpose should be “to properly describe children’s current level of performance in order to point the way to the best intervention possible” (Wolf-Schein, 1998). The assessment should drive the IEP or IFSP. Having said that, assessments should address all of the following areas: (Anderson, Chitwood, & Hayden, 1998)

- Movement
- Communication
- Social Relationships
- Independence/Self-Concept
- Perception/Senses
- Thinking Skills
- Learning Style

In addition, assessments of children with CHARGE should include an evaluation of functional hearing, functional vision, and functional communication skills. How does the child hear, see, and communicate in different contexts?

When possible, assessments should include observation of the child working in familiar environments, with familiar persons. This can provide a wealth of information on learning style, communication, social skills, behaviors, and abilities.

Portfolio assessment can sometimes be an appropriate option for use with children with CHARGE. This can include review of records, interviews with professionals and parents, review of current developmental milestones, and review of progress toward goals.

## **What should assessment reports look like?**

Because children with CHARGE have such complex and multiple needs, assessment reports should state facts and findings in an organized manner. In addition, it is critical that reports include useful recommendations for service providers and parents. These should include recommendations about functional skills and activities in which the child engages.

Functional skills are those that enable a child to function independently in natural environments. Ask yourself, “Is this a skill this child needs to learn in order to function as an adult?” For example, a child could practice the cognitive skill of categorizing by color in many ways. A non-functional way would be to have the child place colored blocks into containers for the sole purpose of the task itself. A functional way to practice this skill would be to have the same child sort construction paper into drawers or files for someone to use at a later time. A child learning one-to-one correspondence could do so by inserting pegs into a pegboard. The same child could learn the same skill if she were passing cookies out to her classmates. It is essential that the assessment itself, the results, and the recommendations address skills that will be functional for the child in his or her natural environment.

## **Quality Indicators and Desired Outcomes of Evaluations of children with CHARGE**

Mar (1998) identifies the following as indicators and outcomes of quality evaluations for children who are deaf-blind. These indicators are also appropriate when evaluating children with CHARGE. A quality psychoeducational evaluation should:

- identify educational/psychosocial concerns;
- use meaningful and relevant tasks;
- use multiple procedures;
- use multiple observations of communication behaviors and social interaction skills in natural routines;
- describe degree of participation in tasks, not failures and inabilities;
- focus on how the student acquires information rather than on test scores;
- involve parents, teachers, and other service providers in information gathering;
- assess pragmatic, functional home and community skills.

A quality evaluation for these children should result in these outcomes:

- positive outcomes for student, parent, team;
- increased understanding of communication behavior;
- a description of competencies, difficulties, progress, and environmental factors affecting performance;
- a description of behavioral concerns and recommendations for supports and services;
- avoidance in use of jargon, technical language, unnecessary references to scores or age levels, and comparisons to the “norm”;
- a description of social interaction skills, involvement with peers, and participation in school and community, and recommended strategies to increase these;
- a description of goals the student might be expected to attain and general strategies to help the student attain them;
- a discussion of strategies for dealing with problem behaviors using positive support such as reinforcers, preferred activities, redirection, and communication training;
- suggestions for age-appropriate and meaningful materials and activities.

Finally, Mar (1998) has developed a “Consumer Test” of assessment outcomes. This is perhaps the most useful set of questions to consider when evaluating a child with CHARGE.

### **CONSUMER TEST OF ASSESSMENT OUTCOMES**

- Will the evaluator be able to describe sufficiently my competencies and the ways that I best communicate?
- Will the assessment indicate what progress I have made?
- Will the assessment be used to determine my needs, desires, choices, and interests in everyday situations and settings?
- Will the assessment help my family, teachers, and schoolmates to interact with me better?
- Will the assessment lead to interventions that make sense to me?
- Will the assessment identify the services and help I really need?
- Will the assessment help other people who work with me?
- Will the assessment contribute to the long-term plan?

### **References**

Anderson, W., Chitwood, S., & Hayden, D. (1997). Negotiating the Special Education Maze: A Guide for Parents and Teachers, 3<sup>rd</sup> Edition. Bethesda, MD: Woodbine House

Hartshorne, N. (1999) The prevalence of mental retardation in CHARGE Association. Paper presented at the 4<sup>th</sup> International CHARGE Syndrome Conference for Families, Houston, TX.

Mar, H. (1998). Psychological Evaluation of Children who are Deaf-Blind: An Overview with Recommendations for Practice. DB – LINK: The National Information Clearinghouse on Children Who Are Deaf-Blind.

Wolf Schein, E. (1998). Considerations in assessment of children with severe disabilities including deaf-blindness and autism. International Journal of Disability, Development and Education, 45, 35-55.

# PSYCHOEDUCATIONAL EVALUATION OF CHILDREN WITH CHARGE SYNDROME

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*"Is it possible to accurately assess a child who does not see or hear, who has had so little exposure to intensive language stimulation that his or her primary means of communication is nonspeech vocalizations, body expression, and approximately 20 manual signs? Are there clues that would tell you he could learn given the right approach? Is it possible that the use of totally inappropriate assessment tools has added significantly to the attitude that this is not only an untestable child, but an unteachable one?"(Wolf-Schein, 1998).*

Under special education law, all children must have an appropriate evaluation in order to be identified as eligible for special education services. The evaluation also serves to identify a child's learning difficulties. During the evaluation process, information will be obtained about a child's present abilities. This information will assist the school team to understand how the child learns, and to plan for his or her educational program. This evaluation should drive the goals and objectives included in the Individualized Education Plan (IEP) or the Individualized Family Service Plan (IFSP).

The Individuals with Disabilities Education Act (IDEA) requires that the assessment be conducted by a Multidisciplinary Team. This means that individuals with different areas of expertise should be involved. The team is often led by a school psychologist. Testing procedures used by schools must meet the following requirements (Anderson, Chitwood, & Hayden, 1997):

- The tests and evaluation materials are administered in your child's native language and primary means of communication.
- The tests must be professionally approved for the specific purposes for which they are used.
- The tests must be given by trained professionals according to the instructions of the publishers of the tests and materials
- Tests and evaluation materials that assess a wide range of educational and developmental needs and capabilities should be used in addition to tests designed to provide a single general intelligence quotient.
- Tests should be selected and administered so as to ensure that they accurately assess the child's aptitude, achievement level, or other factors they are designed to measure, rather than reflecting the child's disabilities.
- The evaluation should be undertaken by a team or group of persons from several professional backgrounds, including at least one teacher or other specialist with knowledge in the area of your child's suspected disability. Your child should be assessed in all areas related to the suspected disability, including, where appropriate, health, vision, hearing, social and emotional status, general intelligence, academic performance, communication skills, and motor abilities.

Depending on the child's age and overall level, assessment can take place in the home, school, or both. Assessment at home may take the form of the examiner making general observations, and asking questions of parents, or the examiner may enlist help from parents to administer test items, so that the best performance possible may be obtained from the child.

One alternative way for examiners to evaluate students with more severe disabilities is through the use of "portfolio assessment". This may include review of the child's records, review of the child's current developmental milestones, and review of the child's progress toward current goals. It will include teacher and parent interviews. Also included may be functional assessments of vision, hearing, and communication.

#### General Tips for Parents:

- Ask for an evaluation meeting with the team prior to the assessment to determine the focus of the evaluation. Evaluation should be driven by questions the educational team has, and not by "what seems like the best test battery to use". Once the team decides what questions they want answered, finding appropriate ways to get these answers during the assessment is much easier. Assessments need not be tests that yield standardized scores. More often than not, IQ testing for a child with CHARGE will be inappropriate, because of vision and hearing losses. An IQ score that is inaccurate tells you nothing about how the child learns, and may lead to an unnecessary and inaccurate label of mental retardation. *"A standardized test score is like a drive-by shooting."*--**Whose quote is this???**
- When you can be present during the evaluation, be sure to watch for your child's fatigue and stress levels. It will help if you can give your child some time to become familiar with the examiners. If allowed, observe the assessment session. Note how well you think your child did, and let the examiner know what you see. You know your child best, and this information will be valuable to the assessment.
- Contribute to the assessment process as much as you are comfortable. Information you provide about your child's abilities, temperament, and medical issues will help the examiners to make sense of assessment information. The examiners may have specific questions for you or evaluation instruments for you to fill out on your child.
- Ask the examiner about his or her familiarity with CHARGE, with vision and hearing impairments, and with deaf-blindness, if appropriate. It may be helpful to connect the examiner with the CHARGE Syndrome Foundation, or with your state's deaf-blind technical assistance center.
- Mar (1998) has developed a "Consumer Test" of assessment outcomes (see next page). This is perhaps the most important set of questions to consider when your child is evaluated. It is from the child's viewpoint. It may be useful to share this list with your child's evaluation team.



## CONSUMER TEST OF ASSESSMENT OUTCOMES

1. Will the evaluator be able to describe sufficiently my competencies and the ways that I best communicate?
2. Will the assessment indicate what progress I have made?
3. Will the assessment be used to determine my needs, desires, choices, and interests in everyday situations and settings?
4. Will the assessment help my family, teachers, and schoolmates to interact with me better?
5. Will the assessment lead to interventions that make sense to me?
6. Will the assessment identify the services and help I really need?
7. Will the assessment help other people who work with me?
8. Will the assessment contribute to the long-term plan?

For more information on psychoeducational evaluation and assessment, consult the following references:

Anderson, W., Chitwood, S., & Hayden, D. (1997). Negotiating the Special Education Maze: A Guide for Parents and Teachers, 3<sup>rd</sup> Edition. Bethesda, MD: Woodbine House

Hartshorne, N. (1999) The prevalence of mental retardation in CHARGE Association. Paper presented at the 4<sup>th</sup> International CHARGE Syndrome Conference for Families, Houston, TX.

Mar, H. (1998). Psychological Evaluation of Children who are Deaf-Blind: An Overview with Recommendations for Practice. DB – LINK: The National Information Clearinghouse on Children Who Are Deaf-Blind.

Wolf Schein, E. (1998). Considerations in assessment of children with severe disabilities including deaf-blindness and autism. International Journal of Disability, Development and Education, 45, 35-55.



## INDIVIDUALIZED EDUCATION PROGRAMS (IEPs)

## INDIVIDUAL FAMILY SERVICE PLANS (IFSPs)

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By law, through the Individuals with Disabilities Education Act (IDEA), any student receiving services through special education must have an IEP or IFSP. These are plans developed by the educational or early intervention team serving the student. Parents must be included in this team, and in all decisions made about the child's education, by federal law. IEPs will include the following components:

- A description of the student's present levels of performance in several areas
- Annual goals for the student and short-term objectives to help them meet those goals
- A description of the specific special education services and related services the student will receive and where and when they will take place
- When the services will start and how long they will last
- How the student's progress on the goals and objectives will be evaluated

An IFSP is written for infants and toddlers and their families, and will include goals and objectives for both the child and the family to assist the child with early development, as well as a plan for transitioning to school-based services after age 2. This plan should always be developed in a family-centered way, meaning professionals make certain the family's priorities come first. For a child with CHARGE, assistance with case coordination (coordinating between medical and educational personnel) may be a high priority. The following components are included in an IFSP:

- A description of your child's development
- Your family's concerns and needs
- What your family hopes and dreams for your child
- What services your family needs to realize these dreams
- In what environment will the services be delivered?
- What other services are needed by your family?
- When will these services begin, and how long will they last?
- Who will coordinate the services?
- What is the plan for transition to school-based services?

These plans are written to assure that each child with a disability will receive a *Free Appropriate Public Education (FAPE)*. This means that public schools are required to provide special education services to a child with disabilities, even if the child attends a private or residential school. Appropriateness is decided upon by the educational planning team, which includes the parents. This team of people designs an individualized plan for the child, based upon the child's needs. This plan is written in the IEP or IFSP document. Related services are also available to students with disabilities, based upon educational need. Related services can include the following, and any or all of these should be considered for a child with CHARGE:

- assistive technology
- audiology
- counseling
- occupational therapy
- parent training
- physical therapy
- psychological services
- recreation
- school health services
- social work services
- speech and language therapy
- transportation
- vision or hearing services
- orientation and mobility

For children under age two, services may be a bit different, and may be provided by an agency other than the school system, such as health departments. However, you and your child will be served through these agencies by specialists in the area of infant and toddlers development.

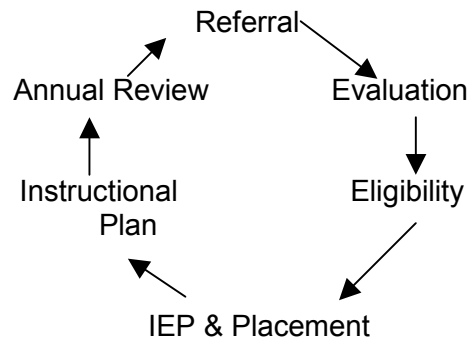
IDEA also requires that students be served in the *Least Restrictive Environment* (LRE). This means that students with disabilities are guaranteed equal access to education, and must receive education where they are least restricted from peers without disabilities. Each IEP team determines how much time the child will spend being educated alongside peers without disabilities. “Students are to be educated in a separate classroom or school only when the nature and severity of their disabilities makes it impossible to meet their educational needs in a less restrictive environment.” (Anderson, Chitwood, & Hayden, 1997). This part of the law is what has led to the recent movement toward inclusive education for children with disabilities.

At age 16, a student’s IEP must include a plan for transition. This can be devised and included as early as age 14. This is most easily accomplished through the use of person-centered-planning techniques. These are a set of activities centered around the student, which take into account the student’s needs, preferences, and interests when planning for the future. Desirable outcomes of a good transition plan can include:

- post-secondary education
- vocational training
- integrated employment (including supported employment)
- continuing and adult education
- adult services
- independent living (including supported living)
- community participation

It is important that a student’s program be written before a placement decision is made. It is somewhat typical of school districts to automatically group students into classrooms that match the students’ labels. For example, a child with autism could be placed in a classroom for children with autism, based on the label, instead of looking at that child’s specific needs, first, and then deciding upon where those needs can best be met. It is important to keep in mind that the label placed on your child should never solely determine their classroom placement. The school team must look at your child’s individual needs, first.

Anderson, Chitwood, & Hayden (1997) refer to the “school system’s cycle” for special education services. In general, it works like this: The cycle begins with **referral** of a student for special education services. The student then undergoes **evaluation** to determine if he or she meets **eligibility** requirements to receive services. If eligible, an **IEP** convenes, and **placement** decisions are made. From the IEP, **instructional plans** are made and carried out during the school year. Each year, an **annual review** IEP must be conducted to review the student’s progress on goals and objectives, and to revise the instructional program. Every three years, a re-evaluation of student eligibility and needs takes place. This is generated by a new **referral**.



### Due Process

IDEA grants each child with a disability the right to legal “Due Process.” More specifically, the following steps are required of school districts:

- Provision of A Free Appropriate Public Education (FAPE) is provided for all children with disabilities between the ages of 3 and 21 in most states
- Children are educated with their peers without disabilities to the maximum extent possible
- Specially designed educational and related services, designed to meet the unique needs of the child, must be written in the IEP
- Parental consent must be obtained before an initial evaluation or a first placement in a special education program takes place
- Reasonable notice must be given to families before an evaluation, placement, or change of placement takes place, or when the school district refuses to carry out these actions
- Evaluations must be carried out by a multi-disciplinary team, including one specialist knowledgeable in the area of the student’s suspected disability
- Tests used for evaluation must not be discriminatory
- Parents may inspect and review all records
- If parents disagree with evaluation results, the school district must pay for an independent evaluation if requested
- If parents believe their child’s rights have been violated, an impartial due process hearing may be requested

Your child’s Due Process rights can empower you to advocate for your child’s best interests.

Finally, here are a few useful tips to consider when preparing for your child's IEP:

- Take a photo of your child with you and place it on the table at the meeting where everyone can see it. This will help to keep the focus on your child as an individual.
- Take an advocate or friend with you. IEPs can be intimidating to parents, with many professionals with professional opinions sitting around a table telling you what they think is best for your child. Just having a friend along to sit next to you and help you listen or take notes can give you more confidence to say what is on your mind and advocate for your child's needs and rights. Alternatively, ask another parent of a child with a disability who has "been there" to attend the meeting with you, and to speak up when they feel advocacy is needed.
- Be assertive, but not aggressive. Repeat yourself concerns if you feel they are not being heard. As difficult as it is not to become very emotional during these meetings, try not to lash out in anger at the team. You are going to have a long-term relationship with these people, who will be teaching your child. It is better to calmly state your concerns, know your rights, and remember that you are the utmost authority on your child. You know your child better than anyone!
- Trust yourself. Trust your instincts. As a parent of a child with CHARGE, you have a wealth of knowledge and are the person most invested in your child's future.
- If, after discussion and advocacy, you do not agree with what is being written in the IEP, you may, on the document itself, indicate your disagreement and either request further discussion, mediation, or a due process hearing.

To read more about IEPs, consult this excellent and user-friendly resource:

Anderson, W., Chitwood, S., & Hayden, D., (1997). Negotiating the Special Education Maze: A guide for Parents and Teachers. Bethesda, MD: Woodbine House

## DOS AND DON'TS FOR EDUCATORS

Cathy Lyle, M.Ed.

Educational Consultant, Minnesota DeafBlind Project

*HOT plus DOG does not equal HOTDOG*

*DEAF plus BLIND does not equal DEAFBLIND*

DO build a relationship with the child. The child that does not trust you cannot learn from you.	DON'T assume that what works for other children will work for this child.
DO celebrate progress, however small.	DON'T underestimate child's ability to learn.
DO help the child develop a positive outlook on life and love learning.	DON'T get so focused on what the child can't do that you fail to see what the child is <u>can</u> do.
DO remember that <b>ALL BEHAVIOR IS COMMUNICATION</b> .  (See: "Behavior as Communication")	DON'T assume a child has to speak or sign words in order to communicate. Body language, facial expressions and gestures can speak volumes. In fact, an infant's understanding of spoken words is based on reinforcing naturally occurring babbling like "mama" or "dada." Likewise, a deaf child's naturally occurring gestures like the thumb of a flat open hand touching the chin or forehead are also reinforced to mean "mama" or "dada."
DO work with parents as partners. Enlist the help of parents to get at the likes and dislikes of the child so the entire team can build on naturally occurring communication.	Don't ignore the parents or belittle their contributions and observations.
DO become familiar with both hearing and vision loss as well as the definition of deafblindness.	DON'T assume that a child with <u>both</u> vision and hearing loss can be served by adding the techniques suggested by the teachers of the deaf/hard of hearing to those by the teachers of the blind/visually impaired.

DO get on the child's "radar," i.e. be sure to communicate within the child's "communication bubble." (See Communication)	DON'T assume a child sees clearly just because he/she can follow movement or understand what you say just because he/she turned toward your voice.
DO understand the significance of medical problems.	DON'T let all the medical "stuff" hinder interaction with the child. These kids don't break. Let the nurse or personal care attendant worry about the whistles and buzzers.
DO teach functional skills.	DON'T teach skills in isolation.
DO teach experiences from beginning to end.	DON'T expect the child to get more than 10-15% by incidental learning. A child who is DeafBlind may think that people and objects appear and disappear by "magic."
DO help the child learn to anticipate what is coming using touch cues, object cues, calendar boxes, etc.	DON'T surprise the child by suddenly picking up the child, taking toys away, putting something in the mouth, suctioning, etc. without letting the child know what is going to happen.
DO find out what the child likes and does not like so they can learn about motivation.	DON'T always expect a child to learn what you want him/her to like.
DO let the child make choices, get messy and learn about consequences.	DON'T make all the decisions <u>for</u> the child.
DO encourage and facilitate social interaction with peers.	DON'T shield the child from hurt feelings. He/she needs to understand what other children think, too.
DO encourage consistent and fluent communication between the child and every person who is in constant contact including parents, nurses, teachers, siblings, interveners, etc.	DON'T assume the responsibility of communication <u>for</u> those people.

**DO LOVE THESE KIDS BECAUSE THEY WILL LOVE YOU BACK!**



## **ORIENTATION AND MOBILITY (O&M)**

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### **What is O&M?**

Orientation is knowing who you are (body image), where you are, where you want to go and planning how to get there. Mobility is the action of moving from place to place.

### **What is an O&M Specialist?**

The Orientation and Mobility Specialist (COMS) is a certified professional who teaches children and adults with visual impairments how to travel safely and independently in familiar and unfamiliar environments.

### **What skills does an O&M Specialist teach?**

- Sensory awareness: learning about the world through the senses (vision, touch, smell, hearing and taste.)
- Body image: awareness and understanding of body parts and how they move.
- Object permanence: knowing that objects exist even when they are not felt, heard or seen.
- Spatial concepts: understanding the relationship between objects in the environment and one's body (in front of/behind, etc.); awareness of both near and distance space (close/far); awareness of distances between objects.
- Searching skills: locating items in all directions, within arm's reach and beyond.
- Independent movement: rolling, scooting, crawling and walking.
- Sighted guide: using a sighted person as a primary means of travel.
- Protective techniques: using one's arms for protection while moving through open space.
- Cane skills: using specific techniques with a cane or adaptive mobility device for more independent and safer travel.
- Trailing: using one or two hands to follow along a surface (wall, furniture) to locate a specific objective and/or maintain a straight line of travel.

### **O&M considerations for the child with CHARGE**

Children with CHARGE need to learn about the world through their residual senses.

**Sight** gives us some of the most important information about our environment. When we enter a room, with a "quick look" we determine how big the room is, the size, colors, objects, etc. A child with CHARGE may have one or more visual problems (see section THE EYES IN CHARGE). Depending on how the eyes are affected, a child may or not have difficulties moving safely in his environment. The child with a retinal coloboma will have an upper field loss, and bring his head up to see better. This head position limits his ability to see low obstacles or changes in the terrain, and may cause stumbling or falling. The child with a coloboma of the macula or optic nerve will have blurry vision as well as large blind spots that affect his ability to see details. Being able to interpret visual cues with residual vision is an important skill for safe travel.

**Hearing** also tells us a lot about the world around us. Two important auditory skills used for orientation and mobility are sound localization and sound discrimination. Sound localization is the ability to determine where sounds are coming from, and can encourage a child to reach and move toward the sources of those sounds. Sound discrimination is the ability to distinguish between sounds. Children learn this skill by having repeated opportunities to hear everyday environmental sounds and pairing them with their sources, e.g., the blender, the radio, the car. Hearing loss is very common in children with CHARGE. Factors that will influence a child's ability to use residual hearing include the type and severity of loss, whether the child wears amplification, how long he has been aided, and the amount of appropriate auditory training the child has received to help him better understand what he is hearing.

**Touch** includes manipulation of objects, dexterity, exploration and identification of objects, as well as discrimination with the feet. A great deal of travel information is gained through the feet to determine the surfaces we are walking on.

**Smell** is an important sense for orientation and mobility. Many familiar locations can be identified through smell: bathroom, kitchen, different aisles in the grocery store, etc. (Because of cranial nerve damage, some children with CHARGE are unable to smell.)

**Movement** is an essential way we learn about our bodies and about our environment. Children with hearing and vision impairments need to feel safe when they are moving. They also need to be motivated and have a purpose for moving. They may have difficulty generalizing concepts and experiences from abstract toys or objects, and need opportunities to play with real objects. They can do this by being given opportunities to manipulate and explore tools used in daily routines such as spoons, cups, purses, boxes, containers, tooth brushes, other brushes, bottles, water, bread, etc.

### **Early Intervention**

Early intervention in O&M for children who are visually impaired has become an important area of programming. It may be difficult at first, to distinguish O&M objectives from those of the OT, PT, or VI Teacher. The O&M and PT often have similar movement related objectives for a young child with CHARGE, and there may be some overlap. Good communication among members of the child's educational team (parents, professionals and paraprofessionals) is crucial. Initially, the OT may be directly involved with the child, while the PT, VI or O&M consult. The O&M specialist may become more directly involve with the child later, as she begins to move through space.

### **References**

Hill & Ponder, Orientation and Mobility Techniques: A guide for the practitioner. American Foundation for the Blind, New York 1976

SKI\*HI Institute: "A Resource Manual for Understanding and Interacting with Infants, Toddlers, and Preschool Age Children with Deaf-Blindness." Department of Communicative Disorders, Utah Sate University, 1993

Pogrund, et al: TAPS, An Orientation and Mobility Curriculum for Students with Visual Impairments. Texas School for the Blind and Visually Impaired. 1993

# THE IMPACT OF SENSORY INTEGRATIVE DYSFUNCTION IN CHARGE

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Sensory integration is the organization of sensation for use. Countless bits of sensory information enter our brain at every moment, not only from our eyes and ears, but also from every place in our body. Sensations are food for the brain that provide energy and knowledge needed to direct our body and mind. The greatest development of sensory integration occurs during an adapted response; a purposeful, goal directed response to a sensory experience. In an adaptive response, we master a challenge and learn something new. At the same time, the information of an adapted response helps the brain to develop and organize itself. The first seven years of life our brain is a sensory processing machine nourished by having fun through play and movement. The child who learns to organize his play is more likely to organize his activities that are required for daily living.

If the brain does a poor job of integrating sensations, this will interfere with many things in life. The brain is not processing or organizing the flow of sensory impulses in a manner that gives the individual good, precise information about himself or his world. Learning is difficult and the individual often feels uncomfortable about himself and cannot easily cope with demands and stress. If the individual is blind or visually impaired his difficulty is compounded when attempting to make sense of his world.

Complex medical problems associated with CHARGE Syndrome at birth may result in delayed sensory integration development. This delay may be due to either neurological disorders or the inability to partake in sensory experiences that nourish the brain because of medical fragileness. Symptoms of irregular sensory processing in the brain are different for each child. There are three basic sensory systems that impact how a child learns and behaves in his environment. They are the tactile, vestibular, and proprioceptive systems. The following outline is a brief description of symptoms observed in each of the three systems when dysfunction of sensory processing is present:

## **The Tactile System:** discriminative touch versus protective touch

Dysfunction in the *discriminative* system may result in:

- Difficulty with fine motor skills for feeding, dressing, and writing
- Problems articulating sounds due to inadequate information from touch receptors in and around the face and mouth (cranial nerve VII abnormalities included)
- Difficulty with accurate visual perception and basic concepts
- Impaired awareness of body scheme
- Inefficiency in how one tactually explores an object or the environment in order to gain additional cues which may give meaning about the object or environment
- Appears to contribute to somato dyspraxia – a specific disorder in motor planning

Dysfunction in the *protective* system may result in:

- Interpreting ordinary contact as threatening
- May be frequently in a state of *Red Alert*
- May react with flight/fright/or fight – these behaviors may be physical or verbal
- May be termed tactually defensive
- Some feel too much; some feel too little. Some may have a high tolerance for pain because they do not accurately know what is happening to them
- They may not react to being too cold or too hot because they are unaware of temperature

**Proprioceptive System:** unconscious awareness of our muscles and joints that constantly send information to our brain to tell us of our body position and posture.

Dysfunction in *proprioception* results in:

- Slower body movements
- More clumsy movements
- Movements involve more effort
- Difficulty grading muscle force – muscle exertion is either too much or not enough when manipulating objects
- Difficulty feeling the weight of objects
- Difficulty planning body movements while performing gross or fine motor activities (getting on or off a riding toy, buttoning clothes, turning on a faucet, etc.)

**Vestibular System:** responds to the position of the head in relation to gravity and accelerated movement (are we moving?) or decelerated movement (are we still?). Vestibular receptors are the most sensitive of all sense organs and are major organizers of sensations in all other sensory channels.

The location of the vestibular system is in the inner ear called the “labyrinth”. Since abnormalities of the ears and hearing loss are common features in CHARGE, the influence of this system plays a major role in the developmental milestones of sensory processing and gross motor skills for these children.

Influence of vestibular system on *eye and neck muscles*:

- Ability to follow objects
- Ability to move eyes from one spot to another
- Ability to interpret -- is it an object, our head, or our whole body that is moving?
- Ability to interpret – is our head moving or tilted?
- Ability to maintain a stable visual field

Influence of vestibular system on *muscles of the body*:

- Helps to generate muscle tone
- Helps us to move smoothly, accurately, and with proper timing

Influence of vestibular system on *postural and equilibrium responses*:

- Helps with balance
- Facilitates spontaneous body adjustments
- Facilitates co-contraction of muscles
- Helps to elicit protective extension

Other areas influenced by the vestibular system:

- *Reticular Interactions* – responsible for arousal of nervous system (calming effects vs. arousal effects); vestibular system keeps level of arousal balanced.
- *Relation to Space* – perception of space; position and orientation within that space.
- *Auditory Processes* – helps the brain process what is being heard; vestibular disorders slow down speech development.
- *Emotional Development/Behavior* – for emotions to be balanced the limbic system (generates emotionally based behavior) must receive well-modulated input from the vestibular system.

## **Two types of vestibular disorders:**

### **1. Underreactive vestibular system**

- Child may tolerate an enormous amount of movement (merry-go-round, swinging, spinning) without getting dizzy or nauseous.
- Has poor integration of the two sides of the body
- Is easily confused by directions or instructions
- Hands do not work well together, nor do his feet

### **2. Overreactive vestibular system: the child is hypersensitive to vestibular input resulting in:**

- *gravitational insecurity* – a feeling of anxiety or stress when assuming a new position, or when someone else tries to control his movement or position; swings, merry-go-rounds, and other playthings that move the body in nonordinary ways is terrifying.
- *intolerance to movement* – discomfort during rapid movement; the child is not necessarily threatened by movement, it just makes him uncomfortable, or possibly even become nauseous.

## The Next Step: Evaluation/Intervention

### Evaluation:

If there is suspicion that a child has dysfunction with sensory motor processing, an evaluation can be conducted by either an *occupational therapist* or *physical therapist*. Evaluation consists of both standardized testing and structured observations of responses to sensory stimulation, posture, balance, coordination, and eye movements. The therapist who conducts the testing may also informally observe spontaneous play, and may ask the parents to provide information about their child's development and typical behavior patterns. A report will follow the evaluation that provides test results and interpretation of what the results indicate. The therapist will then make recommendations regarding the appropriateness of therapy using a sensory integrative approach.

### Intervention:

Providing intervention based on the principles of sensory integration theory requires that the therapist be able to combine a working knowledge of sensory integration theory with an intuitive ability to engender a child's trust and create the just right challenge. Therapy will involve activities that provide vestibular, proprioceptive, and tactile stimulation, and are designed to meet a child's specific needs for development. The activities will also be designed to gradually increase the demands upon a child to make an organized, more mature response. Emphasis is placed on automatic sensory processes in the course of a goal-directed activity, rather than instruction or drilling the child on how to respond. Parent or teacher involvement is also crucial to the success rate of the child's development with sensory processing. The therapist may make suggestions to the parent or teacher about how to help the child in the home or school environment.

For a listing of therapists certified in the evaluation of sensory integration you may contact:

Sensory Integration International  
P.O. Box 9013  
Torrance, California 90508

Telephone: (310) 320-9986

## REFERENCES

Ayres, Jean A. (1979). *Sensory Integration and the Child*

Fisher, Anne G., Murray, Elizabeth A., Bundy, Anita C. (1991). *Sensory Integration Theory and Practice*

Sensory Integration International (1991). *A Parent's Guide to Understanding Sensory Integration*

# PHYSICAL THERAPY AND OCCUPATIONAL THERAPY IN CHARGE SYNDROME

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**Dual Sensory Impairment:** As dual-sensory impaired (deaf-blind, or hearing loss and vision loss), and medically fragile, children with CHARGE are at risk for a variety of motor and sensory delays. Even early on, when medical issues are huge concerns, it is important to begin some early intervention, especially with the child's sensory system. The child with CHARGE will need to learn to rely on his other senses (touch, smell, taste, vestibular and proprioception/pressure sense) to learn about his world.

The pyramid on the following page illustrates how the sensory systems are the basic building blocks for learning motor and cognitive skills. It also shows the importance of early intervention for infants by sensory and motor specialists.

The Central Nervous System represents the newborn, and Academic Learning represents the beginning of school. It becomes obvious that Occupational Therapy' (sensory motor and perceptual motor specialists) evaluations are important very early in an infant's development. Physical Therapy' (gross motor and developmental specialists) evaluations become important slightly later on. Orientation & Mobility Specialist (teachers of safe and independent travel, body concepts, and spatial concepts) evaluations and recommendations become important slightly later on yet. But all have a role quite early in the child's overall development. For example, the occupational therapist role can begin very early, in an infant's life, even before the infant is medically stable. As sensory-motor experts, they can help evaluate and address issues of hypersensitivity, oral-motor skills, mouthing/exploring objects, etc.

Since OT and PT roles often overlap, especially in the pediatric setting, PT can sometimes substitute for an OT evaluation if OT is not available. Also, it is important to note that an early-on O&M (Orientation and Mobility) consult can be very helpful. The whole concept of moving in space as a blind individual relies on pre-existing skills of exploration, using hands functionally, recognizing objects and their use, and recognizing object permanence, all of which the PT and OT are influencing.

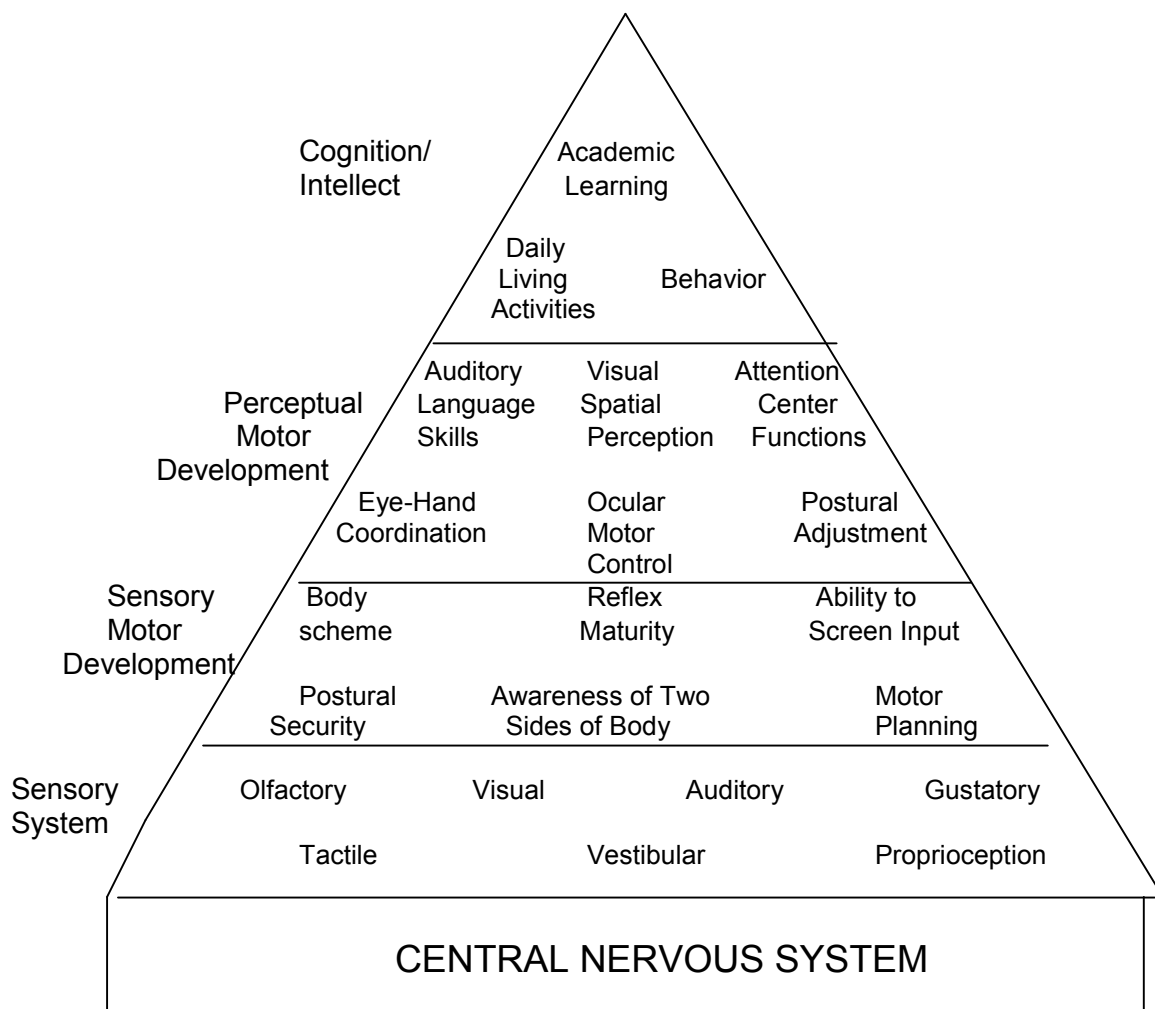


Chart is taken from Williams & Shellenberger: "How does your engine run"

**Once a child enters school**, the following information becomes important:

#### **General Considerations:**

- OT and PT in the school setting must be based on School/Educational issues. OT/PT services support the IEP (Individual Education Plan). School-based OT/PT services support school goals. *It is not the do all, end all, only source of OT or PT services the child should be accessing.*
- Although OT and PT may do certain evaluations without a doctor's referral, *all* PT services (and *some* OT services) *must* be prescribed by a doctor. *But*, not all doctor-prescribed OT/PT services is appropriate for a *School-based* Therapist to perform (school-based therapy is to support school goals).



## **General Roles of the OT and PT**

The OT (Occupational Therapist) primarily supports arm/hand strength and coordination (fine motor) issues, sensory motor issues, sensory integration, activities of daily living, oral motor issues, arm/hand splinting and accompanying issues.

The PT (Physical Therapist) primarily supports gross motor issues of balance, walking, accessing doors, stairs, curbs, posture, wheelchair transfers and other wheelchair issues, leg bracing and accompanying issues.

Both OT and PT can be delivered in a Direct Services model or in a Consultation model (or in a combination of both). *Direct Services* are for those students who need one-on-one intensive physical programming (and are able to participate in such). *Consultative Services* are for those who would learn skills better with a more integrative approach to physical issues. For example, an OT could use direct service to teach a specific motor pattern for scooping, or a PT could teach a pattern for negotiating stairs or they could consult with the teaching and dorm staff on the pattern that the student should utilize for scooping and stairs, so the child could practice the skills daily (not just in a therapy session).

## **Specific Roles of OT/PT**

- Mobility:** This entails evaluating and teaching students the skills needed to physically negotiate campus. It includes negotiating on level surfaces, unlevel surfaces (grass and inclines), doors, stairs, curbs. It includes recommending or evaluating any adaptive equipment needed to accomplish this (wheelchairs, crutches, support canes, etc).
- **Transfers/Transitions:** This entails evaluating and teaching students independent transfers to/from beds, chairs (including wheelchairs), the floor. It includes teaching staff how to safely assist those students who have weak transfer skills. It also includes recommending/evaluating any equipment which may be helpful to the staff and/or the student (footstools, safety belts, grab bars, special transfer devices/lifts etc).
- **Positioning:** This entails evaluating a student's muscle tone and function in relation to how he sits and participates in table-top activities. If a student is not able to rely on his trunk strength to support himself in an upright position, he will be unable to use his hands in a functional, meaningful way. He will need his arms and hands to prop himself and support himself on the table or desktop instead. Many times adaptive seating is indicated to obtain a nice supported, upright posture and to 'free up' the hands and arms for successful tabletop or desk-top activities.
- **Independent Living Skills:** This entails evaluating and teaching students independent skills in a variety of independent living areas. Examples:

**Dressing:** Evaluating a student's skills and assisting w/ setting up a dressing program for improved independence. This includes consideration for adaptive equipment (button hooks, Velcro closures for clothing, adaptive shoe laces, etc). It can also include teaching the student or the staff shoe-tying techniques.

**Eating:** Evaluating a student's skills w/ food. This includes the actual arm/hand skills of eating (how does the child finger feed? how does he hold a utensil? can he physically get his hand to his mouth? can he sit well enough to use his hands

to eat or does he need adaptive seating?). It also includes oral-motor issues (what does he do w/ the food once it is in his mouth? can he chew? does he have a mature swallow? is he overly sensitive to texture?). With all these considerations in mind, a feeding/ eating program is developed and shared w/ all staff involved w/ eating routines w/ the student. Adaptive seating, adaptive utensils, and texture (w/ doctor and/or dietitian consultation) is considered and recommended if appropriate.

**Toothbrushing:** Evaluating a student's motor skill w/ the toothbrush as well as oral sensitivity. This includes making recommendations for adaptations (beginning with toothettes or washcloths or Nuk trainers instead of toothbrushes; allowing the child to sit rather than stand for stability; considering electric toothbrushes, etc.)

**Bed Making:** Evaluating motor-planning, balance and sequencing skills of the student to assist in developing a routine for bed-making. This includes consideration for adaptations to the routine (adding buttons to the bedding to help the child locate top of bed or sheet, etc)

**Bathing:** Evaluating motor-planning, sequencing, hand/arm skills, balance of the student to assist in developing a routine for increased independence in bathing. This includes consideration for any adaptive equipment (bath benches, hand-held showers, long-handled sponges, etc).

**Toileting:** Evaluating standing balance, coordination, clothing manipulation, hand/arm skills for wiping, etc to assist w/ establishing a routine for more independent toileting.

- **Adaptive Equipment:** OT/PT evaluates, recommends and maintains much of the adaptive equipment on campus (eating utensils, braces, hand splints, wheelchairs, canes, walkers, adaptive seating, bathing equipment, dressing adaptations, etc.)
- **Wheelchair Issues:** OT/PT problem-solves most wheelchair issues on campus including negotiating campus when in a wheelchair, modifications needed (one-arm drive, motorized, special seating...), transfer training for student and staff, toileting issues (hand-held urinal use, clothing manipulation, clothing adaptations such as Velcro closures). This includes consultation w/ doctors and/or funding sources and medical supply places as needed.
- **Job-site Evaluations/Modifications:** OT/PT can visit job sites and assist w/ evaluating and recommending adaptations for making a job task more successful. This can entail positioning issues, motor issues, adapting materials (or the task), setting up jigs, etc.
- **Serial Casting & Splinting:** Serial casting and splinting is undertaken only under a physician's referral. It is a process whereby a series of casts is applied for approx. 1 week, then removed and reapplied w/ the joint at a slightly greater angle of stretch than the previous cast. This is usually done for ankle stretching, but can also be done for knees and wrists. Serial casting is generally followed-up with a brace or splint to maintain the joint range gained through casting. The goal for serial casting is to improve

function (balance, coordination, or hand-skills). It is a time-consuming procedure which requires the participation and cooperation of the whole team, as the casts affect walking, bathing and all activities of ADL while they are in place.

- **Some Gross Motor concerns in CHARGE**

- **Balance** is affected by the inner ear mechanism as well as by strength and vision skills. The child w/ CHARGE has disturbances in the ear and vision systems. Inner ear issues can lead to balance problems. Balance needs to be evaluated by a doctor who specializes in inner ear or vestibular problems before a proper treatment regimen can be established.
- **Strength** may not necessarily be directly affected by CHARGE, but is affected by the medical issues and procedures the child is undergoing. It is also affected by the low tone that dominates the trunk and arms.
- **Vision Related** issues of balance: The child with CHARGE has visual field loss which affect his head position and posture. Many children w/ CHARGE have visual field loss which prevent them from seeing in their upper visual fields. They will hold their heads in a raised position in order to see things in front of them. This head position affects what they see (or do not see) near the ground, and leads to missing changes in the ground's surface and stumbling. Some children w/ CHARGE also have difficulties w/ eye dominance (inconsistently use left vs. right eye as the dominant eye). This can lead to a sense of the object moving (as they gaze at it and change eye dominance during the gaze) and also affects balance.



# PHYSICAL ACTIVITY, INCLUDING PHYSICAL EDUCATION, SPORT, AND RECREATION FOR CHILDREN WHO ARE DEAFBLIND

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## INTRODUCTION

### PHYSICAL ACTIVITY

Physical activity defined is any bodily movement produced by skeletal muscles that results in energy expenditure. Health-related fitness includes cardiorespiratory endurance, muscular strength and endurance, flexibility, and body composition (Caspersen, Powell, Christenson, 1985). The International Consensus Conference on Physical Activity Guidelines for Adolescents recommends that “all adolescents... be physically active daily, or nearly every day, as part of play, games, sports, work, transportation, recreation, physical education or planned exercise in the context of family, school, and community activities” (Sallis & Patrick, 1994). Patterns of health-related behaviors develop and become established during childhood and adolescence (Kelder, Perry, Klepp, & Lytle 1994), therefore, young people should be encouraged to become involved in physical activity. In addition, enhanced physical activity is a major component in weight management of obese children and adolescents (Bar-Or, et. al., 1998). According to the U.S. Department of Health and Human Services (1997), physical activity in schools should be promoted as part of a comprehensive school health program. Activities and services should be designed to promote the optimal physical, emotional, social, and educational development of students.

Physical activity is an umbrella term used to describe movement to significantly increase energy expenditure. Physical activity can be many things to many people. It includes physical education, active recreational activities, sports, and fitness activities. Individuals of all ages and abilities need to discover their preferred modes of physical activity.

### PHYSICAL EDUCATION

Physical education “when planned and taught properly is ‘education through the physical’. That is, the activity serves as a medium through which a total learning experience takes place” (Schmottlach & McManama, 1991 p. 1).

Under Public Law 105-17 the Individuals with Disabilities Education Act Amendments of 1997, Physical Education is a direct service under special education (IDEA Amendments of 1997). This means that physical education is **required** for every individual who has a disability. This law also means that physical education programs should be specially designed if necessary to meet unique needs. Under the law physical education consists of:

- a. physical and motor fitness;
- b. fundamental motor skills and patterns; and
- c. skills in aquatics, dance, and individual and group games and sports (including intramural and lifetime sports).

## **SPORT**

The meaning of sport varies according to the individual. Some individuals consider sport to be any physical competition between two or more people. Others consider sport to be any recreational activity such as fishing, canoeing, horseshoes, hunting, trapping or snowshoeing. Regardless of the meaning, it is important to honor the interests of all individuals. In addition, creating an environment in which the individual can be successful is critical. Use the recommendations in this section to ensure the most successful environment for each individual.

## **RECREATION**

Recreation is defined as: "Refreshment of ones mind or body after labor through diverting activity (Morris, 1978 p. 1090). Recreation gives us information about who we are (Haggard & Williams, 1992). It can take the place of inappropriate or self-stimulatory behavior (Honig, 1990). Lastly, it can reduce physical, social, and psychological isolation (Sauerburger, 1993).

## **BENEFITS OF PHYSICAL ACTIVITY**

Regular physical activity can promote health and reduce the risk for all-cause mortality and the development of many chronic diseases in adults (U.S. Department of Health and Human Services, 1997). For example, physical activity may reduce obesity (Greendale, Barrett-Connor, Edelstein, Ingles, & Haile, 1995), depression and anxiety, (Ross & Hayes, 1988; Stephens, 1988) and build bone mass density (Dalsky, Stocke, Ehsani, Slatopolsky, Lee, & Birge, 1995). In addition, it decreases blood pressure in adolescence with borderline hypertension (Alpert & Wilmore, 1994), increases physical fitness in obese children (Gutin, Cucuzzo, Islam, Smith & Stachura, 1996), and decreases the likelihood of obesity among children (Epstein, Valoski & Vara, 1995). According to Calfas & Taylor, (1994), physical activity among adolescents is consistently related to higher levels of self-esteem and self-concept and lower levels of anxiety and stress. In addition, the child with low physical activity levels has a higher risk for "hypertension, hypercholesterolemia, hyperinsulinemia, decreased release of growth hormone, respiratory disorders, and orthopedic problems". Also, "the obese child suffers both psychologically and socially. Self esteem and self image are often damaged by ridicule and scorn" (Bar-Or, et. al, 1998, p 2.)

American children are currently in a physical activity crisis (Kutzleman & Reiff, 1992; Sallis, 1993). The U.S. Surgeon General has determined that the Healthy People 2000 goal of 75% of vigorous activity for all young people has not been met. The goal of 30% for light to moderate physical activity for young people has also fallen short (US Department of Health and Human Services, 1996). The prevalence of overweight is at an all time high among children and adolescents (Nicklas, Webber, Johnson, Srinivasan, & Berenson, 1995; Sallis & Patrick, 1994).

## **CURRENT TRENDS**

National objectives for health promotion set forth by Healthy People 2000 (U.S. Department of Health and Human Services, Public Health Service, 1995) include seven initiatives, which relate directly to improving weight management in children. Three of these relate to the physical activity levels of children:

- a) increase to 30% those who engage regularly in light to moderate physical activity for at least 30 minutes a day:
- b) increase to 75% those children who engage in vigorous physical activity that promotes the development and maintenance of cardiorespiratory fitness three or more days per week for 20 minutes or more per occasion:

c) reduce to 15% those who engage in no leisure-time activity. Increasing the span of a healthy life most effectively and efficiently is the major objective of these initiatives.

### **Physical activity for individuals who are deafblind**

Unfortunately, research indicates that children who are blind tend to have more body fat, less cardiovascular endurance, muscular strength, and muscular endurance than their sighted peers (Lieberman & Carron, 1998; Winnick & Short, 1985). Winnick (1985) has also determined that children who are blind are behind in activities such as throwing, catching, balancing, striking, and body and spatial awareness. *Researchers attribute these various delays not to genetic limitations of performance, but rather to overprotection and discouraging attitudes on the part of the parents or teachers* (Winnick, 1985; Nixon, 1988). To date, research on the fitness and motor ability of individuals who are deafblind is limited yet, one can logically conclude that children who are deafblind will exhibit the same or more severe characteristics due to the additional communication and mobility needs.

It is imperative to encourage individuals who are deafblind to participate in physical activity. The physical, social and psychological benefits of physical activity will increase the likelihood of independence and improve the quality of life for these children. We need to encourage individuals who are deafblind to pursue fitness activities in their recreational time.

Some well-documented problems associated with deafblindness are communication, (Ford & Fredericks, 1995; Greenfield & Ford, 1997; McInnis & Treffry, 1993; Sauerberger, 1993; Sauerberger & Jones 1997; Smith, 1994; Stremmel & Schutz, 1995), mobility (Gee, Houghton, Pogrund, & Rosenburg, 1995; Gense & Gense, 1997; Sauerberger, 1993), isolation, (Enos, 1995; Sauergerber, 1993), and spatial awareness or environmental awareness (Newton & Schafer, 1997). Physical activity is a normal, enjoyable, and productive way to overcome each of these major barriers.

The area of communication is a basic component of physical activity. When an individual is involved in an activity, interest is conveyed through expressive communication, and feedback is expressed through receptive communication. Continued involvement in a variety of physical activities increases normal vocabulary, improves receptive and expressive communication, and allows the individual to understand basic concepts, which are a fabric of everyday life.

Example: Pierre is an 8th grade boy who was congenitally deafblind. He has an intervener and goes through his daily class schedule with her. For physical education, Pierre walked around the gymnasium because the teachers at his school did not think he could do anything else. One day Pierre was walking past the gymnasium and he felt a lot of stomping and cheering. He asked his intervener what was going on. She said "Oh it's just Judo." Pierre was curious "What is Judo?" She explained it, but he was not satisfied. Pierre joined his school's Judo team, learned new terms, made new friends, and recently competed in the 1996 Para Olympics in Atlanta, Georgia representing Canada.

Individuals who are deafblind face the challenge of mobility each day. Mobility is an area of emphasis in most books, conferences, and Individual Education Plans. Mobility for the sake of mobility can be frightening and difficult for many children. When individuals are involved in physical activity, mobility is functional, enjoyable, and can become easier with practice.

Example: Jake is an 11 year old who has Ushers Syndrome. He is losing his vision and has 10 degrees of vision left. Due to his decreasing vision, Jake is very fearful of movement. Jake's mobility instructor and his physical education teacher have decided to infuse many of his mobility concepts into his physical education class. In physical education, he works on running the bases, walking to and from the pool, swimming widths, lengths, and jumping into the pool,

and playing tag with his peers utilizing a peer tutor. He has overcome his fear of mobility, and enjoys movement again.

Many individuals who are deafblind experience isolation. This feeling is devastating especially for youth when their emphasis should be on socialization, and peer involvement. Many individuals who are deafblind even experience isolation when in a room full of people. In order to overcome isolation, individuals who are deafblind need to first develop a common interest with peers with a similar mode of communication to have something to share and participate in together. Physical activity and recreation can be an ongoing bond between a child who is deafblind, and her hearing and sighted or deaf and sighted peers.

Example: Monica is an eight-year-old girl with CHARGE Syndrome. She has some vision, is deaf, and can function independently in her activities of daily living. She uses total communication to communicate, and can be understood by most people who take the time to listen. She has recently entered the second grade and does not have any close relationships with her peers. She has never been to any other kids birthday parties and feels very isolated. Monica is a natural swimmer due to aquatic therapy following several operations. She joined the local YMCA's swim program and has made several new friends. Two friends have spent the night at her house and she was even invited to her first birthday party! She will go to the YMCA's swim camp this summer, and she may earn a spot on the junior varsity swim team.

Spatial awareness and awareness of the environment are concepts which can be difficult even if one is sighted and hearing. Children, born without adequate vision and hearing, will have a developmental delay in this area unless intervention is incorporated early on in development. When early intervention is incorporated early in life, children with deafblindness can appreciate and understand the world around them and increase the likelihood that they will be independent in most activities of daily living. This fact has been recognized and many early intervention programs have been implemented in recent years. There are a few books and articles which recognize early movement experiences as a key to increasing understanding of the world and independence (Haring & Romer, 1995; McInnis & Treffry, 1993).

Example: Jessica is a 2-year-old girl with congenital deafblindness due to prematurity. She has been crawling slowly and recently learned to pull herself up to a stand. Her grandmother had been her guardian, and had watched her every day at home. She was not aware of early intervention services. When the local 307.11 representative discovered her existence, she developed an early intervention program for her four mornings a week. The specialists in the program were trained on strategies that would work with Jessica. Jessica was stimulated to go through obstacle courses, given opportunities to jump on a rebounder, encouraged to feed and dress herself, encouraged to explore the diverse environment, involved in co-active movement, and given immediate positive feedback for her actions. Her grandmother and siblings were taught these strategies and they were also implemented at home. In three months, Jessica was walking, playing and asking for more!

## **DISCUSSION**

Options for physical activity Physical education curriculums differ from school to school. The following are many of the typical units found in physical education classes around the country.

### **Pre-school & Lower Elementary**

Body awareness	Hula hoops
Spatial awareness	Parachute
Locomotor skills	Simple tumbling
Manipulative	
Rhythms	



Object control	catching
kicking	Tag games
throwing (over and underhand)	Scooter boards
batting	Fitness games

### **Upper Elementary & Early Middle School Sports skills lead-up activities**

Tennis	Badminton
Softball	Bowling
Gymnastics	Golf
Soccer	Hiking/Backpacking
Flag football	Cross Country Skiing
Dance	Canoeing
Basketball	Rock climbing
Aerobics	Martial arts
Volleyball	Aquatics
Wrestling	

It is important to note that the activity should be age appropriate therefore, taught at the developmental level. The skills for the activities should be taught as the focus of the unit, and cooperative games and lead-up games should be played as opposed to the sport for a competitive purpose. It is important to focus on the child's skills in order to encourage enjoyment of physical activity. These sport concepts are later applied to competitive games and sports after they have mastered all the skill components.

## **Recreation**

### **Outdoor activities**

Canoeing	Horseshoes
Kayaking	Tether ball
Boating	Rollerblading
Camping	Rollerskating
Hiking/Backpacking	Skateboarding
Rock climbing	Frisbee
Fishing	Parks

### **Indoor activities**

Bowling	Darts
Ice-skating	Roller-skating
Board games	Theater
	Museums

### **Fitness**

Bicycling	Aerobics
Running	Aquatics
Fitness	Dance

## **OPPORTUNITIES FOR PHYSICAL ACTIVITY**

Key Points to remember when getting involved in physical activity involve individual strengths and abilities

### **A. Communication**

It is important to know what form of communication the student uses. As an example, if the child is participating in swimming, the instruction will vary depending on whether the child communicates by visual sign language, hand-over-hand signs or by pictures. Please see the communication chapters for more information on these systems.

Example: Mrs. Mason's beginning dance class. A student, Christine, is a 12-year-old girl who is deafblind. Mrs. Mason observes Christine in her art class at school. She sees Christine using signs and gestures as her receptive form of communication with her peers, and speech as her expressive language. Mrs. Mason then learned some basic signs for each dance step she was teaching, and took the time to listen to feedback from Christine. Because Christine uses sign for her receptive communication, Mrs. Mason understood that Christine had limited hearing. She then utilized strobe lights to set the beat for her beginning dance classes. Christine enjoyed her class so much she enrolled in the intermediate class with the same teacher!

### **B. Ambulation**

It is important that the teacher knows how the child ambulates. Important questions include the following: Does he/she use a wheelchair, a walker, a cane, a guide, or independently? What surfaces are best for him/her? Does he/she use multiple forms of ambulation such as a wheelchair to get places and a cane once he/she gets there? When developing physical activity programs, this is very important information to attain prior to instruction

Example: Jose is a student in Mr. Jones's 8th grade physical education class. Jose's class will be doing a soccer unit. Mr. Jones knows that Jose uses a wheelchair to get places, and usually crutches once he gets to the class. He is totally deaf, yet has some low vision. Mr. Jones gives Jose the option to practice and/or play in the chair or using his crutches. Jose tries different skills utilizing both methods and he decides to practice kicking using his crutches, but when it came to the game, he wanted to be in his chair because he moved much better. With a simple rule adaptation that all children had to touch the ball before they could score, Jose was an active part of the game.

### **C. Range of motion, motor skill and strength**

Determining how much range of motion, motor skill and strength your participant has is imperative in order for the instructor or program director to write individual goals and objectives for class and the IEP. This information can be collected using the Brockport Physical Fitness Test (Winnick & Short, 1998) for flexibility and strength and the Test of Gross Motor Development (Ulrich, 1985).

Example: Julie is a six-year-old girl who is deafblind as a result of prematurity. Her physical therapist and physical education teacher have collaborated to determine her areas of need that should be highlighted in the IEP. She has weakness in the areas of upper body strength, upper body flexibility, and locomotor skills. The therapist and teacher added these components to her IEP, and she worked on these areas daily with her physical education class, family, and therapist. Julie participated in activities such as an obstacle course, monkey bars, tumbling, wheelbarrow races, relay races, and parachute activities with peer tutors in each. At the age of seven Julie has improved her locomotor skills, and has upper body strength and flexibility very close to her same-age-peers.

## D. Independence

The degree of independence for individuals who are deafblind is often determined by how much previous exposure the individual has had in a given activity. Some children will be able to perform all activities independently, while some will need total assistance. Other factors that influence the level of independence include the amount of hearing and vision a child possesses, secondary disabilities, and the instructor's ability to demonstrate. It is imperative that the instructor determines the child's level of independence in order not to give too much or too little assistance in an activity. If an instructor starts an activity with too much assistance, she will not really know how independent the child is, and if the instructor starts out with too little assistance, the child may not reach any significant level of success, and may become frustrated or bored. The instructor can determine what level of assistance to use by task analyzing a skill into its component parts and asking the child to perform each part of the skill with little or no assistance. If the child has difficulty on one or more parts, the instructor may give the child some physical assistance in order to be able to perform all the parts, and decrease assistance as the child becomes more proficient at that skill.

Example: Robert is a 13-year-old boy in 6th grade. He is in summer camp and his camp counselor; Mr. Winnick wants to teach all the boys how to cast a fishing rod. Robert has been in camp before, but Mr. Winnick was not there and had no idea if he had experienced fishing. The other kids in the bunk did not remember how independent Robert was, but Robert said that he had fished before, but could not describe his level of independence. Mr. Winnick wanted Robert to be involved to the fullest extent possible, but did not want him to hurt himself or others, or become frustrated. Mr. Winnick task analyzed a cast and tested Robert to see what he could do. The following is the task analysis by used Mr. Winnick:

### Fishing Task Analysis

- \_\_\_\_\_ Bait the hook with a worm
- \_\_\_\_\_ Reel in the remaining line
- \_\_\_\_\_ Bring the rod back over the shoulder
- \_\_\_\_\_ Release and cast the rod forward
- \_\_\_\_\_ Lock the release on the reel

Mr. Winnick walked through each step with Robert to determine what he could do independently. When he finished the task analysis, it was apparent that Robert could bait the hook, reel in the line, bring the rod back over his shoulder and lock the release independently. He needed assistance with the timing of the casting and release of the reel. After some hand-over-hand assistance with the cast and release, Robert was able to perform the task independently. On his next fishing trip Robert caught two fish! Now, taking the fish off the line is a different story!

## IMPORTANT RULES

A few important rules of thumb to follow when developing and adapting activities for children or adults who are deaf-blind include the following:

- 1) Ecological Task Analysis (Davis & Burton, 1991). Ecological Task Analysis theory suggests that aspects of motor performance emerge from the constraints of the performer, the environment, and the task. Manipulating one or more of these three types of constraints will improve success for the individual. Participants should be afforded the opportunity to use a variety of equipment, and have choices about how to optimally perform the desired activity. This will allow the individual, when possible, to have some input on the type and extent of adaptations made.

- 2) Link movement to language (Van Dijk, 1966). Once the child knows the movement and what it is called, he/she has the potential to execute the skill independently.
- 3) Utilize Partial participation. Partial participation is better than no participation (Block, 1992). If a child needs moderate to total physical assistance to participate in an activity, that is better than a lack of participation. The person assisting can be a peer, a sibling, the teacher, or a volunteer.
- 4) The fewer adaptations the better. Always begin with the amount of assistance that will ensure desired performance and success (Lieberman, 1995).
- 5) Adaptations are not a one-time occurrence. Adaptations must be constantly monitored to determine whether further adaptations are necessary and to ensure success of the individual.
- 6) Utilize the name of the sport, skill or activity even if it is drastically modified from the original version. As an example, if you have made several adaptations to a game such as golf, with a different ball, a different club, and holes closer together, you can still call it golf. If we call activities different names because they look different than the original or "normal" version, then individuals who are deaf-blind will not have the satisfaction of knowing that they can really play golf. For example if the individual goes to a group home and is asked if he plays golf, he can say "Yes but I use this type of ball, and this type of club, and I will try to golf on your course". If we call golf something else he would say "no" and perhaps be excluded unnecessarily.

Example: Cory, a 17-year-old young Deaf woman with a visual impairment and cerebral palsy. She uses a wheelchair for ambulation, and has travel vision with corrective lenses. Cory lives in an environment which has long winters. Cory was introduced to cross-country skiing by her high school physical education teacher. She sat in a sled, and used cut-off poles for propulsion. Cory was given occasional directional cues by her teacher or a friend in order to cross-country ski successfully. When she moved to a group home, they offered cross-country skiing every week. Cory became involved because she knew she could ski with her minor adaptations. She now cross-country skis regularly with her friends and family.

- 7) Incorporate disability awareness simulation activities. Modify activities in a way which students without disabilities assume the impairment of the individual with a sensory impairment (Winnick, 1978).
- 8) Emphasize activities which force the participant to use their remaining hearing and sight. It is imperative that individuals with deafblindness learn to use their remaining senses to the best of their ability and any activity presented should encourage them to do so.
- 9) Emphasize activities which promote movement. Individuals who are deafblind are often reluctant to move. It is imperative that these individuals are encouraged to reach their physical potential and eventually govern their own movement experiences. For many individuals with deafblindness, this will not occur unless it is taught. Beginning activities which promote movement are:
  - a. Parachute swing: two people hold on to the parachute and swing the child inside the parachute.
  - b. Incline roll: place the child on top of a low incline and allow him/her to roll or crawl to a motivating sound source at the bottom.
  - c. Scooter pull: child sits on a scooter and allows the teacher to pull him/her around the gymnasium with a hula hoop. The child must hold on to one end of the hula hoop.
  - d. Scooter push: child sits on a scooter and pushes him/herself around cones or from one sound source to another. The child can also lie on his/her stomach and pull him/herself with their hands around the room.

- e. Therapy ball push: in an assisted sitting position, the child pushes a large heavy therapy ball to a sound source or a person. This builds upper body strength to assist in crawling and creeping.
- f. Rebounder heaven: the child jumps on a rebounder while holding the wall, the teacher, or a bar.
- g. Bell Balloon bash: Child chases a bell balloon around the room crawling, walking, or running guided if necessary and kicking it when he/she can (Lieberman & Cowart, 1996).

\* Material taken in part from :Lieberman, L.J. (1996). Adapting games, sports and recreation for children and adults who are deaf-blind. Deaf-Blind Perspectives, 3(3).

## **OPTIONS FOR INSTRUCTIONAL MODIFICATION:**

Explanation- explaining what you want the child to do in simple terms.

- should be done in the mode of communication the child understands
- should be repeated in a different way if the child does not understand the first time
- should be used with demonstration to ensure understanding if the child has any usable vision

Demonstration/Modeling- showing the child what you want him/her to do.

- should be done in the child's field of vision
- should be done by someone as close as possible to the child's size and ability
- should be done whole-part-whole when possible. This means that you should demonstrate the whole skill, then parts (task analysis), then the whole thing again.

Physical assistance/Guidance- assisting the child physically through the desired skill or movement.

- should be documented where you physically assisted, how much assistance was given, and for how long.
- should tell the child you are going to touch him/her before you physically assist
- should try to fade assistance to "normal" touch cues when possible

Brailing- allowing the learner to feel a peer or the instructor execute a skill or movement which was previously difficult to learn with the three previous approaches.(Lieberman & Cowart, 1996)

- should tell the child where and when to feel you or a peer executing a skill
- should document when and where the child touched you or a peer and why
- should be repeated as many times as necessary to ensure understanding
- should be combined with the other teaching methods to increase understanding

**Teaching Techniques**- These involve utilizing different teaching approaches depending upon the students learning strengths.

Command Style: This is when the teacher is in total control of all decisions.

Task Style: The teacher develops a series of task cards (may be brailled or in pictures) that progressively lead to the achievement of an instructional objective.

Guided Discovery: Students are encouraged to discover movement solutions that meet the criteria stated by the teacher. Using questions or short statements, the teacher guides the students in a progressive series of steps.

Problem Solving: Similar to Guided discovery style, the Problem Solving style emphasizes the development of multiple solutions to a given problem posed by the teacher. (Winnick, 1995)

Example: Mrs. Merritt is a horseback riding instructor and has taught at her farm for 18 years. Every year she gives eight weeks of lessons to the local church camp group. This year as part of her extended summer school recommendation Annette joined the church camp. Annette is

13 years old and has congenital Rubella. She has some balance difficulties, uses gestures and some signs, is totally deaf, and has some usable vision close up. In camp Annette used an intervener for most activities. The intervener, Mary, came along for the outing to interpret during the experience. Mrs. Merritt has taught in the same manner since she started (command style) and considers herself very successful. On the first day of class she began to teach using the methods she had been using for years. As a result, the girls quickly remembered the skills they had acquired in previous years. They all wore a velvet horseback riding helmet, stepped with the left leg, swung the right leg over, and rode with the reigns in their palms with thumbs up and sitting upright. When it was Annette's turn, she insisted on wearing her biking helmet, stepping with the right and swinging the left leg over, wearing the reigns around her wrist so she could sign, and leaning over hugging the horse's neck. Mrs. Merritt was very confused and frustrated with Annette's technique and attempted to change the way Annette rode. Her intervener Mary explained why she needed to ride this way and after great consideration, Mrs. Merritt decided that there were other ways to ride, and that Annette's way was best for her and it was fine. From that day on Mrs. Merritt gave more options in her lessons (guided discovery, problem solving) and allowed her students to explore what was best for them. Annette attends camp every summer and she now rides with her own helmet, sitting up, and with the reigns around her wrists.

### **Adaptations**

Many children who are deafblind will not be able to be successful in regular activities without some type of modification. Modifications may include changing the rules, equipment and environment.

Rules: A rule modification can be anything that deviates from the original or culturally acceptable rules of the game. Individuals who are deafblind may need the rules adapted in order to be successfully included.

Example: Adam is a 13-year-old boy in seventh grade. His physical education class is starting a hockey unit. Adam is deaf and has low vision with cerebral palsy and mild mental retardation. Adam, along with his physical therapist, mother, and physical education teacher, collectively determined what would make him most successful in this unit. They decided that he should start the unit with minor physical assistance and use a frisbee instead of a small puck. His class worked on skill development for two weeks, and Adam worked successfully with his peer tutor. When it came time for the game, Adam was allowed ten seconds to hit the frisbee. In addition, Adam was required to touch the frisbee before his team could score.

Equipment: Equipment modification is any modification that would make the participant more successful than using the pre-existing equipment. Individuals who are deafblind may need equipment adapted for a number of reasons such as assistance in grip, increased auditory awareness, limited range of motion, increased tactile cues, increased visual stimulation, increased independence, etc..

Example: Hanna is an 8-year-old girl. She is visually impaired, hard of hearing and has mild cerebral palsy in her legs. Hanna uses a wheelchair as her primary means of ambulation. Hanna can push her chair independently, yet is slow, and often has a hard time gripping her wheel rims. She uses signs, gestures, and speech as her primary means of communication. Hanna's classmates are learning signs and enjoy her participation in all activities. Hanna's physical education class is working on a parachute unit. Hanna's teacher was thrilled by this because one of Hanna's priorities on her IEP

was upper body and grip strength. In order for Hanna to fully participate in the parachute unit, her teacher made some modifications with the input of Hanna's mother, the teacher, and the physical therapist. The handle on the parachute was small for Hanna, so the teacher wrapped duct tape around it to increase the size. The teacher used a bright flashlight for Hanna to follow, when the light went up, she was to lift the parachute up, and when the light went down, she brought the chute down. When they played a ball game with the parachute the teacher had big, bright balls so she could track them in the game. In addition, Hanna had a trained peer tutor with her during class to assist in understanding, feedback, and physical assistance when needed. Hanna and her peers enjoyed the game so much, they asked to borrow the parachute at recess and successfully included Hanna in each game.

**Environment:** Environmental modifications may be needed in order to successfully include a child with deafblindness. Often times these modifications are not noticed until they become a problem. It is never too late to modify the environment to increase the success of an individual. In order to know what needs to be modified the instructor may ask the parent, previous teachers, or the participant himself. Environmental modifications include: decreasing distractions, increasing visual cues, limiting noise, changing lighting, increasing accessibility of the playing area, etc.

**Example:** Sal is nine years old and was recently diagnosed with Usher Syndrome. He is adjusting to his change in vision, but often finds certain things annoying or frustrating. He has been best friends with four friends in his neighborhood since he was young. They know the majority of signs and use gestures for communication. Before he was diagnosed with Ushers he played kickball every weekend at his friends house in the back yard. Sal found it more and more frustrating to kick their faded red ball on the grass, and run on the uneven grass to the shirt they laid down for a base. He was even more distracted in the outfield with the two birdhouses and kite in the trees, the houses, and the satellite dish in the neighbors yard. It was hard to distinguish between the ball, the bases and all the distractions in the background. Sal's friends noticed this and thought maybe they should stop playing kickball, but this thought made Sal even more frustrated. As a group they decided how Sal could participate more successfully. They moved their game to a cul-de-sac about 3 blocks away. The blacktop was smooth, and there were only green trees and one dark house in the background. Their parents bought them a yellow ball and four orange cones. They used chalk to draw a white line from home to first, second and third so Sal could run without confusion. They continued to play kickball and now even play baseball with the same yellow ball. Although Sal was losing more and more of his vision, his friends understood his needs and changed what was necessary to include him as much as possible.

### **Curriculum modification examples**

The tables at the end of this section are adapted units. Each unit has a title and four variables; equipment, instruction, environment, and rules. Listed under each variable are a variety of options from which the teacher can choose. The teacher determines what adaptations will enable the child to become most successful and checks all that apply. The adaptations should be documented and kept in the student's file for the parents, administrator, future teachers, and the IEP.

It is important to keep in mind that the order of teaching for successful learning and maximum enjoyment is skill development, cooperative games, and finally, once these two components are mastered, competition. In addition, adaptations, modifications, and variations are not a one-time occurrence. They need to be continually monitored to ensure the success of the child.

Tables are at the end of this section

### **Fitness modification examples (Lieberman & Taule, 1998)**

#### **Bicycling:**

- \* Independently: Individuals who have some usable vision may be able to ride a bicycle independently in a quiet park or around a track. It is always safer if there are peers or individuals with sight to ensure safety if this is the chosen mode of fitness.
- \* Tandem bicycles: Tandem bikes allow the sighted participant to ride in the front of the bike, while the participant who is deafblind rides in the back. The person in front is responsible for steering, peddling and stopping. The person in the back is responsible for peddling. Tandem bikes can be purchased through most bicycle stores and range from \$400-\$2,000. Be sure to try out several bikes before you purchase one. If you are not sure who to ride with, contact your local bicycling club, university, or deaf club. When riding be sure to develop specific signals for turning, stopping, or emergencies.
- \* Surrey or duo bikes: Surrey, or duo bikes are bikes where the participants ride side by side. The sighted participant is responsible for steering and stopping. This is more conducive to communication for individuals who are deafblind because the riders are side-by-side. The purchase of these bikes usually takes place through specialty bike stores. Your local bike store will give you directions about where to get a catalogue for these neat bikes.
- \* Stationary bicycles: These bicycles can be used independently by anyone who has some functional use of their legs. Many stationary bikes will read the distances traveled and amount of time ridden. These can be purchased for \$100-\$1,000 in most sporting good stores. Be sure that you have a way to record the distance traveled if this interests you. You may also wish to ride for a certain period of time.
- \* Bicycle stand: Bicycle stands can turn an ordinary ten -speed into a stationary bike. These are now similar to stationary bicycles and can be purchased for under \$100 from any sporting good stores.

#### **Jogging:**

- \* Sighted Guide: The runner grasps the guides elbow, shoulder or hand depending upon what is most comfortable for the runner and guide.
- \* Tether: The runner and guide grasp a tether, which is a short string, towel or shoelace. This allows the runner full range of motion of the arms, while remaining in close proximity to the sighted runner.
- \* Sound source: The guide rings bells or shakes a noisemaker for the runner to hear while they run side-by-side. This works best in areas with limited background noise and for runners with enough residual hearing.
- \* Guidewire: The runner holds onto a guidewire and runs independently for time or distance. Guidewires can be set up permanently, or temporarily. A guidewire is a rope or wire pulled tightly across a gymnasium, or track. A rope loop, metal ring, or metal handle ensures that the individual will not receive a rope burn and allows for optimal performance. The runner holds onto the sliding device and can run for as long as he or she wishes independently.
- \* Sound source from a distance: The runner runs to a sound source such as a clap or a bell. This can be done as a one time sprint or continued for a distance run.
- \* Sighted Guides shirt: The runner with partial vision runs behind a guide with a bright shirt. This must be done in areas where it is not too crowded. Be sure to pick a color which is recognized easily by the runner.
- \* Independently around a track: Runners with low vision can follow white lines on a track when the track is relatively empty. A dark track with bright lines should be used.
- \* Treadmill: Running on a treadmill provides a controlled and safe environment. Treadmills can be purchased at most sporting good stores. They will cost anywhere from \$200-over



\$1,000. Be sure to purchase a treadmill with a safety feature in case you want to stop quickly. Also, many will come with timers, or mileage readers. If you want this feature, be sure to tell the sales person.

- \* Wheelchair racing: An individual in a chair can use any of the above adaptations if necessary and appropriate. The individual can push him/herself in the chair for long distances to gain the same aerobic effect. Again, this can be around a track, their neighborhood, or on a paved path with assistance.

### **Fitness/Exercise Training**

This section will discuss the bulk of a fitness program, the exercise phase, specifically focusing on techniques for teaching individuals who are deafblind muscular strength and endurance training. The safest way to perform muscular strength and endurance training is to utilize a circuit of stationary machines. The machines may consist of a bench press, or a leg flexion-extension machines, a sit-up board, or a universal machine. The following are some strategies to introduce and instruct circuit training.

- \* Allow time for exploration: When introducing an individual to a machine allow time for tactile and/or visual exploration.
- \* Demonstration: The instructor should demonstrate the movement and link the movement to language, including the name of the exercise and muscle involved.
- \* Option to perform: The individual is encouraged to try everything yet, it is important that they understand that they can decline.

The following are some adaptations to the physical environment:

- \* Pictorials/Braille instructions: Allow time for the person to look at performance pictorials and/or the opportunity to read about it.
- \* Visual/Tactual perimeter: For safety, mark the perimeter of the exercise machines with rope or contrasting colored tape on the floor.
- \* Adaptations to equipment: Use large print, hi-mark and braille on/off switches. Vibrating timers can be worn around the neck or in the pocket.
  - Number stations: Use large print, hi-mark and braille each piece of equipment with a designated number. You could also have a rope, tape or tactile guide from one piece of equipment to the next one.

### **Aerobics:**

The term aerobics means literally “with oxygen” or the “steady state transport of oxygen to the working muscles” (Shephard, 1990 p. 5). The fitness activity called aerobics involves sustained physical activity to a point where the body is utilizing oxygen. This means that the body has to utilize 60%-80% of its maximum heart-rate over a period of time. To determine your working heart-rate, subtract your age from 220, then multiply that number by .60, .70, or .80 depending upon how hard you want to work (American College of Sports Medicine, 1991).

Example: Nancy, who is 32 wants to work at 60% of her maximum heart-rate. She would take  $220-32=188$ .  $188 \times .60 = 112.8$ . This means that Nancy’s working heart-rate is 110-120 beats per minute. To ensure that Nancy is working up to this level, she would sustain an activity such as aerobic dancing, cycling, or jogging in place for 5-10 minutes. While continuing this activity, she would take her pulse for 6 seconds, then add a 0 to her pulse score and compare that to 120. If she came up with 14, that translates to 140 heartbeats per minute and she should slow down a little. If she came up with 10, that translates to 100 beats per minute and she needs to step up the pace a little to maintain her desired heart-rate. Try to sustain the working heart rate for over 15 minutes, preferably 30 minutes. Remember to start out slowly.

Some examples of aerobic activities include:

- \* Step aerobics: This is sustained stepping on and off a 4, 6 or 8-inch high platform at varying tempos and in different directions. This type of activity is adaptable to any level of ability. If an individual cannot step onto a step he/she can do the same activities without a step.
- \* Low impact aerobics: sustained activity keeping one foot on the ground at all times. You can march with high knees, kick to the front, bring your knee up and clap under your leg, march in place and bring your arms up and down, do toe touches to the front, right and left, or just walk briskly around the room. As long as the individual is moving and keeping his/her heart rate up this activity can be executed successfully by anyone who is ambulatory.
- \* High Impact aerobics: This is sustained activity with both feet leaving the floor at some point during the movement. You can do jumping jacks, kicks to the front, jog in place, bring your knee up and clap under your leg with a jump with the other leg, pendulum leg swings out to the sides, side jumps and front jumps alternating directions, etc..... An individual obviously has to be in condition to sustain this activity for a long amount of time.
- \* Wheelchair aerobics: This is aerobics done in a wheelchair. The individual moves his/her arms up in the air, out to the sides, punches down, or twists at the hips for eight counts or more to elevate the heart rate. If the individual can move his/her legs they can move their legs at the same time as their arms. The idea is to increase the heart rate and amount of energy expenditure. Any amount of movement can elevate the heart and if this is continued for over 5 minutes it is considered aerobic. Make sure the point is to elevate the heart rate and have fun!
- \* Physical assistance and/or Braille (Lieberman & Cowart, 1996): This can be used when the individual does not have enough vision and/or hearing to understand the movement. The physical assistance and braille needs to be explained so the participant will know what to do. The instructor can then simplify all the moves to one touch cue or a sign cue that the participant will understand. An example would be if the instructor wants the participant to march in place as part of a low impact aerobics routine. Once the concept is understood the instructor does the sign for soldier, or taps the individual's knee to signal marching. The participant now knows he will march for eight counts then a new cue will be given for the next move.
- \* Others: Any activity which brings your heart-rate up for a sustained period of time is considered an aerobic activity. Cycling, running, swimming, walking, or aerobics can accomplish this.

The instructor can also set up routines so that one eight count move is always followed by the next eight count move and so on. This will depend upon the ability and level of condition of the participant. Once the moves are understood the instructor should try to fade out the touch cues for physical assistance and braille to promote independence.

**\*\*Note** these activities can be enjoyed with or without music. It is much easier to practice them without music first, then add that variable when you so desire.

**Aquatics**: Swimming is one of the best activities for individuals who are deafblind. There are few barriers, and the swimmer can move freely without worrying too much about obstacles. Water can aid in range of motion, balance, stability, locomotion, and socialization. A few adaptations for the aquatics area include:

- \* Flotation devices: Utilize a variety of flotation devices when needed. An individual can still receive an aerobic workout with a flotation device. If the individual is afraid to swim in the deep end without a flotation device, that is fine. He/she can swim laps with the flotation

device if this makes them more comfortable. Kickboards are helpful because the board hits the side of the pool before your head does!

- \* Trailing: Use the wall and lane lines as guides for lap swimming the length of the pool.
- \* Distance traveled: Use some type of counter devices such as flip cards, counters, or rings to assist in understanding of distance traveled or number of laps.
- \* Improved skills: The pool is a wonderful medium for teaching locomotor and object control skills because water provides full-body support and balance is naturally enhanced.
- \* Beginner swimmers: Utilize a lot of hand-over-hand teaching and braille for the instruction of beginners (Lieberman & Cowart, 1996)
- \* Tread water: For individuals who do not feel comfortable swimming laps, treading water is a good aerobic workout and you don't have to worry about bumping your head on the wall of the pool!

**Dance**: Dancing is an opportunity for free movement and exercise as well as a chance to be involved in a crowd, bumping bodies, moving, holding hands, and socializing (Smith, 1994).

- \* Decibels: Play music very loud and turn up the bass. Be sure to inform those with hearing aids before you turn the music up so they can adjust the setting if necessary.
- \* Beats of music: Utilize strobe lights that reflect the beat of the music. You could also have participants hold a balloon which can pick-up the vibrations from the music (Smith, 1994).
- \* Wooden floor: Dance on a wooden floor to increase vibrations and tactile cues.
- \* Peer tutors: Utilize peer tutors to give modeling, and physical assistance (Lieberman & Cowart, 1996; Houston-Wilson, Lieberman, Horton, Kasser, 1997). Deaf peer tutors are also excellent role models!

## **SUMMARY**

The benefits of physical activity have been well documented, yet many children who are deafblind are excluded from participation. In addition, the success or failure of the individual in physical fitness, or aquatics is largely dependent upon the attitude of the physical educator, teacher, parent, support staff, and individual him/herself (Downs & Williams, 1994; Rizzo & Kirkendall, 1995). It is important to remember that individuals who are deafblind can achieve the same benefits and success as their hearing and sighted peers as long as those working with these individuals creatively adapt the activities in order to meet each individual's unique needs.

- \* The previous material was taken directly from: Lieberman, L.J., & Taule, J. (1998). Including physical fitness into the lives of individuals who are deafblind, Deafblind Perspectives, 5(2), 6-10.

## INCLUDING PHYSICAL EDUCATION ON THE INDIVIDUAL EDUCATION PLAN (IEP)

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Physical education is found in Public Law 105-17, the Individuals with Disabilities Education Act (IDEA) under special education. Specifically, in the law physical education covers fundamental motor skills, physical and motor fitness, play, games and sport activities, rhythms, dance, aquatics, leisure and recreation, and community based activities. Because physical education is listed in the law it is a **direct service**. This means that it must be included in every child's schedule and on every IEP. Many physical educators and teachers do not include physical education on the IEP because they do not know how to test the student who is deafblind. There are no specific tests to evaluate a child who is deafblind. This minor problem can be overcome by following the principle of the three P's.

### ASSESSING FOR PHYSICAL EDUCATION: THE 3 P'S

One way to use authentic assessment data in the development of IEP's is to follow the procedure known as the 3 P's. This procedure has the teacher analyze the *Process* of the movement, the *Product* or outcome of the performance and the *Parameters* under which the performance is exhibited. The following provides a description as well as an example of each term.

#### Process

Process information relates to the form or quality of a movement. Skills can be analyzed into component parts through task analysis or through the use of ready-made checklists. Fronske (1997) provides a variety of task analyzed sport skills which teachers can utilize in authentic assessment. For example, Fronske (1997) task analyzes the underhand volleyball serve as follows:

##### Underhand Volleyball Serve

- \_\_\_step toward net with opposite foot of serving arm
- \_\_\_palm up making fist with serving hand
- \_\_\_weight shift forward
- \_\_\_contact ball in front of body
- \_\_\_follow through

This example could be used as authentic assessment data on the ability of students to master the underhand serve. Furthermore, it could be used to develop IEP goals and objectives for students with disabilities in inclusive classes without additional expenditure of time as all students would be assessed on this particular skill

### **Product**

The product of the skill relates to the quantitative value produced by the student's performance. Skills will be quantified differently depending upon the desired outcome, age of the student and goals of the class. Using the underhand volleyball serve, the product could be quantified by:

1. the number of components of the skill the student was able to master;
2. the number of times the student served the ball;
3. the number of times the ball went over the net or (4) the number of times the ball lands in a pre-determined placement.

### **Parameter**

Parameter refers to the conditions under which the skill was performed. Examples of parameters which could be examined include: (1) type of equipment used; (2) distance from where the skill is executed; (3) environmental arrangement including indoors, outdoors, group or 1:1 setting; and (4) levels of assistance (independent, verbal cue, demonstration, physical assistance) if required. Again, using the underhand volleyball serve, the equipment parameters, which could be chosen, include a regulation volleyball, a volleyball trainer or a beach ball. The distance parameter may include the distance served from the net. The environmental parameter in which the serve takes place could be either indoors or outdoors, during a game situation or drill situation, or in small or large groups. Lastly, the level of assistance required will depend on the individual student's functional level. Students may require verbal cues, demonstration or physical assistance. Assistance may come from a peer, teachers aid or the teacher directly.

Taken in part from: Houston-Wilson, C., & Lieberman, L.J. (in press). Becoming involved in the Individualized Education Plan process: A guide for regular physical educators. Journal of Physical Education Recreation & Dance.

## **IMPORTANCE OF BECOMING INVOLVED IN THE IEP PROCESS**

Every child needs stimulation in all behavioral domains for healthy development. It is imperative that the psychomotor domain be represented on the IEP. By documenting in a measurable way what the child can do, the teacher is diagnosing strengths and weaknesses. This information is imperative in future programming and goal setting for the child and their family. In addition, by documenting what the child can do in the psychomotor domain, the school, physical educator, parent, and administrators are committing to improving weaknesses which will eventually assist in a better quality of life. A teacher who documents progress and learning is held accountable for her teaching, and will benefit in the future by knowing that she made a difference, no matter how big or small.

Many children with deafblindness do not have physical education on the IEP. The following is a checklist to assist in IEP involvement:

Checklist for IEP involvement:

- ☐ Assess the current IEP for any information which may help you
- ☐ Determine who would know about the child's psychomotor abilities (parents, teachers interveners)
- ☐ Test the child on at least two areas and document present level of performance (utilizing the 3 P's)
- ☐ Document strength and weak areas in positive terms
- ☐ Develop short term objectives
- ☐ Meet with the multidisciplinary team to ensure common goals
- ☐ Attend the IEP meeting when possible
- ☐ Re-evaluate often to ensure goals and objectives are being met

## **EQUIPMENT COMPANIES:**

**Access to Recreation:-** Adaptive Recreation Equipment for the Physically Challenged.

1-800-634-4351- to order catalogue

1-805-498-8186-fax

**Community Play Things:-** Safe and adapted playground equipment for young children.

1-800-777-4244-customer service

1-914-658-8065-fax

**Jesana Ltd.-** Adapted communication, recreation, play, and game equipment specifically for individuals with more involved disabilities. (walkers, bikes, tricycles, prone standards, etc.)

1-800-443-4728-to order catalogue

1-914-376-0021-fax

**Project Adventure Equipment Catalogue-** A source book for ropes courses and adventure programs.

1-508-524-4556 in MA or 1-770-784-9310 in GA

**Sporttime/Abilitations-** Sports, recreation, and games equipment for all individuals including those with sensory impairments and physical disabilities. (varieties of textures, shapes and colors)

1-800-850-8602- to order catalogue

1-800-845-1535 fax

**S&S World Wide Games-** Sports equipment for every occasion

1-800-243-9232- to order catalogue

1-800-566-6678- fax

**Snitz-** Sports, recreation and games adapted equipment for every occasion.

1-800-558-2224-to order catalogue

1-800-432-2842-fax

**Team Rehab-** Innovative assistive technology to increase independence for individuals with involved disabilities.

1-310-317-4522- for catalogue and journal

1-310-317-9644- fax

**Things From Bell-** A variety of sports, recreation and game equipment in many colors, shapes and textures.

1-800-642-7337- to order catalogue

1-800-543-1458- for orders only

1-800-432-2842-fax

**Toledo-** Physical Education supply company with inexpensive sports and playground equipment.

1-800-225-7749-to order catalogue

**Wolverine Sports-** sports, gymnastics, & coaching equipment.

1-800-521-2832-to order catalogue

1-800-654-4321-fax

**Vital Signs-** Instrumentation for rehabilitation, sports medicine, and physical fitness

1-608-735-4718-to order catalogue

1-608-735-4859-fax

## **REFERENCES**

- Alpert, B.S. & Wilmore, J.H. (1994). Physical activity and blood pressure in adolescents. Pediatric Exercise Science, 6, 361-380.
- American College of Sports Medicine (1991). Guidelines for Exercise Testing and Prescription. (4th ed.) Philadelphia, PA: Lea & Febiger Publishers.
- Bar-Or, O., Foreyt, J., Bouchard, C., Brownell, K.D., Dietz, W.H., Ravussin, E., Salbe, A.D., Schwenger, S., St Jeor, S., & Torun, B. (1998). Physical activity, genetic, and nutritional considerations in childhood weight management. American College of Sports Medicine, 31(1), 2-10.
- Block, M.E. (1992). What is appropriate physical education for students with profound disabilities? Adapted Physical Activity Quarterly, 59, 48-52.
- Calfas, K.J. & Taylor, W.C. (1994). Effects of physical activity on psychological variables in adolescents. Pediatric Exercise Science, 6, 406-423.
- Caspersen, C.J., Powell, K.E., & Christenson, G.M. ((1985). Physical activity, exercise and fitness: definitions and distinctions for health-related research. Public Health Report, 100(2), 126-131.
- Dalsky, G.P., Stocke, K.S., Ehsani, A.A., Slatopolsky, E., Lee, W.C. & Birge, S.J. (1988). Weight-bearing exercise training and lumbar bone mineral content in post menopause women. Annals of Internal Medicine, 108, 824-828.
- Davis, W.E., & Burton, A.W. (1991). Ecological task analysis: Translating movement behavior theory into practice. Adapted Physical Activity Quarterly, 8, 154-177.
- Downs, P., & Williams, T.(1994). Student attitudes toward integration of people with disabilities in activity settings. Adapted Physical Activity Quarterly, 11, 32-43.
- Enos, J. (1995). Building relationships with friends and other community members. In J.M. Everson, Supporting young adults who are deaf-blind in their communities. Baltimore, MD: Paul H. Brookes Publishing.
- Epstein, L.H., Valoski, A.M. & Vara, L.S. (1995). Effects of decreasing sedentary behavior and increasing activity on weight change in obese children. Health Psychology, 14(2), 109-115.
- Ford, J. & Fredericks, B. (1995). Using interpreter-tutors in school programs for students who are deafblind. Journal of Visual Impairments, 89(3), 229-234.
- Fronske, H. (1997). Teaching cues for sports skills. Needham Heights, MA: Allyn and Bacon Publishers.
- Gense, J.D.& Gense, M. (1997). Making it happen: functional ideas for orientation and mobility instruction for students with multi-impairments and/or deafblindness. Paper presented at the National Conference on Deafblindness, Washington, DC, The Individual in a Changing Society, June 6-9.
- Greendale, G.A., Barrett-Connor, E., Edelstein, S. Ingles, S. & Haile, R. (1995). Lifetime leisure exercise and osteoporosis: the Rancho Bernardo Study. American Journal of Epidemiology, 141(10), 951-959.
- Greenfield, R.& Ford, J.(1997). Designing effective communication systems for children who are deafblind. Paper presented at the National Conference on Deafblindness, Washington, DC, The Individual in a Changing Society, June 6-9.
- Gutin, B., Cucuzzo, N., Islam, S., Smith, C., & Stachura, M.E. (1996). Physical training, lifestyle education, and coronary risk factors in obese girls. Medicine Science in Sport and Exercise, 28(1), 19-23.
- Haggard, L.M., & Williams, D.R. (1992). Identity affirmation through leisure activities: Leisure symbols of the self. Journal of Leisure Research, 24, 1-18.
- Honig, B. (1990). Program guidelines for individuals who are deafblind. Sacramento: California Department of Education.

- Houston-Wilson, C., Lieberman, L.J., Horton, M. Kasser, S. (1997). Peer tutoring: A plan for instructing students of all abilities. Journal of Physical Education, Recreation, & Dance, 6, 39-44.
- Houston-Wilson, C. & Lieberman, L.J. (in press). Becoming Involved in the Individualized Education Plan Process: A Guide for Regular Physical Educators. Journal of Physical Education, Recreation & Dance.
- Individuals with Disabilities Education Act of 1997 (IDEA), PL 105-17. (January 7, 1997). Individuals with Disabilities Education Act Amendments of 1997. Washington D.C.: U.S. Government Printing Office.
- Kelder, S.H., Perry C.L., Klepp, K.I., & Lytle, L.L. (1994). Longitudinal tracking of adolescent smoking, physical activity and food choice behaviors. American Journal of Public Health, 84(7), 1121-1126.
- Kuntzleman, C.T., & Reiff, G.G. (1992). The decline in American children's fitness levels. Research Quarterly in Exercise and Sport, 63, 107-111.
- Lieberman, L.J. (1995). Recreation and leisure. Deaf-Blind Perspectives, 2, 10-13.
- Lieberman, L.J. (1996). Adapting games, sports and recreation for children and adults who are deafblind. Deaf-Blind Perspectives, 3, 5-8.
- Lieberman, L.J. (1997). Fundamentals for developing successful recreation programs. Canadian Institute for the Blind's Conference for Teachers of the Visually Impaired. October, 1997, Kelowna, Canada.
- Lieberman, L.J. & Carron, M.F. (1998). Health Related Fitness of Children who are Visually Impaired. Research Presented at the National American Association for Health, Physical Education, Recreation & Dance Conference, Reno, Nevada.
- Lieberman, L.J. & Cowart, J.F. (1996). Games for people with sensory impairments. Champaign, IL. Human Kinetics Publishers.
- Lieberman, L.J. & Downs, S.B. (1995). Physical education for students who are deaf-blind: A tutorial. Brazilian International Journal of Adapted Physical Education, 2, 125-143.
- Lieberman, L.J., & Taule, J. (1998). Including physical fitness into the lives of individuals who are deafblind, Deafblind Perspectives, 5(2), 6-10.
- McInnis, J.M. & Treffry, J.A. (1993). Deaf-Blind infants and children. Toronto: University of Toronto Press.
- Morris, W. (ed.) (1978). The American Heritage Dictionary of the English language. Boston, MA: Houghton Mifflin Company, pp. 1090.
- Newton, G. & Shafer, S. (1997). Active learning by means of the little room: An adaptation/sharing of Lilli Nielsen's work with environmental intervention with young children with deafblindness. Paper presented at the National Conference on Deafblindness, Washington, DC, The Individual in a Changing Society, June 6-9.
- Nicklas, T.A., Webber, L.S. Johnson, C.C., Srinivasan, S.R. & Berenson, G.S. (1995). Foundations for health promotion with youth: a review of observations from the Bogalusa Heart Study. Journal of Health Education, 26(2 supplement), S18-S26.
- Ross, C.E. & Hayes, D. (1988). Exercise and psychologic well-being in the community. American Journal of Epidemiology, 127(4), 762-771.
- Rizzo, T.L. & Kirkendall, D.R. (1995). Teaching students with mild disabilities: What effects attitudes of future physical educators? Adapted Physical Activity Quarterly, 12, 205-216.
- Sallis, J.F. (1993). Epidemiology of physical activity and fitness in children and adolescents. Critical Reviews in Food Science and Nutrition, 33, 403-408.
- Sallis, J.F. & Patrick, K. (1994). Physical activity guidelines foadolescents: consensus statement. Pediatric Exercise Science, 6, 302-314.
- Schmottlach, N. & McManama, J. (1997). The physical education handbook (9th edition). Needham Heights, MA: Simon & Schuster Company.



- Shephard, R.J. (1990). Fitness in Special Populations. Champaign, IL: Human Kinetics Publishers.
- Smith, T.B. (1994). Guidelines: Practical tips for working and socializing with deaf-blind people. Burtonsville, MD: Sign media Inc.
- Stephens, T. (1988). Physical activity and mental in the United States and Canada: evidence from four population surveys. Preventive Medicine, 17, 35-47.
- Sauerberger, D. (1993). Independence Without Sight or Sound. New York, NY: American Foundation for the Blind.
- Sauerberger, D. & Jones, S. (1997). Communication of deafblind people with the public. Paper presented at the National Conference on Deafblindness, Washington, DC, The Individual in a Changing Society, June 6-9.
- Smith, T.B. (1994). Guidelines: Practical Tips for Working and Socializing with Deaf-Blind People. Burtonsville, MD: Sign Media Inc.
- Stremel, K., & Schutz, R. (1995). Functional communication in inclusive settings for students who are deaf-blind. In N.G. Haring, & L.T. Romer, Welcoming Students who are Deaf-Blind into Typical Classrooms, Baltimore, MD: Paul H. Brookes.
- Ulrich, D.A. (1985). The Test of Gross Motor Development. Austin, TX: Pro ED Publishers.
- U.S. Department of Health and Human Services, Public Health Service. (1995). Healthy people 2000: Midcourse review and 1995 revisions. Maryland: National Center for Health Statistics.
- U.S. Department of Health and Human Services. (1996). Physical activity and health: A report of the surgeon general. Atlanta, GA: U.S. Department of Health and Human Services, Centers for Disease Control and Prevention, National Center for Chronic Disease Prevention and Health Promotion.
- Van Dijk, J. (1966). The first steps of the deaf-blind child towards language. International Journal for the Education of the Blind, 15(1), 112-115.
- Winnick, J.P. (1985). Performance of visually impaired youngsters in physical education activities: Implications for mainstreaming. Adapted Physical Activity Quarterly, 3, 58-66.
- Winnick, J.P., & Short, F.X. (1985). Physical fitness testing for the disabled: Project UNIQUE. Champaign, IL: Human Kinetics.
- Winnick, J.P. & Short, F.X. (1998). The Brockport Health Related Physical Fitness Test. Champaign, IL: Human Kinetics Publishers.



## ELEMENTARY PHYSICAL ACTIVITY EXAMPLES

### Jump rope

<u>Equipment</u>	<u>Rules</u>	<u>Environment</u>	<u>Instruction</u>
<input type="checkbox"/> Light rope	<input type="checkbox"/> Roll over rope	<input type="checkbox"/> Lay rope on floor	<input type="checkbox"/> Physical assistance
<input type="checkbox"/> Heavier rope	<input type="checkbox"/> Crawl over rope	<input type="checkbox"/> Student holds one side of rope	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Shorter rope	<input type="checkbox"/> Step over rope	<input type="checkbox"/> Student holds both sides of rope	<input type="checkbox"/> Peer holds one side
<input type="checkbox"/> Longer rope	<input type="checkbox"/> Leap over rope	<input type="checkbox"/> Student in wheelchair	<input type="checkbox"/> 1:1
<input type="checkbox"/> Velcro glove	<input type="checkbox"/> Jump forward over rope	<input type="checkbox"/> Student rolls over rope while holding rope	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Bells on rope	<input type="checkbox"/> Jump sideways over rope	<input type="checkbox"/> Use music	<input type="checkbox"/> Slow down
<input type="checkbox"/> Bright rope	<input type="checkbox"/> Swing rope and step over	<input type="checkbox"/> No music	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Handled rope	<input type="checkbox"/> Swing rope and jump over	<input type="checkbox"/> Limit stimulation	<input type="checkbox"/> Feedback
<input type="checkbox"/> Cloth rope	<input type="checkbox"/> Swing rope and jump consecutively	<input type="checkbox"/> Increase stimulation	<input type="checkbox"/> Braille
<input type="checkbox"/> Plastic Rope	<input type="checkbox"/> Swing rope and jump double		<input type="checkbox"/> Homework
<input type="checkbox"/> No rope	<input type="checkbox"/> Swing rope and jump crossed in front		<input type="checkbox"/> Guided Discovery
<input type="checkbox"/> Rope cut 1/2	<input type="checkbox"/> Swing rope and jump back & double back		<input type="checkbox"/> Command Style

### Spatial Awareness

<u>Equipment</u>	<u>Rules</u>	<u>Environment</u>	<u>Instruction</u>
<input type="checkbox"/> Mats	<input type="checkbox"/> Personal space	<input type="checkbox"/> Guided discovery	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Beanbags	<input type="checkbox"/> Control speed	<input type="checkbox"/> Visual boundaries	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Balls	<input type="checkbox"/> Right	<input type="checkbox"/> Sequence centers	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Ropes	<input type="checkbox"/> Left	<input type="checkbox"/> Play stations	<input type="checkbox"/> Verbal command
<input type="checkbox"/> Tunnels	<input type="checkbox"/> Up	<input type="checkbox"/> Give and get	<input type="checkbox"/> Braille
<input type="checkbox"/> Hoops	<input type="checkbox"/> Down	<input type="checkbox"/> Exploration	<input type="checkbox"/> Stations/Tasks
<input type="checkbox"/> Scooters		<input type="checkbox"/> Obstacle	<input type="checkbox"/> Demonstration
<input type="checkbox"/> Tires		<input type="checkbox"/> Course	<input type="checkbox"/> Explanation
<input type="checkbox"/> Yarn balls			<input type="checkbox"/> Guided Discovery
<input type="checkbox"/> Balloons			<input type="checkbox"/> Problem Solving
			<input type="checkbox"/> Tactile Discovery
			<input type="checkbox"/> Intervener Assistance

## Laterality and Verticality

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Mats	<input type="checkbox"/> Up hill	<input type="checkbox"/> Play stations	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Cones	<input type="checkbox"/> Down hill	<input type="checkbox"/> Visual boundaries	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Ropes	<input type="checkbox"/> Fast	<input type="checkbox"/> Stations	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Stairs	<input type="checkbox"/> Slow	<input type="checkbox"/> Small space	<input type="checkbox"/> Brailleing
<input type="checkbox"/> Tunnels	<input type="checkbox"/> Distance	<input type="checkbox"/> Give and get	<input type="checkbox"/> Verbal command
<input type="checkbox"/> Hoops	<input type="checkbox"/> Obstacles	<input type="checkbox"/> Exploration	<input type="checkbox"/> Stations
<input type="checkbox"/> Scooters	<input type="checkbox"/> Right	<input type="checkbox"/> Smooth surface	<input type="checkbox"/> Enlarged task cards
	<input type="checkbox"/> Left		<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Tires		<input type="checkbox"/> Textured surface	<input type="checkbox"/> Guided Discovery
<input type="checkbox"/> Ramps		<input type="checkbox"/> Obstacle courses	<input type="checkbox"/> Problem Solving
<input type="checkbox"/> Rhythm instruments			<input type="checkbox"/> Tactile
<input type="checkbox"/> Foot prints			<input type="checkbox"/> Discovery
<input type="checkbox"/> Climbing ropes			<input type="checkbox"/> Intervener
<input type="checkbox"/> Balls			
<input type="checkbox"/> Bridges			

## Balance

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Balance beam	<input type="checkbox"/> Holding objects	<input type="checkbox"/> Play stations	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> High beam	<input type="checkbox"/> Forward	<input type="checkbox"/> Sequence centers	<input type="checkbox"/> Verbal command
<input type="checkbox"/> Low beam	<input type="checkbox"/> Backwards	<input type="checkbox"/> Give and get	<input type="checkbox"/> Brailleing
<input type="checkbox"/> Narrow beam	<input type="checkbox"/> Dodging	<input type="checkbox"/> Stations	<input type="checkbox"/> Indirect
<input type="checkbox"/> Wide beam	<input type="checkbox"/> One foot	<input type="checkbox"/> Boundaries	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Balance board	<input type="checkbox"/> Two foot	<input type="checkbox"/> Small space	<input type="checkbox"/> Enlarged task cards
<input type="checkbox"/> Bean bags	<input type="checkbox"/> Hands free	<input type="checkbox"/> Large space	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Jump rope	<input type="checkbox"/> Sideways	<input type="checkbox"/> Obstacle	<input type="checkbox"/> Guided Discovery
<input type="checkbox"/> Mat	<input type="checkbox"/> Step over		<input type="checkbox"/> Task Style
<input type="checkbox"/> Ladder	<input type="checkbox"/> Step through		<input type="checkbox"/> Problem Solving
<input type="checkbox"/> Chair	<input type="checkbox"/> Step in		
<input type="checkbox"/> Tilt boards	<input type="checkbox"/> Step out		
<input type="checkbox"/> Bench	<input type="checkbox"/> Moving		
<input type="checkbox"/> Lines	<input type="checkbox"/> Stations		
<input type="checkbox"/> Rebounder			
<input type="checkbox"/> Steps			
<input type="checkbox"/> Tires			
<input type="checkbox"/> Hula Hoops			

## Locomotor skills

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Cones	<input type="checkbox"/> Fast	<input type="checkbox"/> Boundaries/Tactile	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Scooters	<input type="checkbox"/> Slow	<input type="checkbox"/> Small space	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Foot prints	<input type="checkbox"/> Directions	<input type="checkbox"/> Large space	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Dots	<input type="checkbox"/> Rhythm	<input type="checkbox"/> Cones	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Ropes	<input type="checkbox"/> Holding objects	<input type="checkbox"/> Secluded	<input type="checkbox"/> Feedback
<input type="checkbox"/> Hoops	<input type="checkbox"/> Left	<input type="checkbox"/> Use music	<input type="checkbox"/> Brailleing
<input type="checkbox"/> Tires	<input type="checkbox"/> Right	<input type="checkbox"/> No music	<input type="checkbox"/> Verbal command
<input type="checkbox"/> Mats	<input type="checkbox"/> Up	<input type="checkbox"/> Play stations	<input type="checkbox"/> Indirect
<input type="checkbox"/> Tunnel	<input type="checkbox"/> Down	<input type="checkbox"/> Sequence centers	<input type="checkbox"/> Small group
<input type="checkbox"/> Stairs	<input type="checkbox"/> Low	<input type="checkbox"/> Give and gets	<input type="checkbox"/> Problem Solving
<input type="checkbox"/> Sound source	<input type="checkbox"/> High	<input type="checkbox"/> Bright Lines	<input type="checkbox"/> Guided discovery
<input type="checkbox"/> Video	<input type="checkbox"/> Down		

## Object Control Skills

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Large balls	<input type="checkbox"/> Varied distance	<input type="checkbox"/> Non stimulatory	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Small balls	<input type="checkbox"/> Against a wall	<input type="checkbox"/> Boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Auditory balls	<input type="checkbox"/> One hand	<input type="checkbox"/> Small space	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Yarn balls	<input type="checkbox"/> Two hand	<input type="checkbox"/> Large space	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Wiffle balls	<input type="checkbox"/> Dominate hand	<input type="checkbox"/> Cones	<input type="checkbox"/> Feedback
<input type="checkbox"/> Tactile balls	<input type="checkbox"/> Nondom hand	<input type="checkbox"/> Secluded	<input type="checkbox"/> Brailleing
<input type="checkbox"/> Heavy balls	<input type="checkbox"/> Dominate foot	<input type="checkbox"/> Use music	<input type="checkbox"/> Verbal command
<input type="checkbox"/> Light balls	<input type="checkbox"/> Non dominate foot	<input type="checkbox"/> No music	<input type="checkbox"/> Indirect
<input type="checkbox"/> Bright balls	<input type="checkbox"/> Underhand	<input type="checkbox"/> Play stations	<input type="checkbox"/> Small group
<input type="checkbox"/> Ball on a rope	<input type="checkbox"/> Overhand	<input type="checkbox"/> Sequence centers	<input type="checkbox"/> Problem Solving
<input type="checkbox"/> Hockey puck	<input type="checkbox"/> Sidearm	<input type="checkbox"/> Give and gets	<input type="checkbox"/> Guided Discovery
<input type="checkbox"/> Rackets	<input type="checkbox"/> Stationary		
<input type="checkbox"/> Bats	<input type="checkbox"/> Traveling		
<input type="checkbox"/> Paddles			
<input type="checkbox"/> Tee			
<input type="checkbox"/> Hockey stick			
<input type="checkbox"/> Bean bags			
<input type="checkbox"/> Sponges			
<input type="checkbox"/> Baskets			
<input type="checkbox"/> Buckets			
<input type="checkbox"/> Goals			
<input type="checkbox"/> Bases			
<input type="checkbox"/> Auditory Bases			

## Parachute

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Large parachute	<input type="checkbox"/> Limit range	<input type="checkbox"/> Smooth surface	<input type="checkbox"/> Explanation
<input type="checkbox"/> Small parachute	<input type="checkbox"/> of motion	<input type="checkbox"/> Grass	<input type="checkbox"/> Physical assistance
<input type="checkbox"/> Towel	<input type="checkbox"/> Increase range	<input type="checkbox"/> Mats	<input type="checkbox"/> Guided discovery
<input type="checkbox"/> Straps on chute	<input type="checkbox"/> of motion	<input type="checkbox"/> Lighting dim/bright	<input type="checkbox"/> Problem solving
<input type="checkbox"/> Ace bandage	<input type="checkbox"/> Change objective	<input type="checkbox"/> Stimulating	<input type="checkbox"/> Demonstration
<input type="checkbox"/> around hand	<input type="checkbox"/> Limit # of shakes	<input type="checkbox"/> Limit distractions	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Bells on chute	<input type="checkbox"/> Increase # of	<input type="checkbox"/> Consistency	<input type="checkbox"/> Peer tutoring
<input type="checkbox"/> center/perimeter	<input type="checkbox"/> shakes		
<input type="checkbox"/> Balls in chute	<input type="checkbox"/> Limit amount		
<input type="checkbox"/> bright or auditory	<input type="checkbox"/> of movement		
<input type="checkbox"/> Parachute bright			
<input type="checkbox"/> Parachute tactile			

## Twister

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Bed sheet	<input type="checkbox"/> Use only hands	<input type="checkbox"/> Limit space	<input type="checkbox"/> Explanation
<input type="checkbox"/> Blanket	<input type="checkbox"/> Use only feet	<input type="checkbox"/> Add space	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Raised shapes	<input type="checkbox"/> Use right and left	<input type="checkbox"/> Limit distractions	<input type="checkbox"/> Physical
<input type="checkbox"/> Shapes in	<input type="checkbox"/> Count number found	<input type="checkbox"/> Add support staff	<input type="checkbox"/> assist
<input type="checkbox"/> different textures	<input type="checkbox"/> Play with partner	<input type="checkbox"/> Give commands	<input type="checkbox"/> Braille
<input type="checkbox"/> Raised shapes	<input type="checkbox"/> Change rules	<input type="checkbox"/> on a board	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Different colors	<input type="checkbox"/> Change objective	<input type="checkbox"/> Lighting dim/bright	<input type="checkbox"/> Problem solving
<input type="checkbox"/> Numbers in rows	<input type="checkbox"/> Limit space/objects	<input type="checkbox"/> Limit sound distractions	<input type="checkbox"/> Teach signs for
<input type="checkbox"/> Letters in rows	<input type="checkbox"/> to touch	<input type="checkbox"/> Cues given ex:	<input type="checkbox"/> for shapes, colors,
<input type="checkbox"/> Braille spinner	<input type="checkbox"/> Add space/objects	<input type="checkbox"/> light for right and left	<input type="checkbox"/> textures
<input type="checkbox"/> Large print spinner	<input type="checkbox"/> to touch		
<input type="checkbox"/> Tactile spinner	<input type="checkbox"/> Commands written		
	<input type="checkbox"/> Increase time allotted		

## Roller-skating/Rollerblading

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Velcro skates	<input type="checkbox"/> Skate slow	<input type="checkbox"/> Smooth surface	<input type="checkbox"/> Explanation
<input type="checkbox"/> Brakes front	<input type="checkbox"/> Skate forward	<input type="checkbox"/> Soft surface	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> & back	<input type="checkbox"/> Skate backwards	<input type="checkbox"/> Inclined surface	<input type="checkbox"/> Physical Assistance
<input type="checkbox"/> Wrist, knee pads	<input type="checkbox"/> Skate in a circle	<input type="checkbox"/> Limited space	<input type="checkbox"/> Problem solving
<input type="checkbox"/> helmet	<input type="checkbox"/> Skate w/partner	<input type="checkbox"/> Larger space	<input type="checkbox"/> Guided discovery
<input type="checkbox"/> Trash can on	<input type="checkbox"/> Skate in a line	<input type="checkbox"/> Lighting dim/bright	
<input type="checkbox"/> wheels for	<input type="checkbox"/> with group	<input type="checkbox"/> Limit distractions	
<input type="checkbox"/> support	<input type="checkbox"/> Obstacle course	<input type="checkbox"/> Challenging	
<input type="checkbox"/> Shopping cart	<input type="checkbox"/> Skate for distance	<input type="checkbox"/> using obstacles	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> for support	<input type="checkbox"/> Skate for time	<input type="checkbox"/> Limited people	<input type="checkbox"/> Small group
<input type="checkbox"/> Walker on wheels	<input type="checkbox"/> Skate & dance	<input type="checkbox"/> in the way	<input type="checkbox"/> 1:1
<input type="checkbox"/> Tether	<input type="checkbox"/> Roller hockey	<input type="checkbox"/> Supportive	<input type="checkbox"/> Intervener
<input type="checkbox"/> Bar held by peer	<input type="checkbox"/> Skate for		<input type="checkbox"/> explanation
<input type="checkbox"/> Hula hoop held	<input type="checkbox"/> transportation		<input type="checkbox"/> by peer

## SECONDARY PHYSICAL ACTIVITY EXAMPLES

### Volleyball

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Volleyball	<input type="checkbox"/> No change	<input type="checkbox"/> Tactile boundaries	<input type="checkbox"/> Physical assistance
<input type="checkbox"/> Larger ball	<input type="checkbox"/> One bounce	<input type="checkbox"/> Visual boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Smaller ball	<input type="checkbox"/> Two bounces	<input type="checkbox"/> Limit distractions	<input type="checkbox"/> 1:1
<input type="checkbox"/> Lighter ball	<input type="checkbox"/> Catch the ball	<input type="checkbox"/> Success oriented	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Beach ball	<input type="checkbox"/> Walk with ball	<input type="checkbox"/> Limit space	<input type="checkbox"/> Slow down
<input type="checkbox"/> Balloon	<input type="checkbox"/> 3 passes		<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Heavier ball	<input type="checkbox"/> 4 passes		<input type="checkbox"/> Feedback
<input type="checkbox"/> V-ball trainer	<input type="checkbox"/> Unlimited passes		<input type="checkbox"/> Task style
<input type="checkbox"/> Lower net	<input type="checkbox"/> Serve closer to net		<input type="checkbox"/> Intervener
<input type="checkbox"/> Higher the net	<input type="checkbox"/> No dunking		<input type="checkbox"/> play by play
<input type="checkbox"/> Auditory ball	<input type="checkbox"/> Limited space		<input type="checkbox"/> explanation
	<input type="checkbox"/> Cooperation vs. Competition		
	<input type="checkbox"/> More than 1 hit		

### Swimming

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Personal floatation	<input type="checkbox"/> Cooperation vs competition	<input type="checkbox"/> Small space	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Kick boards	<input type="checkbox"/> Elementary stroke	<input type="checkbox"/> Large space	<input type="checkbox"/> Brailing
<input type="checkbox"/> Lane lines	<input type="checkbox"/> Side stroke	<input type="checkbox"/> Shallow end	<input type="checkbox"/> Verbal cues
<input type="checkbox"/> Deck rings	<input type="checkbox"/> Breast stroke	<input type="checkbox"/> Deep end	<input type="checkbox"/> 1:1
<input type="checkbox"/> Inner tubes	<input type="checkbox"/> Back stroke	<input type="checkbox"/> Lane assignments	<input type="checkbox"/> Direct
<input type="checkbox"/> Rafts	<input type="checkbox"/> Floating	<input type="checkbox"/> Tactile	<input type="checkbox"/> Indirect
<input type="checkbox"/> Mats	<input type="checkbox"/> Diving	<input type="checkbox"/> Auditory	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Bright balls	<input type="checkbox"/> Surface diving	<input type="checkbox"/> Boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Bell balls	<input type="checkbox"/> Breath control	<input type="checkbox"/> Bright tape	<input type="checkbox"/> Proximity
<input type="checkbox"/> Radio	<input type="checkbox"/> Propulsion	<input type="checkbox"/> Roped areas	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Extension pole	<input type="checkbox"/> Kicking	<input type="checkbox"/> Padding	<input type="checkbox"/> Feedback
<input type="checkbox"/> Whistle	<input type="checkbox"/> Arm stroke	<input type="checkbox"/> Lights	<input type="checkbox"/> Guided discovery
<input type="checkbox"/> Beach ball	<input type="checkbox"/> games	<input type="checkbox"/> lighter/dimmer	<input type="checkbox"/> Task style
	<input type="checkbox"/> Object control skills in pool		<input type="checkbox"/> Small group

## Basketball

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Auditory balls	<input type="checkbox"/> Bounce pass	<input type="checkbox"/> Bright boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Bright balls	<input type="checkbox"/> Double dribble	<input type="checkbox"/> Carpet squares	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Tactile balls	<input type="checkbox"/> Undefined	<input type="checkbox"/> Shooting line	<input type="checkbox"/> Verbal cues
<input type="checkbox"/> Bell balls	<input type="checkbox"/> Time limitations	<input type="checkbox"/> Tactile lines	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Buzzer basket	<input type="checkbox"/> Boundary limits	<input type="checkbox"/> Auditory lines	<input type="checkbox"/> Braille
<input type="checkbox"/> High basket	<input type="checkbox"/> Free shooting	<input type="checkbox"/> Cones	<input type="checkbox"/> Bright clothing
<input type="checkbox"/> Low basket	<input type="checkbox"/> Throw-ins	<input type="checkbox"/> Ropes	<input type="checkbox"/> 1:1
<input type="checkbox"/> Bright basket	<input type="checkbox"/> Walk with ball	<input type="checkbox"/> Clap under basket	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Whistle	<input type="checkbox"/> Run with ball		<input type="checkbox"/> Slow down
	<input type="checkbox"/> Cooperation vs. competition		<input type="checkbox"/> Demonstrate
	<input type="checkbox"/> Space limit		<input type="checkbox"/> Feedback
	<input type="checkbox"/> Keep away		<input type="checkbox"/> Indirect
			<input type="checkbox"/> Direct
			<input type="checkbox"/> Task style

## Soccer

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Auditory balls	<input type="checkbox"/> Ground pass	<input type="checkbox"/> Bright boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Bright balls	<input type="checkbox"/> Timed dribble	<input type="checkbox"/> Carpet squares	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Tactile balls	<input type="checkbox"/> Undefined	<input type="checkbox"/> Shooting line	<input type="checkbox"/> Verbal cues
<input type="checkbox"/> Bell balls	<input type="checkbox"/> Time limitations	<input type="checkbox"/> Tactile lines	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Nerf balls	<input type="checkbox"/> Boundary limits	<input type="checkbox"/> Auditory lines	<input type="checkbox"/> Braille
<input type="checkbox"/> Bells on net	<input type="checkbox"/> Free shooting	<input type="checkbox"/> Cones	<input type="checkbox"/> Bright clothing
<input type="checkbox"/> Buzzer on net	<input type="checkbox"/> Throw-ins	<input type="checkbox"/> Ropes	<input type="checkbox"/> 1:1
<input type="checkbox"/> Radio	<input type="checkbox"/> Walk with ball	<input type="checkbox"/> Clap behind goal	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Fan	<input type="checkbox"/> Run with ball		<input type="checkbox"/> Slow down
	<input type="checkbox"/> Cooperation vs. Competition		<input type="checkbox"/> Demonstrate
	<input type="checkbox"/> Peer place ball		<input type="checkbox"/> Feedback
	<input type="checkbox"/> Free kicks		<input type="checkbox"/> Indirect
			<input type="checkbox"/> Direct
			<input type="checkbox"/> Task style

## Track and Field

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Bright lane lines	<input type="checkbox"/> Set # of trials	<input type="checkbox"/> Bright boundaries	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Auditory start/finish	<input type="checkbox"/> Time limitations	<input type="checkbox"/> Carpet squares	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Soft hurdles	<input type="checkbox"/> Boundary limits	<input type="checkbox"/> Throwing line	<input type="checkbox"/> Verbal cues
<input type="checkbox"/> Low hurdles	<input type="checkbox"/> Guide runner	<input type="checkbox"/> Tactile lines	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Ropes	<input type="checkbox"/> Cooperation vs. competition	<input type="checkbox"/> Auditory lines	<input type="checkbox"/> Braille
	<input type="checkbox"/> Peer runners	<input type="checkbox"/> Cones	<input type="checkbox"/> Bright clothing
<input type="checkbox"/> Tethers	<input type="checkbox"/> Limited space	<input type="checkbox"/> Ropes	<input type="checkbox"/> 1:1
<input type="checkbox"/> Sand pits	<input type="checkbox"/> Clap in target areas	<input type="checkbox"/> Jumping line	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Auditory jump board	<input type="checkbox"/> Form vs. time	<input type="checkbox"/> Success oriented	<input type="checkbox"/> Slow down
<input type="checkbox"/> Tactile jump boards		<input type="checkbox"/> Limited distractions	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Auditory high jump			<input type="checkbox"/> Feedback
<input type="checkbox"/> Bright colored bars			<input type="checkbox"/> Indirect
<input type="checkbox"/> Various throwing objects			<input type="checkbox"/> Direct
			<input type="checkbox"/> Tandem run
			<input type="checkbox"/> Play by play action
			<input type="checkbox"/> Task style



## Baseball/Softball

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Beep ball	<input type="checkbox"/> Set # of strikes	<input type="checkbox"/> Guide rails	<input type="checkbox"/> Peer tutor
<input type="checkbox"/> Auditory balls	<input type="checkbox"/> Set # of batting trials	<input type="checkbox"/> Carpet squares	<input type="checkbox"/> Physical assist
<input type="checkbox"/> Bright balls	<input type="checkbox"/> Hit off ground	<input type="checkbox"/> Shooting line	<input type="checkbox"/> Verbal cues
<input type="checkbox"/> Tactile balls	<input type="checkbox"/> Hit off tee	<input type="checkbox"/> Tactile lines	<input type="checkbox"/> Close proximity
<input type="checkbox"/> Bell balls	<input type="checkbox"/> Hit off fly	<input type="checkbox"/> Auditory lines	<input type="checkbox"/> Braille
<input type="checkbox"/> Nerf balls	<input type="checkbox"/> No tag-outs	<input type="checkbox"/> Cones	<input type="checkbox"/> Bright clothing
<input type="checkbox"/> Whiffle ball	<input type="checkbox"/> Tag-outs on bases	<input type="checkbox"/> Ropes	<input type="checkbox"/> 1:1
<input type="checkbox"/> Bells on bases	<input type="checkbox"/> Ground pass	<input type="checkbox"/> Clap behind goal	<input type="checkbox"/> Task analyze
<input type="checkbox"/> Buzzer on bases	<input type="checkbox"/> Time limitations	<input type="checkbox"/> Bright lines	<input type="checkbox"/> Slow down
<input type="checkbox"/> Radio	<input type="checkbox"/> Boundary limits	<input type="checkbox"/> Open space	<input type="checkbox"/> Demonstrate
<input type="checkbox"/> Fan	<input type="checkbox"/> Throw ball out	<input type="checkbox"/> Success oriented	<input type="checkbox"/> Feedback
<input type="checkbox"/> Tee	<input type="checkbox"/> Cooperation vs. competition	<input type="checkbox"/> Increase stimulation	<input type="checkbox"/> Indirect
	<input type="checkbox"/> Peer place ball	<input type="checkbox"/> continuous explanation	<input type="checkbox"/> Intervener
	<input type="checkbox"/> Peer runs		<input type="checkbox"/> Direct
	<input type="checkbox"/> 2 bases only		<input type="checkbox"/> Tandem run
	<input type="checkbox"/> Limited space		<input type="checkbox"/> Play by play action
	<input type="checkbox"/> Hit hanging ball		<input type="checkbox"/> Task style

## Golf

Equipment	Rules	Environment	Instruction
<input type="checkbox"/> Larger heads	<input type="checkbox"/> Limit distance	<input type="checkbox"/> Uncluttered-	<input type="checkbox"/> Explanation
<input type="checkbox"/> Larger balls	<input type="checkbox"/> Add distance	<input type="checkbox"/> no sand or ponds	<input type="checkbox"/> Demonstration
<input type="checkbox"/> Auditory balls	<input type="checkbox"/> Count every other	<input type="checkbox"/> Limit distractions	<input type="checkbox"/> Physical assistance
<input type="checkbox"/> Brighter balls	<input type="checkbox"/> hit or every third	<input type="checkbox"/> Predictable	<input type="checkbox"/> Braille
<input type="checkbox"/> Larger grip	<input type="checkbox"/> Move ball to	<input type="checkbox"/> Braille description	<input type="checkbox"/> Peer tutor/buddy
<input type="checkbox"/> Shorter club	<input type="checkbox"/> preferred spot	<input type="checkbox"/> of course	<input type="checkbox"/> Command style
<input type="checkbox"/> Hockey stick	<input type="checkbox"/> Pair up w/buddy	<input type="checkbox"/> Consistent	<input type="checkbox"/> Task style
<input type="checkbox"/> in place of club	<input type="checkbox"/> Play in teams	<input type="checkbox"/> Play on cloudy day	<input type="checkbox"/> Problem solving
<input type="checkbox"/> String tied to	<input type="checkbox"/> Play for fun w/	<input type="checkbox"/> Play at night w/	<input type="checkbox"/> Intervener explain
<input type="checkbox"/> ball and club	<input type="checkbox"/> no score	<input type="checkbox"/> bright ball	<input type="checkbox"/> game play by play
<input type="checkbox"/> Golf cart	<input type="checkbox"/> Play frisbee golf	<input type="checkbox"/> Practice at a range	<input type="checkbox"/> Task analyze
<input type="checkbox"/> w/driver	<input type="checkbox"/> Allow participant	<input type="checkbox"/> Supportive	<input type="checkbox"/> Cooperative games
<input type="checkbox"/> Lighter clubs	<input type="checkbox"/> to make up own course		<input type="checkbox"/> Show course on
	<input type="checkbox"/> Allow more than one attempt at each hole		

\*The previous curriculum concept was created by Michele Carron, Assistant Professor at SUNY Brockport.

\*Contributors to the above curriculum were: Wendy Kohler Graduate students, Annette Scorse, Sal Estremo undergraduate students



## FINDING HELP

Check out the Resources Section (IV). When asked, these are some of the things parents said were helpful to them (special thanks to Michael Schwartz, Michelle Westmaas, Bonnie Haggerty, Lisa Weir, and Jeanne McMullen for their input)

### **Other Parents:**

Finding/getting help from *other parents* has been the most beneficial to us if for no other reason than knowing we are not alone in our struggle. *A good way to contact other parents is through the CHARGE listserv. See the website [www.chargesyndrome.org](http://www.chargesyndrome.org) for a link to the list. Going to the biennial conferences is another good way to network with parents.* Ask the nurses or doctors if they know of another family you can contact.

### **Early Intervention:**

Try to find early intervention from birth. Our hospital identified Patty from the PICU and they came to the house once a week from the time she was sent home. *Ask the **hospital social workers** about early intervention or Birth to Three program referrals.*

### **Parent to Parent:**

Parent to Parent is a wonderful organization that provides parents with assistance in areas that only a parent of a special needs child can know about. They have coordinators who can match up parents with other parents with similar struggles. These parents can provide assistance with educational battles, insurance battles, emotional support, and much more. They have a national 800# as well as regional chapters throughout the country.

### **Educational consultants and Case Managers**

Identify educational consultants and case managers as early as possible. *Again, hospital social workers may help. Also try calling your local school district special services program.*

### **Community Agencies:**

Finding community agencies. Look through your phone book blue pages page for page under "Human Services," paying particular attention to the headings like "Services for the Disabled." Your local Easter Seals, ARC, and Parent to Parent Chapter can also steer you to your providers of early intervention resources. Request any pamphlets any organizations you speak with have. Often they have pamphlets that can steer you towards other services such as respite or legal aid.

Danny has been getting services through the school district, Easter Seals, the Regional Center (for developmentally disabled persons) and California Children's Services. *All states have similar programs, including DeafBlind services.*

### **Ask Lots of Questions:**

Ask questions and follow up on programs, even those that don't sound like they would help (at first). For example, several people at the hospital told us to register with California Children's Services, and one doctor even referred us there. I didn't think we would qualify because of our income, but I made an appointment anyway. Sure enough, we make too much money for their main program (which would have been of little interest to me anyway since we have health insurance). But on the way out, the eligibility worker said, "the only thing you would qualify for is occupational therapy and physical therapy." When I got home, I started to think that that "only thing" could be a very helpful thing. I called back (it took a few calls on my part) and got Danny in a therapy program that has been a big help.

**Ask about other services:**

Ask each agency if they have any other programs, do they provide, e.g., speech therapy, respite care, etc., or do they know some agency that would. Some of these agencies are worried about their budgets and don't volunteer info unless you ask. Some will even lie or say know if you don't ask exactly right, so it helps to ask around a bit.

**DeafBlind services:**

Every state has DeafBlind services. You can find out more from DBLink and NTAC (see at Resources)

**Special Education services:**

In most states, special education services start at birth or age three – long before “school age.” One place to start is to get a copy of the federal IDEA law and your state's special education regulations. The state document should be free at your request from the State Board of Education. I read several books on IEP law, parents' rights, how to advocate, etc, but I can't recall any titles. I searched on Amazon.com and Barnes & Noble.com then took the titles I found to the local library. They got them for me through the inter-library loan system (we're in a small rural community). Another source I used for access to reading material was my local special ed and early intervention offices and our state spec ed library clearinghouse.

Request your first case study and IEP meeting as soon as possible. Plan to discuss the team's findings and ideas and then adjourn. Take time to mull it all over, research some more, and reconvene when you are armed with all the info you need to get what your child needs. This strategy worked well for me, but that's because it fits my personality.

**Parent Training:**

I learned a lot from reading but also found parent trainings to be essential. Find out who would sponsor these trainings in your area. If you can get on the right mailing list, you will be aware of advocacy trainings, parent retreats, etc. I met many professionals who have served as wonderful sources of information by networking at various trainings.

Go to advocacy workshops to learn and network. Contact Parent to Parent and go to their social functions. You'll learn through your contacts the people and agencies who are particularly helpful for their families and who do not

Press the professionals for as much information as you can. Social workers are often overworked and will often dedicate their limited time to the families that they deem "most needy" so you really need to effectively communicate exactly what your needs are and keep on them for answers.

**Other Financial help:**

The things most helpful have been Family Support Services (FSS) funds which in some form are in every state. These funds can defray respite costs and assistive technology costs. The FSS case manager can be instrumental in helping us obtain emergency respite care. *Ask the hospital or other social workers about these programs.*

**Other helpful things:**

The Massachusetts loophole (nationally known as the Katie Beckett Waiver) and the Health Insurance Premium Plan (HIPP) program. Through HIPP, the state of Pennsylvania pays the private health care premiums of KB waiver recipients when private healthcare is available through the parent's employer at less cost than the waiver (saving us much \$\$\$).

## CHARGE SYNDROME: GLOSSARY

Compiled by Meg Hefner, M.S.

### A

**Accutane:** (isotretinoin, retinoic acid) prescription medication used to treat severe cystic acne. It is a synthetic derivative of Vitamin A. When taken during pregnancy, it can cause very serious birth defects, including hydrocephalus, microcephaly (very small head), mental retardation, small and malformed ears and other facial abnormalities, and heart defects. Although the ear malformations can be similar to those seen in CHARGE, the face is distinct.

**amblyopia:** "lazy eye" - poor vision in one eye without detectable cause. Often treated by patching the stronger eye.

**anesthesia:** 1. Loss of sensation resulting from pharmacologic depression of nerve function or from neurological dysfunction. 2. Broad term for anesthesiology as a clinical specialty. Sedation used during surgery

**anophthalmia:** absence of the eye or eyes. Anophthalmia can be considered the most severe form of a coloboma.

**aorta:** the large artery arising from the base of the left ventricle of the heart. The aortic arch is formed by the ascending aorta and the descending aorta.

**aortic arch:** The curved portion between the ascending and descending parts of the aorta. Aortic arch anomalies are common in CHARGE

**apnea:** A potentially life-threatening condition in which breathing stops abnormally, usually during sleep. CPR is sometimes needed to start the breathing again.

**arhinencephaly:** a term used to refer to an absence of some or all areas of the forebrain, or certain areas of the anterior (front) portions of the brain, particularly the olfactory bulbs and nerves. The defect may occur relatively late in fetal differentiation, so that only a small area of the brain is involved. One result can be the lack of a sense of smell.

**aspiration:** The inspiratory sucking into the airways of fluid or foreign body, as in vomitus. Children with CHARGE are at risk for aspiration of food or liquid which is not adequately swallowed or which is refluxed up from the stomach.

**aspiration pneumonia:** A lung inflammation caused by inhaling a foreign body, such as food, into the lungs.

**association:** A connection of persons, things or ideas by some common factor. In genetics: the occurrence together in a population, more often than can be readily explained by chance, of two or more traits of which at least one is known to be genetic. For example VACTERAL association.

**atresia:** A general term for closure of a structure that should be open, such as a canal, passage or tube. See choanal atresia, esophageal atresia, and ear anomalies.

**atrial septic defect (ASD):** A hole between the two upper chambers (atria) of the heart. Surgery is usually required to close an ASD.

**audiogram:** The graphic record drawn from the results of hearing tests with audiometer. The audiogram charts the threshold of hearing at various frequencies (pitch) against sound intensity in decibels (loudness). Pure tone audiogram: a chart of the threshold for hearing acuity at various frequencies usually expressed in decibels above normal threshold and usually covering frequencies from 128 to 8000 Hz. Speech audiogram: the record of thresholds for spondaic word lists and scores for phonetically balanced word lists. See Ears and Hearing Section.

**autistic disorder:** Impaired development of social interaction and communication along with repetitive and stereotyped patterns of behavior, interests, and activities, with an onset prior to age three.

**autistic behaviors** may include withdrawal from social contact, avoidance of eye contact, and failure to develop friendships; delay or total lack of communication, or communication is abnormal; self-stimulation, preoccupation with objects or parts of objects, obsession with certain routines or rituals. Autistic-like behaviors are observed in some children with CHARGE. It is not clear whether to attribute them to central nervous system structural abnormalities, to dual sensory impairment, or whether a separate diagnosis of autism is appropriate.

## **B**

**BAER:** Abbreviation for "brainstem auditory evoked response." Measurement of alteration in the electrical activity of the auditory system of the brain brought about by presenting sounds through earphones. This can be done with the child sedated and without the cooperation of the child.

**bilateral** Two-sided, or affecting both sides.

## **C**

**cartilage** connective tissues found primarily in joints, the walls of the thorax, and tubular structures such as the larynx, air passages, and ears; comprises most of the skeleton in early fetal life, but is slowly replaced by bone. Cartilage is often weak in children with CHARGE, resulting in floppy ears, laryngomalacia and other complications.

**Cardiologist:** physician who specializes in the heart.

**central auditory processing** A central hearing loss that may prevent sound from being interpreted meaningfully by the brain. Sound may be picked up by the ears and signals transmitted to the brain, but the brain has difficulty making sense of the input.

**central nervous system (CNS) abnormalities:** CNS abnormalities seen in children with CHARGE Syndrome include: structural brain abnormalities (diagnosed by CT scan or

MRI), microcephaly, seizures, apnea, and central processing problems (including a central hearing loss). See also: cranial nerve abnormalities and Brain Section.

**cerebellum**      The large back portion of the brain. It consists of the two lateral hemispheres united by a narrow middle portion, the vermis.

**cerebrum**      Originally referred to the largest portion of the brain, including practically all parts within the skull except the medulla, pons, and cerebellum; it now usually refers only to the parts derived from the telencephalon and includes mainly the cerebral hemispheres (cerebral cortex and basal ganglia).

**cerumen:**      ear wax

**CHARGE facial features:**      Children with CHARGE Syndrome resemble their parents, but they may also have some of the following features that make them look similar to other children with CHARGE Syndrome: square shape of the face and head, flat cheekbones, facial asymmetry (with or without facial palsy), wide nose with high bridge, and unusual ears (see Ear anomalies). The facial features do not cause any health problems, but can be very helpful in making the diagnosis of CHARGE Syndrome. See Diagnosis Section and photos with stories.

**choroid (eye):**      The portion of the eye lying between the retina and the sclera.

**chromosomes:**      Microscopic structures found in the nucleus of cells which contain the genetic information (DNA). Possible chromosome tests include routine (g-banded, including most prenatal chromosome testing), fragile X (to diagnose a particular cause of mental retardation), and FISH (to look for specific microdeletions or other abnormalities). No specific chromosome abnormalities have yet been shown to cause CHARGE. In VCF, there is a small deletion (microdeletion) of chromosome 22.

**choanal atresia or stenosis:**      The choanae are the passages from the back of the nose to the throat which allow breathing through the nose. Choanal atresia is a birth defect in which this passage is completely blocked; stenosis refers to a narrowed passage. The atresia or stenosis can be on one or both sides. If a baby is born with bilateral choanal atresia, immediate surgery is needed to permit the baby to breathe through the nose. Choanal stenosis can lead to problems with breathing or increased nasal stuffiness. See Choanal atresia Section.

**cleft lip and/or cleft palate:**      Many children with CHARGE Syndrome have a cleft lip (hare lip) and/or cleft palate (an opening in the roof of the mouth). Children with a cleft palate may have problems with ear infections and speech even after surgical correction. Submucous cleft palate may be difficult to diagnose in children with CHARGE. See Cleft lip and palate Section.

**cochlea:**      part of the inner ear, responsible for transmitting sound to the auditory nerve.

**cochlear implant**      A device that changes sound into electrical signals. These signals are then sent through the skin to an electrode array surgically implanted within the cochlea. Some individuals with severe to profound sensorineural hearing loss can get some hearing through a cochlear implant. Experience with cochlear implants in children with

CHARGE is limited.

**coloboma:** A cleft or keyhole-shaped defect of the eyeball. Colobomas can occur anywhere in the eye (any combination of iris, retina, or disc) and can affect one or both eyes. A coloboma of the iris (colored part of the eye) will result in a keyhole-shaped pupil, but probably will not affect vision. A coloboma of the retina or disc in the back of the eye can only be detected by an eye exam done by an experienced ophthalmologist. Retinal or disc colobomas can cause significant vision loss, both by restriction of the visual field (large blind spots across the top of normal vision) and by decreased acuity (blurred vision). See Eye Section.

**columella:** the part of the nose between the nostrils. It is sometimes prominent (sort of long) in children with CHARGE. See CHARGE face photos in Diagnosis Section.

**conductive hearing loss:** Hearing loss due to abnormalities of the middle ear bones (ossicles) and/or to fluid accumulation in the middle ear. If the loss is due to fluid accumulation, it may get better with use of PE tubes and/or decongestants. If the loss is due to malformed ossicles, surgery might be considered, but this is controversial.

**conotruncal anomalies of the heart:** Most common heart defects in CHARGE, including, double outlet right ventricle, tetralogy of Fallot, interrupted aortic arch, and VSD.

**corpus callosum:** the plate of nerve fibers which connect the two cortical hemispheres of the brain. Some individuals with CHARGE have absent or hypoplastic (underdeveloped) corpus callosum.

**cranial nerves** those nerves that emerge from, or enter, the cranium or skull, in contrast to the spinal nerves, which emerge from the spine or vertebral column. The twelve paired (one on each side) cranial nerves are the I - olfactory, II - optic, III - oculomotor, IV - trochlear, V - trigeminal, VI - abducent, VII - facial, VIII - vestibulocochlear, IX - glossopharyngeal, X - vagal, XI - accessory, and XII - hypoglossal. Cranial nerve abnormalities are very common in CHARGE.

**cryptorchidism:** undescended testicles. If the testes do not descend into the scrotum, they can be lowered surgically.

**CT scan:** computerized tomography. A special X-ray of the head used to look at the structure of the brain.

## **D**

**Dandy-Walker malformation or cyst:** A developmental anomaly of the fourth ventricle of the brain. It can result in cerebellar hypoplasia, hydrocephalus, and posterior fossa cyst formation. Occasionally seen in children with CHARGE.

**developmental delay:** Most children with CHARGE Syndrome will have delayed or retarded development. Vision loss causes delays in motor development. hypotonia (low muscle tone) and balance problems also delay motor development. Hearing loss can cause delays in speech and language. As development depends on the combination of intelligence, hearing, and the ability to see, early developmental delay does not always



mean mental retardation (see mental retardation). Many children with CHARGE have normal intelligence.

DiGeorge sequence: congenital absence of the thymus and parathyroid glands in combination with heart defects. The thymus problems lead to increased infections. Common in VCF syndrome (22q deletion), rare in CHARGE.

dysfunction: difficult or abnormal function

## E

ear anomalies: Children with CHARGE Syndrome often have misshapen ears. Often the ears have characteristics which are unique to CHARGE. Typical "CHARGE ears" are small and wide, with little or no ear lobe. Often the outer fold of the ear (helix) is missing or may appear clipped-off. The child's two ears often look different. The unusually shaped ears do not cause hearing loss unless there is stenosis (narrowing) or atresia (collapse) of the ear canal, which is rare in CHARGE Syndrome. If a hearing aid is required, the ear anomalies may make it difficult to fit the ear mold properly. The ear anomalies can be very helpful in making the diagnosis of CHARGE Syndrome because they look different from ear anomalies seen in any other syndrome. See EARS and Hearing and Diagnosis Sections.

echocardiogram the ultrasonic record obtained by echocardiography. Sound waves are used to get an image of the heart to diagnose heart defects.

EEG (electroencephalogram): brain wave test used to look for seizure activity.

Endocrinologist: physician who specializes in treatment of hormone abnormalities.

esophageal atresia: the esophagus (food pipe) ends in a pouch instead of connecting to the stomach. Babies with this abnormality spit up all their food until it is surgically corrected. Even after surgery, feeding may be difficult for some time due to other swallowing problems which are seen in CHARGE Syndrome. See TEF/EA Section.

Eustachian tube: the tube leading from the tympanic cavity (behind the eardrum) to the nasopharynx (back of the throat). It enables equalization of pressure within the tympanic cavity with ambient air pressure, referred to commonly as "popping of the ears." Blocked Eustachian tubes predispose to ear infections.

evoked potentials auditory screening for newborns using auditory evoked potential measures, including auditory brainstem response (ABR, BAER)

exotropia: outward turning of the eyes.

## F

facial palsy or paralysis: The facial nerve (cranial nerve VII, which controls facial muscles) does not work in many children with CHARGE. This can cause a lopsided smile and trouble blinking the eye on the affected side. The affected eye may not produce tears very well. It may be unilateral or bilateral. If bilateral, it leads to a very blank expression. Facial

nerve palsy can affect both eating and speaking.

**FISH** Fluorescent in-situ hybridization. A specialized chromosome test using fluorescent dyes to identify very small deletions or other abnormalities of chromosomes. Many children with CHARGE will have chromosome tests done including FISH for deletion of chromosome 22 seen in VCF.

**FM Trainer** FM (frequency-modulated) signal transmission, represents the most successful and largest market of assistive listening devices for children. FM systems have an advantage of an improved signal-to-noise ratio over hearing aids.

**fundoplication** An operation in which the opening from the esophagus to the stomach is tightened.

## **G**

**G-tube** gastrostomy tube (see below).

**Gastroenterologist:** physician who specializes in the esophagus, liver, spleen, bowel and pancreas.

**gastrostomy** An operation in which an artificial opening is made into the stomach through the wall of the abdomen to place a g-tube.

**gastrostomy tube:** a tube which is surgically placed in the stomach through the abdominal wall for feeding. This is done when the child cannot eat by mouth because of a severe cleft palate, tracheo-esophageal (T-E) fistula, or swallowing problems. Often, a gastrostomy button or Mic-key is used.

**GE Reflux** gastroesophageal reflux, the backward flow of food from the stomach into the esophagus, possibly into the pharynx where they can be aspirated between the vocal cords and down into the trachea; symptoms of burning pain and acid taste result; pulmonary complications of aspiration are dependent upon the amount, content, and acidity of the aspirate. This is very common in CHARGE.

**growth deficiency** small stature or size. In CHARGE, this can be due to multiple medical problems, growth hormone deficiency, or some other cause

**growth hormone deficiency:** Growth hormone (GH) is produced in the pituitary gland. It promotes body growth, fat mobilization, and the inhibition of glucose utilization. Most children with CHARGE are small. Some have documented GH deficiency. The frequency of GH deficiency in CHARGE is not yet known.

## **H**

**hearing loss:** About 85% of children with CHARGE Syndrome have some hearing loss. The loss can be conductive, sensorineural (nerve), or mixed (both) and can range from a mild hearing loss to profound deafness. The loss may be progressive. A CHARGE syndrome hearing loss is typically a mixed loss with a large conductive component in the low frequencies and a sensorineural loss or mixed loss for high frequency sounds. The

losses can be severe and very difficult to measure completely and accurately, especially in infants and young children. As a result, repeated testing may be required before a satisfactory set of results is obtained. See Ears and Hearing Section.

**heart disease, congenital (CHD):** About 2/3 of children with CHARGE are born with some kind of heart defect. Some have only a murmur, while others may have a life-threatening heart defect which requires surgery. Heart abnormalities described in children with CHARGE include tetralogy of Fallot, ASD, VSD, PDA, aortic arch anomalies, double outlet right ventricle, pulmonic stenosis, and others. All children in whom a diagnosis of CHARGE is suspected should have a cardiac evaluation.

**hockey-stick crease:** crease on the palm of the hand which bends up to form a deep crease between the index and middle fingers. Although this has no medical significance, it is very common in children with CHARGE and can help confirm the diagnosis. See photo in Diagnosis Section.

**holoprosencephaly:** failure of the forebrain or prosencephalon to divide into hemispheres or lobes; cyclopia occurs in the severest form. It is often accompanied by a deficit in midline facial development. Often diagnosed by MRI or CT. Rare in CHARGE.

**horseshoe kidney:** union of the lower or occasionally the upper extremities of the two kidneys by a band of tissue extending across the vertebral column, resulting in a single horseshoe-shaped kidney. See Renal Section for illustration.

**hydrocephalus :** a condition marked by an excessive accumulation of fluid in the brain, resulting in dilation of the ventricles of the brain and raised intracranial pressure; may also result in enlargement of the cranium and atrophy of the brain. Treated by surgical placement of a shunt. Rare in CHARGE.

**hydronephrosis ("water on the kidneys"):** Dilation (enlargement) of the pelvis and calices of one or both kidneys resulting from obstruction or backflow of urine.

**hypogonadism:** underdeveloped genital system. In boys, there may be a small penis or cryptorchidism (undescended testicles). In girls, the labia (external skin folds) may be small or absent. In children of both sexes, hypogonadism may prevent puberty unless hormones are given.

**hypoplasia:** underdevelopment of a tissue or organ.

**hypospadias:** a genital problem in males. The urethral opening is not at the end of the penis and needs to be corrected surgically.

**hypotonia:** low muscle tone. This can lead to a "floppy" baby. Many children with CHARGE have hypotonia, especially of the trunk (upper body), contributing to delay of some motor milestones such as sitting and walking and predisposing the children to scoliosis.

**IEP** Individualized Education Plan

**imperforate anus:** the anus is closed over and needs to be opened surgically. Sometimes only a thin membrane needs to be opened. More often the blind end of the large bowel needs to be connected to the skin on the belly as a colostomy. Later the end of the bowel is put down through an artificial opening created where the anus ought to be. Not the same as choanal atresia. This is not usually seen in CHARGE.

**incontinence:** inability to prevent the discharge of any of the excretions, especially of urine or feces. Includes bedwetting. Common in CHARGE.

**infections:** Children with CHARGE Syndrome tend to be very sickly, especially in the early years. The most common illnesses are recurrent ear infections and pneumonia. The pneumonias tend to become less frequent after two to three years of age. The ear infections and ear drainage may last well into the teens.

**inguinal hernia:** also known as a "rupture". This is a lump noted in the groin and is actually a small loop of intestine sticking out a small hole connecting the inside of the abdomen to the groin. If the intestine gets stuck there, gangrene can occur, so preventive surgery is necessary.

**iris:** the colored part of the eye, with the pupil in the center. A coloboma of the iris results in a keyhole-shaped pupil. Common in CHARGE.

**IUGR:** Intrauterine growth retardation. Slower than expected growth of a baby before birth. At birth, it will show up as low birth weight. Rare in CHARGE.

## **K**

**kidney abnormalities:** 40% of children with CHARGE have kidney abnormalities. Kidney abnormalities seen in CHARGE include hydronephrosis, small or absent kidney, posterior urethral valves, and kidney reflux. See Renal Section.

**kidney reflux:** back flow of urine into the kidney. This can result in eventual damage to the kidney. IVP is often needed to confirm reflux in the kidneys.

## **L**

**labia:** female genital folds. May be smaller than normal in girls with CHARGE.

**laryngomalacia or laryngotracheomalacia:** floppy airway. The presence of soft laryngeal cartilage, most often seen in the epiglottis of young children. Very common in CHARGE, and contributes to surgical risks of anesthesia, breathing problems and swallowing problems.

**larynx:** the organ of voice production; the part of the respiratory tract between the pharynx and the trachea; it consists of a framework of cartilages and elastic membranes housing the vocal folds and the muscles which control the position and tension of these elements. May be weak in CHARGE.

**limb defects** abnormalities of arms, legs, hands, or feet. Occasional limb defects seen in CHARGE include thumb and forearm abnormalities. See Muscles and Bones Section.

## M

**macula:** the center of the retina of the eye. The macula is responsible for seeing details. Coloboma of the retina can result in legal blindness.

**Medical Geneticist:** physician with medical training (most often in pediatrics or OB/Gyn) with subspecialty training in Medical Genetics or (more recently) physician with residency training in medical genetics and certification by the American Board of Medical Genetics of the AMA.

**mental retardation:** The intelligence in children with CHARGE syndrome ranges from normal to severe mental retardation. Intelligence is very hard to estimate and in fact is often underestimated, especially when children are young. Vision and hearing problems can delay speech and development. Special testing methods are needed to evaluate children with sensory deficits (See Assessment Sections). In addition to the sensory deficits, so many children with CHARGE spend a large portion of their early years in the hospital. Nevertheless, it is important to have your child evaluated early and often to help set up the most appropriate educational program possible. Evaluations should be done by specialists with experience testing children with sensory deficits (hearing and vision loss).

**microcephaly:** unusually small head

**micrognathia:** small jaw or chin

**micropenis:** abnormally small penis. See Genital Section.

**microphthalmia/microphthalmos:** abnormal smallness of the eye. A severe coloboma can result in microphthalmia or anophthalmia.

**midface:** the middle of the face, especially the cheekbone area. May be flattened or small in CHARGE.

**Mondini defect:** an abnormal opening from the semicircular canal into the middle ear. This is a potentially treatable (with surgery) cause of balance problems.

**MRI:** magnetic resonance imaging; a diagnostic form of imaging (of the brain or other body parts) Unlike conventional radiography or CT, MRI does not expose patients to ionizing radiation. In addition, it can provide superior 3-D images of the body's interior, delineating muscle, bone, blood vessel, nerve, organ, and tumor tissue. Often used to describe the exact choanal atresia and/or inner ear abnormalities in CHARGE.

## N

**nares:** nostrils. May be small or appear pinched in CHARGE.

**Neurologist:** physician who specializes in the nervous system.

NG tube: nasogastric tube. Feeding tube which is put through the nose, down the throat into the stomach for feeding.

nystagmus: involuntary movements of the eyeball, most often side to side or in circles.

## O

olfactory nerve: cranial nerve I, responsible for smell. Often absent or abnormal in children with CHARGE.

omphalocele: failure of the abdominal wall to close properly around the umbilical cord during fetal development. Often some of the intestines are outside the body. This can usually be corrected surgically. An umbilical hernia is a very mild omphalocele.

Ophthalmologist: MD who specializes in the eyes. Children with CHARGE should be evaluated by a Pediatric Ophthalmologist.

optic nerve: cranial nerve VII. Abnormalities of cranial nerve VII result in facial palsy.

optometrist: technician who specializes in detecting problems of visual acuity.

ossicles: tiny bones in the middle ear: anvil, stapes and hammer.

ossicular malformation: malformation of the small bones of the middle ear (hammer, anvil and stapes). Ossicular malformations are very common in CHARGE and result in conductive hearing loss.

OT: occupational therapist. A specialist in the development of fine motor, social, and adaptive skills.

otitis media: ear infections. These can occur when fluid accumulates in the middle ears, behind the eardrums. If the fluid is infected, the child will complain of pain and there will be hearing loss. If the fluid is not infected, the child usually will have no pain, but may still have significant hearing loss. In CHARGE Syndrome, otitis media often lasts into the teenage years and requires constant medical care (see PE tubes).

Otolaryngologist: specialist in hearing, either MD (ENT specialist) or PhD (Audiologist)

## P

PAX2: group of genes responsible for orchestrating development of eyes, ears, and kidney. No PAX2 gene abnormalities have yet been detected in children with CHARGE.

palmar crease: referring to the palm of the hand. Children with CHARGE often have an unusual "hockey-stick" palmar crease. See photos in Diagnosis Section.

parathyroid gland: adjacent to the thyroid gland; one of two small paired endocrine glands. They secrete parathyroid hormone that regulates the metabolism of calcium and phosphorus.

**PE Tubes:** tiny polyethylene (plastic) tubes which can be surgically placed in the eardrum to drain the excess fluid from behind the drum. This will help prevent hearing loss caused by recurrent otitis media. Often several sets are needed over many years in children with CHARGE.

**perseverative behavior:** repetitive, repeated behavior, often seen in children with sensory deficits.

**photophobia:** intolerance to light, especially bright lights

**pinna:** external ear. Often very unusual in CHARGE.

**pituitary abnormalities:** The pituitary is a gland at the base of the brain which produces several important hormones which help control growth, thyroid and sex gland function, and steroid production. The pituitary gland does not function properly in some children with CHARGE. This can result in deficiencies in growth hormone and/or in the sex hormones which cause puberty. If left untreated, these children will be short and will not develop secondary sex characteristics. Hormone therapy is available for these problems.

**polyhydramnios:** excess amount of amniotic fluid. In pregnancies where the baby has CHARGE, polyhydramnios can related to choanal atresia and/or swallowing abnormalities.

**postnatal:** occurring after birth

**posterior urethral valves:** small pieces of tissue which prevent urine from flowing out of the bladder through the urethra to the outside. This can cause back-up of urine and damage to the kidneys. Surgery is usually necessary.

**prenatal:** preceding birth; antenatal

**proprioception:** A sense or perception, usually at a subconscious level, of the movements and position of the body and especially its limbs, independent of vision; this sense is gained primarily from input from muscles and the vestibular (balance) apparatus.

**ptosis:** droopy eyelids, caused by a facial nerve problem in CHARGE syndrome.

## **R**

**renal:** nephric; relating to the kidney.

**retarded development:** see Developmental delay and Mental retardation.

**retina:** The part of the eye which receives light and transmits electrical signals to the brain, resulting in vision. Colobomas of the retina will result in blind spots and/or visual acuity (sharpness) abnormalities.

**retinal detachment:** separation of the retina from the choroid. This can result in blindness.

retinoic acid      Vitamin A acid; used topically in the treatment of acne. See Accutane.

## **S**

sclera:              whites of the eye

scoliosis:           curvature of the spine. Common in CHARGE at any age.

sedation:           medication given to calm and/or quiet a child to make testing easier and/or more accurate. Children with CHARGE can have unusual (more or less extreme) reactions to sedation.

semicircular canals:   part of the inner ear involved in balance

sensorineural hearing loss (nerve deafness):              hearing impairment due to disorders of the cochlear division of cranial nerve VIII (auditory nerve), the cochlea, or the mitochondrial nerve tracts, as opposed to conductive hearing loss. Very common in CHARGE.

sensory deficit:           vision loss and/or hearing loss.

sonogram:           ultrasound. These terms can be used interchangeably

short stature:      can be caused by a lack of growth hormone (see pituitary abnormalities). Some children with CHARGE Syndrome have normal levels of growth hormone but still have short stature from an unknown cause.

stapes:              the smallest of the three auditory bones in the middle ear.

stapedius tendon:      stapedius muscle; dampens vibration of the stapes by drawing head of stapes backward as a result of a protective reflex stimulated by loud noise. The stapedius tendon is often abnormal in CHARGE.

strabismus:      crossed eyes

stridor:              a high-pitched, noisy respiration, like the blowing of the wind; a sign of respiratory obstruction, especially in the trachea or larynx.

swallowing difficulties:   Many children with CHARGE Syndrome have trouble coordinating the muscles used for normal sucking and swallowing, even in the absence of other problems such as esophageal atresia or T-E fistula. This may be due to abnormalities in cranial nerves IX and X. The incoordination can lead to gagging, apnea (breathing stops), and pneumonia (food inhaled into the lungs, causing infection). In some cases, tube feeding (gastrostomy) is used until the child is able to learn to swallow. Children with CHARGE may not learn to swallow effectively until they are 5 or 6 years old. See Feeding Section.

syndrome:           the aggregate of signs and symptoms associated with any morbid process, and constituting together the picture of the disease. As in CHARGE syndrome.



## T

teratogen: a drug or other agent that causes abnormal fetal development, for example, Accutane. There are no teratogens known to cause CHARGE.

thymus: a gland in the lower part of the neck, that is necessary in early life for the normal development of immunological functions. It reaches its greatest relative weight shortly after birth and its greatest absolute weight at puberty; it then begins to involute, and much of the lymphoid tissue is replaced by fat. It is abnormal in children with DiGeorge sequence.

ToF: Tetralogy of Fallot. Complex heart defect often seen in CHARGE.

tracheo-esophageal (T-E) fistula: an abnormal connection between the trachea (wind pipe) and esophagus (food pipe). If it is not corrected surgically, food will get into the lungs and cause choking and/or pneumonia.

tracheomalacia: floppy airway due to weakness of elastic and connective tissue of the trachea. Common in CHARGE.

tracheostomy/tracheotomy: the operation of opening into the trachea to permit breathing directly into the throat, usually intended to be temporary. Often necessary in CHARGE.

## U

ultrasound: imaging procedure which uses high-frequency sound waves to get an image of a fetus during pregnancy. Echocardiography also uses ultrasound to image the heart.

umbilical hernia: see omphalocele.

unilateral: affecting one side only.

URI: upper respiratory infection

urinary tract malformations: The urinary tract malformations seen in CHARGE Syndrome include posterior urethral valves, abnormal kidney shape or location, and backup of urine from the bladder into the kidney (reflux).

UTI: urinary tract infection

## V

VACTERAL/VATER association Acronym for Vertebral defects, Anal atresia, Cardiac defects, TracheoEsophageal fistula and/or esophageal atresia, Renal anomalies and Limb defects. Many features overlap with those of CHARGE, but can usually be distinguished by a Medical Geneticist.

**VCF:** velocardiofacial syndrome; Shprintzen syndrome. Syndrome consisting of cleft palate, heart defects, learning disabilities, and distinct physical features. The overwhelming majority of children with VCF have a microdeletion of chromosome 22 detectable by FISH. Although many features overlap, it is distinct from CHARGE. The shapes of the ears, face, and hands are distinct from CHARGE. To date, no one with a definite diagnosis of CHARGE has been positive for the chromosome 22 deletion associated with VCF.

**velopharyngeal:** pertaining to the soft palate (velum palatinum) and the posterior nasopharyngeal wall.

**ventricles (head, heart)** A normal cavity or space. In the heart, the spaces through which blood is pumped (left and right ventricle). In the brain, fluid-filled spaces (lateral, third, and fourth ventricles - see hydrocephalus).

**vestibular:** relating to a vestibule, especially the vestibule of the ear. The vestibule of the ear is responsible for one component of balance. Vestibular abnormalities are common in CHARGE and contribute to delay in motor milestones such as sitting and walking.

**VER:** visual evoked responses; used to measure electrical activity of the parts of the brain used in vision.

**videofluoroscopy:** examination x-ray, using the fluoroscope, using an image intensifier and television camera for image detection and a video monitor for display to get live action pictures of , e.g. swallowing.

**visual field:** the portion of side view, and top and bottom view, in which a child can see or has functional vision. Colobomas cause visual field defects (blind spots).

**webbed neck:** wide or broad neck, often somewhat short. Many children with CHARGE have a webbed neck or just a short neck.

## CHARGE SYNDROME RESOURCES

Compiled by: Wendy Keedy and Betsy McGinnity

CHARGE Syndrome Foundation, Inc.  
2004 Parkade Blvd.  
Columbia, MO 65202-3121  
phone: (800)442-7604 Parent line (USA callers only)  
(573)499-4694 all others  
FAX: (573)499-4694  
e-mail: [marion@chargesyndrome.org](mailto:marion@chargesyndrome.org)  
<http://www.chargesyndrome.org>

### \*\*INTERNATIONAL CHARGE ORGANIZATIONS\*\*

CHARGE Family Support Group  
82 Gwendolen Ave  
London E13 0RD  
United Kingdom  
phone: (01)020 8552 6961  
<http://www.widerworld.co.uk/charge/>

The Australasian CHARGE Association  
P.O. Box 91  
Glenfield, N.S.W. 2167  
Australia  
phone: (02) 9829-4801  
e-mail: [austcharge@hotmail.com](mailto:austcharge@hotmail.com)

Queensland CHARGE Association  
16 Ronmack Street  
Chermside QLD 4032  
Australia  
phone: (07)3359-6297  
e-mail: [charge\\_qld@hotmail.com](mailto:charge_qld@hotmail.com)  
<http://www.users.bigpond.com/arrone/>

### \*\* CHARGE WEB PAGE and LISTSERVE\*\*

CHARGE Syndrome Foundation, Inc.  
<http://www.chargesyndrome.org>

Minnow's CHARGE WebPage:  
<http://www.geocities.com/Heartland/1220/charge@saber.net>

**\*\*DEAF AND DEAFBLIND RESOURCES\*\***

A.G. Bell Association for the Deaf  
3417 Volta Place, N.W.  
Washington, DC 20007-2778  
phone: (202) 337-5220 (v, TTY)  
<http://www.agbell.org/>

American Society for Deaf Children  
PO Box 1510  
Olney, MD 20830  
Attn: Mr. Ken Silverstein  
1-800-942-ASDC Parent line  
[ASDC1@aol.com](mailto:ASDC1@aol.com)  
<http://www.deafchildren.org/>

DB-LINK, The National Information Clearinghouse on Children who are Deaf-Blind.  
(Also lists 307.11 State grantees Deaf-Blind projects)  
Teaching Research  
345 N. Monmouth Ave.  
Monmouth, OR 97361  
phone: (800)438-9376 (voice), (800)854-7013 (TTY), FAX: (503)838-8150  
e-mail: [dblink@tr.wou.edu](mailto:dblink@tr.wou.edu)  
<http://www.tr.wou.edu/dblink/index.htm>

Deafblindness Web Resource  
<http://www.deafblind.co.uk>  
Includes terminology, communication, journals, periodicals

The Family Centre  
86 Cleveland Road  
Ealing, London W13 OHE  
United Kingdom  
phone: (01)81 991-0513, FAX: (01)81 810-5298

The Foundation for Fighting Blindness  
Executive Plaza 1, Suite 800  
11350 McCormick Road  
Hunt Valley, MD 21031-1014  
phone: (800)683-5555 (voice), (800)683-5551 (TTY), FAX: (410) 771-9470  
<http://www.blindness.org>

John Tracy Clinic  
806 West Adams Boulevard  
Los Angeles, CA 90007  
phone: (213) 748-5481 (v) , (213) 747-2924 (TTY), (800) 522-4582, FAX: (213) 749-1651  
<http://www.johntracyclinic.org>

Helen Keller National Center for Deaf-Blind Youths and Adults (HKNC)  
111 Middle Neck Road  
Sand Points, NY 11050-1299  
phone: (516) 944-8900 (v), (516) 944-8637 (TTY), FAX: (516) 944-7302  
e-mail: [abigailp@aol.com](mailto:abigailp@aol.com)  
<http://www.helenkeller.org/national/index.htm>

HOUSE Ear Institute  
2100 West 3rd Street  
Los Angeles, CA 90057  
phone: (800)287-4763 (v, TTY) California, (800)352-8888 (v, TTY) all other 50 states  
<http://www.hei.org/>

Katlyn's HOPE  
303 South Elm Street  
Wellington, KS 67152  
phone: (316)326-6118, (877)677-HOPE – Toll-free for parents  
e-mail: [khope@idir.net](mailto:khope@idir.net)  
<http://www.idir.net/~khope/links.html>

National Association for Parents of the Visually Impaired, Inc. (NAPVI)  
P.O. BOX 317  
Watertown, MA 02471  
phone: (617) 972-7441, (800) 562-6265, FAX: (617) 972-7444  
<http://www.spedex.com/napvi>

National Coalition on Deaf-Blindness (NCDB)  
175 North Beacon Street  
Watertown, MA 02472  
phone: (617) 972-7347, FAX: (617) 923-8076  
e-mail: [davies@perkins.pvt.k12.ma.us](mailto:davies@perkins.pvt.k12.ma.us)

National Cued Speech Center  
PO Box 31345  
Raleigh, NC 27622-1345  
phone: (919) 828-1218 (v, TTY)  
e-mail: [ncsa@aol.com](mailto:ncsa@aol.com)

National Family Association for Deaf-Blind (NFADB)  
111 Middle Neck Road  
Sand Point, NY 11050  
phone: (800) 255-0411, FAX: (516) 944-5984  
e-mail: [pajomac@aol.com](mailto:pajomac@aol.com)

National Technical Assistance Consortium for Children and Young Adults who are Deaf-Blind (NTAC)

345 N. Monmouth

Monmouth OR 97361

phone: (503) 838-8096 (v) (OR), (503) 838-9623 (TTY) (OR), FAX: (503) 838-8150 (OR), (516) 944-8900 x 273 (v) (NY), (516) 883-9059 (TTY) (NY), FAX: (516) 883-9060 (NY)

<http://www.tr.wou.edu.ntac>

NOR-CAL Center on Deafness

Supported Employment Program

1820 Tribute Rd, Suite A

Sacramento, CA 98515

Phone: (916) 921-1045 (v, TTY), FAX: (916) 921-1177

Partners for Progress, Pre-College National Mission Program

Gallaudet University

800 Florida Ave., NE

Washington, DC 20002, FAX: (202) 651-5435

<http://www.gallaudet.edu/~parweb/pfp.html>

SENSE The National Deafblind and Rubella Association

11-13 Clifton Terrace

Finsbury Park, London N435R

United Kingdom

phone: (01)71 272-7774, FAX: (01)71 272-6012

e-mail: [enquiries@sense.org.uk](mailto:enquiries@sense.org.uk)

<http://www.vois.org.uk/sense/>

U.S. Department of Education Office of Special Education Programs, Division of Education Services For Children with Deaf-Blindness Program

Switzer Bldg., Room 4613

330 C Street, SW

Washington, DC 20202-2734

phone: (202)205-8165, FAX: (202)205-8971

e-mail: [charles\\_freeman@ed.gov](mailto:charles_freeman@ed.gov)

## **\*\*DEVELOPMENTAL and SENSORY INTEGRATION RESOURCES\*\***

Sandra L.H. Davenport, M.D.

Sensory Genetics/Neuro-Development

5801 Southwood Drive

Bloomington, MN 55437-1739

phone: (952)831-5522 (v, TTY), FAX: (952)831-0381

e-mail: [slhdaven@tc.umn.edu](mailto:slhdaven@tc.umn.edu)

The HANDLE Institute  
1530 Eastlake Avenue E., Suite 100  
Seattle, WA 98102  
phone: (206) 860-2665, FAX: (206) 860-3505  
e-mail: [support@handle.org](mailto:support@handle.org)  
<http://www.handle.org>

National Academy for Child Development  
P.O. BOX 380  
Huntsville, UT 84317  
phone: (801) 621-8606, FAX: (801) 621-8389  
e-mail: [nacdinfo@nacd.org](mailto:nacdinfo@nacd.org)  
<http://www.nacd.org/>

Sensory Integration International (SII)  
The Ayres Clinic  
1514 Cabrillo Avenue  
Torrance, CA 90501  
phone: (310) 320-2335, FAX: (310) 320-9982  
e-mail: [sensoryint@earthlink.net](mailto:sensoryint@earthlink.net)  
<http://home.earthlink.net/~sensoryint/>

**\*\*DISABILITY RESOURCES\*\***

Beach Center on Families and Disabilities  
3111 Haworth Hall  
University of Kansas  
Lawrence, KS 66045-7516  
phone: (913) 864-7600 (v), (913) 864-7600 (TTY) FAX: (913) 864-7605  
email: [beach@dole.lsi.ukans.edu](mailto:beach@dole.lsi.ukans.edu)  
<http://www.lsi.ukans.edu/beach/beachhp.htm>

ERIC Educational Resources Information Center  
Clearinghouse on Disabilities and Gifted Children  
The Council for Exceptional Children  
1920 Association Drive  
Reston, VA 20191  
phone: (800)328-0272 (v) (703) 264-9449 (TTY)  
e-mail: [ericec@cec.sped.org](mailto:ericec@cec.sped.org)  
<http://www.cec.sped.org/ericec/digests.htm>

ESCO Hearing Aid Insurance  
Ear Service Corporation  
3650 Annapolis Lane, Suite 107  
Plymouth, MN 55447  
phone: (800) 992-3726  
e-mail: [info@earserv.com](mailto:info@earserv.com)  
<http://www.earserv.com>

MEDICAID Waivers - Katie Beckett Waivers  
Health Care Financing Administration (HCFA)  
Within the Department of Health and Human Services (DHHS)  
Family Voices National Office  
PO Box 769  
Algodones, NM 87001  
Toll-free (888) 835-5669  
Voice (505) 867-2368  
FAX (505) 867-6517  
<http://www.familyvoices.org/~vrosales/KATIEBEC.html>

NICHCY (National Information Center for Children and Youth with Disabilities)  
P.O. BOX 1492  
Washington, DC 20013-1492  
phone: (800) 695-0285 (v, TTY), 800-695-0285 (v, TTY) FAX: (202)884-8441  
<http://www.nichcy.org>  
(Among resources are: State chapters of parent and disability groups, Protection and Advocacy Agencies per State, Parent Training and Information Projects, and IDEA 1997)

NORD (National Organization for Rare Disorders)  
100 Rt. 37, P.O. BOX 8923  
New Fairfield, CT 06812-8923  
phone: (203) 746-6518 (v), (203) 746-6927 (TTY), (800) 999-6673 FAX: (203) 746-6841  
e-mail: [orphan@nord-rdb.com](mailto:orphan@nord-rdb.com)  
<http://www.nord-rdb.com/orphan>

Special Olympics  
1325 G. Street NW, Suite 500  
Washington, DC 20005  
phone: (202) 628-3630, FAX: (202) 824-0200  
<http://www.specialolympics.org/>

TASH Disability Advocacy Worldwide  
29 W. Susquehanna Ave. Baltimore, MD 21204  
phone: (410) 828-8274, FAX: (410) 828-6706  
e-mail: [nweiss@tash.org](mailto:nweiss@tash.org)  
<http://www.tash.org/>

Wide Smiles Web Page  
<http://www.widesmiles.org/WS-58.html>

Wright's Law - The Special Ed Advocate  
Peter W.D. Wright, Esq.  
P.O.BOX 1008  
Deltaville, VA 23043  
phone: (804)257-0857  
e-mail: [webmaster@wrightslaw.com](mailto:webmaster@wrightslaw.com)  
<http://www.wrightslaw.com/>



**\*\*SPEECH, LANGUAGE AND COMMUNICATION RESOURCES\*\***

American Speech - Language and Hearing Association  
10801 Rockville Pike  
Rockville, Maryland, 20852  
phone: (301) 897-5700 (v), (301) 897-0157 (TTY) FAX: (301) 571-0457  
<http://www.asha.org/contents.htm>

APRAXIA-Kids website  
<http://www.jump.net/~gmikel/apraxia/>

Listenup Web Page Kay Powell  
[listenup@texas.net](mailto:listenup@texas.net)  
<http://members.tripod.com/~listenup/>

The Little Room  
"Space and Self" book by Lilli Nielsen  
Sikon Publisher  
<http://members.aol.com/LSWebDesgn/LNroom.html>

Mayer-Johnson (Picture Communication Symbols)  
PO BOX 1579  
Solana Beach, California 92075  
(800) 588-4548, FAX: (858) 550-0449  
<http://www.mayerjohnson.com/>