9th International

CHARGE Syndrome Conference

July 24-26, 2009

Indian Lakes Resort

Bloomingdale, IL

CONFERENCE PROGRAM AND HANDOUTS

The CHARGE Syndrome Foundation, Inc.

www.chargesyndrome.org

THURSDAY - July 23, 2009

8:00 – 5:30	**1ST CHARGE Syndrome Conference for Professionals: (Conference Center: Trillium 2)**
12:00 – 6:00	**9th International CHARGE Syndrome Conference:** **Conference Registration and Childcare Registration in the Hotel Lobby**
7:00 – 9:00	**9th International CHARGE Syndrome Conference:** **Reception for Families and Professionals (Conference Center: Trillium 1)** *[Light snacks, soft drinks & cash bar]*

FRIDAY - July 24th, 2009

6:30 – 6:00	**Conference Registration & Sales Center open**
7:00 – 9:00	**Breakfast—In Trillium Ballrooms**
6:30 – 8:30	**Registration for Camp Discovery & Camp Explorer – Conference Center**
8:00 – 12:00	**Camp Discovery (Downstairs in Conference Center) & Camp Explorer (Downstairs in hotel) Morning session**
9:00 – 6:00	**Vendor Fair—Fireplace & Falling Waters Lounges**
9:00 – 9:45	**Welcome and opening remarks—Trillium Ballrooms**
9:45 – 10:15	**Special Guest Presentation: Bryan D. Hall – "How the CHARGE Association (Syndrome) Was Identified and Expanded: A Personal Scenario"**
10:15 – 10:45	**Break**
10:45 – 11:45	**Breakout Session #1—See below**
11:45 – 1:00	**Lunch—In Trillium Ballrooms**
12:15 – 3:30	**Sib Shop #1 – Sorrell II**
12:45 – 5:45	**Camp Discovery & Camp Explorer afternoon session**
1:00 – 2:00	**Breakout Session #2—See below**
2:00 – 2:10	**Break**
2:10 – 3:10	**Breakout Session #3—See below**
3:10 – 3:30	**Break**
3:30 – 5:30	**Breakout Session #4—Interact at Information Central**
4:00 – 6:00	**Registration for Camp Discovery & Camp Explorer – Conference Center (for children not yet registered)**
7:00	**1st Annual CHARGE Syndrome "CONFERENCE IDOL" Talent Extravaganza – Trillium Ballrooms**

	Trillium I	Trillium II	Trillium III	Trillium IV	Byrne	Jensen	Cyperus IV
Breakout Session #1 10:45 – 11:45		**Bone Anchored Implants: Baha**	**Minding the Gap**	**CHARGE 101 – New Families**	**Grandparent Meeting**	**Father's Forum**	**CHARGE into Vocational Training (Young adults with CHARGE only !!)**
Breakout Session #2 1:00 – 2:00		**Cochlear Implants for children with CHARGE syndrome**	**Genetic Diagnosis of CHARGE syndrome**	**CHARGE 102 – New Families**	**Sib Shop # 1** **In Sorrell II from 12:15 – 3:30**		**Young Adults with CHARGE syndrome** **LOUNGE**
Breakout Session #3 2:10 – 3:10	**Resources for Physical Education, Sports & Fitness for children with CHARGE**	**Impact of Cochlear implants in CHARGE syndrome – Preliminary findings**	**Special Needs Future Planning: Trusts, Government Benefits, Guardianship, etc.**	**CHARGE 103 – New Families**			
Breakout Session #4 3:30 – 5:30	**Interact at INFORMATION CENTRAL** **Conference presenters will be available to talk to conference participants.** **17 Poster Presentations will be available.** **FUN Chi demonstration** **Researchers will be available to talk about current projects.**						

SATURDAY - July 25th, 2009

7:00 – 4:00	**Conference Registration & Sales Center open**
7:00 – 9:00	**Breakfast—In Trillium Ballrooms**
8:00 – 12:00	**Camp Discovery & Camp Explorer morning session**
9:00 – 9:10	**Welcome and housekeeping**
9:10 – 9:45	**Foundation general membership meeting—Updates and Awards**
9:45 – 10:15	**Committee Reports**
10:15 – 10:45	**Break**
10:45 – 11:45	**Breakout Session #5—See below**
11:45 – 1:00	**Lunch—In Trillium Ballrooms**
12:45 – 5:00	**Camp Discovery & Camp Explorer afternoon session**
1:00 – 2:00	**Breakout Session #6—See below**
2:00 – 2:10	**Break**
2:10 – 3:10	**Breakout Session #7—See below**
3:10 – 3:40	**Break**
3:40 – 4:40	**Breakout Session #8—See below**
6:00 – 9:30	**Dinner, carnival and silent auction – Trillium Ballrooms**

	Trillium I	Trillium II	Trillium III	Trillium IV	Cypress IV	Sorrell II
Breakout Session #5 10:45 – 11:45		**Smell and puberty in CHARGE syndrome**	**Parenting children with CHARGE syndrome: Perils & Promise**	**Constructing Meaningful Conversations**	**Going to college: What do I need to know? (Young adults with CHARGE only !!)**	**Sib Shop #2 8:45 – 12:00**
Breakout Session #6 1:00 – 2:00		**The "R" & "G" in CHARGE**	**Person Centered Planning as an adjunct to psychoeducational evaluation for individuals with CHARGE**	**Balance: What is it and how does it work?**	**Young Adults with CHARGE syndrome**	
Breakout Session #7 2:10 – 3:10	**What's there to stress about? The case of CHARGE**	**The NICU experience: It's impact & implications**	**Successful teaching and key components for use in educational and community settings**	**Taking charge of your life, developing a positive vision for employment**	**LOUNGE**	**Sib Shop #2 1:00 – 4:15**
Breakout Session #8 3:40 – 4:40	**Look at me now: The impact of early inclusion in outcomes**	**Anesthesia management of Individuals with CHARGE syndrome**	**Nanny 411: Top 10 strategies for promoting positive behavior in children with CHARGE**	**Vision issues for people with CHARGE syndrome**		
EVENING 6:00 – 9:30	**Carnival, Dinner and Silent Auction (Trillium Ballrooms)**					

SUNDAY - July 26th, 2009

7:00 – 9:00	**Breakfast—In Trillium Ballrooms**
8:30 – 12:30	**Camp Discovery & Camp Explorer morning session**
9:00 – 9:30	**Welcome and housekeeping**
9:30 – 10:00	**Special Presentation by Cynthia Antaya**
10:00 – 10:30	**Tribute to Marion Norbury**
10:30 – 11:00	**Break**
11:00 – 11:15	**Message from the National Coalition on Deafblindness**
11:15 – 11:45	**Conference farewell message from Michelle Westmaas**
11:45 – 11:55	**Deep Thoughts by David Brown**
11:55 – 12:00	**2009 Conference Farewell**

9th International CHARGE Syndrome Conference

GUIDE TO PLATFORM & POSTER PRESENTATIONS WITH ABSTRACTS

Abstracts of Friday Breakout Sessions

SE=Sensory FS=Family Support NF=New Families TA=Transition to adulthood

MG=Medical/genetic ED=Education, communication, development, and behavior GI=General interest

10:45-11:45 BREAKOUT SESSION #1

II SE Lisa Christensen: Bone anchored Implants: Baha

Presentation Abstract: Bone anchored implants are becoming more popular as a treatment for inoperable conductive and mixed hearing loss in children. This presentation will discuss the Baha system manufactured by Cochlear Americas, which at the present time is the only FDA approved bone anchored implant in the United States. Candidacy and criteria for implanting in children, complications, currentavailable processors, and how a bone anchored implant works.

III GI Gail Deuce & David Brown: Minding the gap

Presentation Abstract: This session will be based on interviews undertaken by David and Gail with parents of young people/ adults to consider: (1)Whether or not parents have talked to their child about their having CHARGE or about them having certain CHARGE anomalies; (2) If they did this what made them decide to do it and when did they do it? If not, why not?; (3) If they did this, how was it done and were any other persons involved (e.g. a professional, another family member, another person with CHARGE); (4) What worked and what didn't work in this process?; (5) Has it been an ongoing process and what determined the timing and nature of the steps in the process?; (6) How has this issue been addressed to meet the communicative and cognitive needs of the individual?

IV NF CHARGE 101

Meg Hefner: CHARGE syndrome: diagnosis and features

Presentation Abstract: A brief history of CHARGE: where did the name come from? How often does it happen? How often does it happen again? Diagnosis: What else can look like CHARGE? What about the DNA test? What are the features? Which ones are helpful in diagnosis and which ones are most important in management? What are the consequences of the features of CHARGE?

Kim Blake: Anesthesia complications in CHARGE syndrome

Presentation Abstract: Why is anesthesia important? How many surgeries is my child likely to have? Why is it important to combine procedures with one anesthesia?

Byrne FS Leonard Stanger (Moderator): Grandparent Meeting

Purpose of the Grandparent Meeting: CHARGE syndrome affects every member of the family. This meeting is only for grandparents. It is an opportunity for those who have a grandchild with CHARGE to meet and discuss whatever is of importance to those in attendance. By request of grandparents at earlier CHARGE Conferences, the Grandparent Meeting is held at the beginning of the Conference so that the grandparents can meet each other early and socialize and share information for the next two-and-half days. This informal discussion session will be led by Leonard Stanger who is very familiar with the issues of importance to those who have a grandchild with CHARGE.

Jensen FS John Reiman & Tim Hartshorne (Moderators): Father's Forum

Purpose of the Fathers' Forum: CHARGE syndrome affects every member of the family. This meeting is only for fathers who have a child with CHARGE. It is their opportunity to have an open discussion and share experiences with other fathers who have had to deal with the issues associated with CHARGE syndrome. The moderators of the Fathers' Forum are two fathers with extensive personal and professional experience with special needs children and adults.

Cyperus IV TA Wendy Bridgeo & Christa Giklhorn: CHARGE into vocational training *[Adults with CHARGE only]*

Presentation Abstract: This presentation will highlight the importance of providing students with significant disabilities with a preparatory vocational curriculum that starts with the end in mind. Functional and creative vocational education and training enhances vocational exploration, skill development and assessment. Seasoned vocational educators from the Deafblind Program at Perkins School for the Blind will share ideas, activities and strategies that have been implemented and refined in school and community-based vocational settings.

1:00-2:00 BREAKOUT SESSION #2

II SE Beth Tournis: Cochlear implants for children with CHARGE syndrome

Presentation Abstract: This presentation will describe the procedures associated cochlear implantation the criteria for candidacy for implantation, the audiologic information needed; expectations and success; and post-surgical services. Case studies will be presented.

III MG Conny van Ravenwaaij-Arts & Jorieke Bergman: Genetic testing in CHARGE syndrome

Presentation Abstract: he gene involved in CHARGE syndrome was identified in 2004. Mutations in this gene, the CHD7 gene, are found in the majority of CHARGE syndrome patients. Different techniques are used to analyze this large gene. Sequence analysis is the method of first choice and will identify mutations in the CHD7 gene. Two other techniques, MLPA and array CGH, can be used to find deletions or duplications of the CHD7 gene.

IV NF Sandra Davenport: CHARGE 102: Sensory loss: how does that change early development?

Presentation Abstract: Development: How do all those features of CHARGE influence development? What are sensory deficits? What does DeafBlind mean? What does balance have to do with it? What about weakness? How well can my child do?

2:10-3:10 BREAKOUT SESSION #3

I ED Lauren Lieberman: Resources for physical education, sports & fitness for children with CHARGE syndrome

Presentation Abstract: Children with CHARGE Syndrome are often medically fragile, and have ongoing health issues. Often times parents, teachers and physicians do not believe they can be physically active within their schools and communities. The lack of physical involvement can have detrimental effects on their physical, social and emotional growth. This presentation will give a variety of resources so teachers, caregivers and physicians can help them access a physically active and social lifestyle regardless of their medical involvement.

II SE Susan Bashinski & Kathleen Stermel Thomas: Impact of cochlear implants in CHARGE syndrome: preliminary findings

Presentation Abstract: Presenters will share preliminary findings from their ongoing research study investigating outcomes for children with deaf-blindness, who have received a cochlear implant. Participants in this study are diverse; the second largest group, by etiology, is children with CHARGE syndrome. In addition to sharing research findings they have to date, researchers will present suggestions parents might utilize to promote their children's communication development and listening skills. Sound inventories for home, school, and community environments, developed by the researchers, will be discussed.

III FS Brian Rubin: Special need future planning: trusts, government benefits, guardianship, etc.

Presentation Abstract: Mr. Rubin is an attorney whose practice is devoted to services for individuals with special needs. He will present on the topics listed in the presentation title as well as others.

IV NF Robert Last: CHARGE 103: Setting the scene – communication with babies and toddlers

Presentation Abstract: "For children who have CHARGE syndrome there is strong evidence that diagnosis and intervention before six months of age significantly improves outcomes in communication, behaviour and learning" Jan van Dijk and Arno de Kort. This presentation explores what communication means, the early considerations in communication with babies and toddlers, the strategies in acquiring communication skills and exploring outcomes.

3:30-5:30 BREAKOUT SESSION #4: Interact at INFORMATION CENTRAL

DEMONSTRATION

FS Sharon Barrey Grassick:

FUN-Chi (see handout at the end of the section for platform presentations)

Demonstration Abstract: Stress! We all have it at times; individuals with CHARGE often have too much of it. So, how to destress?Challenging behaviours, to varying degrees, can emerge in children withCHARGE; stress is a probable key cause. There are no magical answers to reduce stress and self-regulate behaviours. Let's think outside the square - to the circle…Yin Yang or Tai Chi. This presentation willdemonstrate a fun approach to exercise and stress management - called *FUN Chi*!

POSTER PRESENTATIONS

TX Nikki Anderson, Shawn Herrick, Mikal, Folstaedt, Jaymie Barker & Amber Hamilton *[Spalding University]*

Parental attitudes toward the use of neuromuscular electrical stimulation (NMES) for the treatment of dysphagia in children with CHARGE syndrome: a pilot study

Poster Abstract: The purpose of this pilot study is to determine the attitudes of parents toward Neuromuscular Electrical Stimulation (NMES), trade name VitalStim, as an intervention to treat swallowing difficulties (dysphagia) in children who have CHARGE Syndrome. A questionnaire has been completed by parents and an unstructured phone interview will be conducted if additional information is needed. The results of this study are currently being analyzed and will be completed by June of 2009.

ED Alana Zambone, Susan Bashinski & Melissa Darrow Englemann *[East Carolina University]*

Preparation/certification of professionals in the area of deaf-blindness

Poster Abstract: East Carolina University's (ECU) new Graduate Certificate in Deafblindness and Intervener Certificate effectively prepare personnel to meet the new national standards and attain national accreditation through distance education. The research and development process for the national teacher and intervener standards, accreditation, and ECU's courses and practica through distance education will be shared. The ways in which ECU's certificate programs apply principles of Universal Design to accommodate participants' diverse knowledge, experience, and learning styles are described.

TX Kim Blake & Jill MacCuspie *[Dalhousie University]*

Botox for dysphagia in CHARGE syndrome: a case study

Poster Abstract: Our patient's neonatal surgeries included Tracheooesophageal fistula repair, PDA and vascular ring ligation. He remained ventilation dependent because of excessive oral secretions and was awaiting tracheostomy. At 2 months of age, botox was injected into two pairs of his salivary glands and within 24 hours he was extubated. Five months later increasing oral secretions and aspiration pneumonia resulted in our patient requiring mechanical ventilation. Botox was successfully used again to reduce oral secretions.

POSTER PRESENTATIONS *(continued)*

ED Donna Consacro, Linda Balderson & Julie Brandrup *[TREDS – Tennessee Deaf-Blind Project]*

Cued speech: What is it? Would it be useful to our family?

Poster Abstract: This presentation will explore Cued Speech's effectiveness as a tool for receptive and expressive language development for children. Participants will gain an understanding of what Cued Speech is, how it might be used alone or to supplement sign language, the speed with which it can be acquired by parents and the subsequent breadth of language exposure it offers to their children. Possible physical reasons that might prevent the use of Cued Speech will also be explored.

MG Maryann Girardi *[Perkins School for the Blind]*

CHARGE feet: fact or fiction (Part II)

Poster Abstract: This poster will present the results from the study of foot anomalies at the 2007 conference

ED Nancy Salem-Hartshorne *[Central Michigan University]*

Person centered planning as an adjunct to psychoeducational evaluation for individuals with CHARGE

Poster Abstract: This poster will describe useful Person-Centered Planning techniques the presenter has used to assist families and professionals to come together to meet the needs of students with CHARGE syndrome. In both situations, the family members and professionals were at odds prior to the evaluation and Person-Centered Plan. The presentation will be highlighted with photographs and descriptions of the specific plans created, the stories behind the plans, and the positive outcomes for the students. The families of these students may be available to answer questions as well.

ED Diane Haynes *[Kentucky Deaf-Blind Project]*

Constructing meaningful conversations

Poster Abstract: The basic premise of this presentation is that any interaction between human beings is the basis for a conversation. Successful interactions / conversations depend on our ability as communication partners to perform the steps necessary to complete a basic dyadic interaction. Steps encompass accurate identification of receptive functions and receptive forms that reflect knowledge of a partner's functioning within all seven sensory systems. The presenter will share strategies for developing an intervention plan.

ED Emily King Miller[1], Lori Swanson[2], Nancy Steele[3], Ilsa Schwarz[1], Sara Thelin & James Thelin[1]
[1 University of Tennessee;2 University of Wisconsin-River Falls, 3 National Consortium forDeaf-Blindness]

Communicative rate, forms, and functions in CHARGE syndrome

Poster Abstract: A method of analyzing communicative rate, forms (pre-symbolic or symbolic), and functions (purposes) was developed to describe communication development in individuals with CHARGE. Video-taped communication samples were analyzed of 21 individuals with CHARGE syndrome (1:8 to 20:5 years: months). The analyses provide a means for describing specific communication abilities for an individual and for planning therapy to develop communication abilities. The results of the analyses were also used to describe the communication of the participants as a group.

POSTER PRESENTATIONS *(continued)*

SE Sarah Krivenki & James Thelin *[University of Tennessee]*

Vestibulo- and cervico-ocular reflexes in CHARGE syndrome

Poster Abstract In CHARGE syndrome, critical structures of the VOR and COR (which stabilize visual images with head motion) are often abnormal: incomplete or missing semicircular canals and ocular colobomas. A VNG procedure was used to measure the VOR and COR in subjects who could only provide minimal cooperation for brief periods. In subjects with CHARGE syndrome, horizontal VOR was absent for 29 of 29 subjects and horizontal COR was absent for 12 of 13 subjects.

ED Martha Majors & Sharon Stelzer *[Perkins School for the Blind]*

A multi-media tour of classrooms with students with CHARGE syndrome

Poster Abstract: This session will include a photo board of the key educaetional components of the Deafblind Program at Perkins School for the Blind; this will be a visual presentation to support discussion related to families questions about the Program; Martha Majors and Sharon Stelzer will be at the Poster Session.

ED Lori Swanson *[University of Wisconsin- River Falls] & Shawn Herrick {Spalding University]*

Use of manual signs promotes speech: a case study

Poster Abstract: This case study describes the development of oral language skills in a boy with CHARGE syndrome. Fluent manual sign and speech input were provided to this child at an early age. He relied on manual signs for language production while his tracheostomy was in place. When his trach was removed, he made any easy transition to speech. The importance of early intervention to facilitate language development will be discussed.

ED-MG Kasee K. Stratton and Timothy S. Hartshorne *[Central Michigan University]*

Identification of pain in CHARGE syndrome

Poster Abstract: Parents and professionals working with children with CHARGE have long suspected that these children experience considerable pain that is related to some of their behavioral difficulties. Pain may result from some of the physical anomalies of CHARGE and from multiple, extensive surgeries, as well as on-going health issues such as ear infections and headaches. To better understand the relationship between pain and behavior, we have proposed a method for measuring pain in CHARGE

MG Timothy S. Hartshorne & Kasee K. Stratton *[Central Michigan University]*

Prevalence of Genetic Testing in CHARGE Syndrome

GI Simon Howard *[CHARGE Family Support Group, UK]*

The CHARGE Family Support Group in the United Kingdom

GI Gail Deuce *[Sense, UK]*

"Sense for deaf-blind people" – an organization in the UK and Europe

POSTER PRESENTATIONS *(continued)*

ED Beth Marie Shaver Kennedy *[Deafblind Central],* Barbara Purvis *[National Consortium for Deaf-Blindness*], & Nicole Bruce *[Deafblind Central]*

Using person-centered planning for students with low-incidence disabilities who are transitioning from school

Poster Abstract: DB Central and the National Consortium on Deaf-Blindness are working in collaboration with Michigan School for the Deaf on an initiative to improve post-secondary outcomes for students with low incidence disabilities. Participants involved in the initiative are trained in the philosophy of person-centered planning (PCP) and the many ways in which the PCP can be used to enhance post-secondary transition planning. Participants are also encouraged, throughout the process, to function as a team, sharing common goals and desired outcomes. To increase local capacity, efforts are also made to train local person-centered planning facilitators and to initiate a person-centered planning community of practice.

TA Christa Giklhorn & Wendy Bridgeo *[Perkins School for the Blind]*

Vocational Education and Training: Preparing Students for Meaningful Adult Lives Through School and Transition Experiences

Poster Abstract: This presentation will highlight the importance of providing students with significant disabilities with a preparatory vocational curriculum that starts with the end in mind. Functional and creative vocational education and training enhances vocational exploration, skill development and assessment. Seasoned vocational educators from the Deafblind Program at Perkins School for the Blind will share ideas, activities and strategies that have been implemented and refined in school and community-based vocational settings.

Abstracts of Saturday Breakout Sessions

SE=Sensory FS=Family Support NF=New Families TA=Transition to adulthood

MG=Medical/genetic ED=Education, communication, development, and behavior GI=General interest

10:45-11:45 BREAKOUT SESSION #5

II MG Jorieke Bergman & Conny van Ravenswaaij-Arts: **Smell and puberty in CHARGE syndrome**

Presentation Abstract: Smell deficiency and delayed/absent puberty often occur in CHARGE syndrome, but few studies have looked at these features in adolescent patients. Therefore, we studied smell and puberty development in 22 adolescent CHARGE patients and showed that puberty and smell problems always co-occur. Therefore, a smell test can possibly predict whether puberty will occur spontaneously or not. This will prevent delay of hormonal pubertal induction, resulting in an age-appropriate puberty in smell deficient CHARGE patients.

III GI Timothy S. Hartshorne: **Parenting children with CHARGE syndrome: perils & promise**

Presentation Abstract: Parenting children with CHARGE is a huge challenge, and parenting methods for "regular" kids don't seem to apply. This presentation suggests overall strategies for parenting children with CHARGE. The presentation topics include typical characteristics, parent experiences, courage, executive functions, family meetings, encouragement, behavior & misbehavior, self-regulation, and punishment & rewards.

IV ED Diane Haynes: **Constructing meaningful conversations**

Presentation Abstract: The basic premise of this presentation is that any interaction between human beings is the basis for a conversation. Successful interactions / conversations depend on our ability as communication partners to perform the steps necessary to complete a basic dyadic interaction. Steps encompass accurate identification of receptive functions and receptive forms that reflect knowledge of a partner's functioning within all seven sensory systems. The presenter will share strategies for developing an intervention plan.

Cyperus IV TA Beth Jordan & Belinda Arnell: **Going to college: What do I need to know?** *[Adults with CHARGE only]*

Presentation Abstract: Applying to a college or training program after high school can be an overwhelming task. This session is designed to help a young adult consider what supports he or she might need during the college application process or when in a classroom on a campus. Participants will receive specially designed tool that provides a simple and easy way to determine the level and types of accommodations that a college offers a student with a vision and hearing impairment (e.g. interpreter services, notetakers, readers etc.).

1:00-2:00 BREAKOUT SESSION #6

II MG Jeremy Kirk: **The "R" and "G" in CHARGE**

Presentation Abstract: Both growth problems (failure to thrive, short stature, delayed/absent puberty) and also genital problems (micropenis/undescended testicles) are part of the original acronym of CHARGE. Whilst these problems are commonly seen in CHARGE, other more pressing medical and surgical problems often mean that they are not always recognized or treated appropriately. We have been collecting data on these problems from within our local clinic, and also national/international groups, in order to provide best practice.

III ED Nancy Salem Hartshorne: **PERSON CENTERED PLANNING AS AN ADJUNCT TO PSYCHOEDUCATIONAL EVALUATION FOR INDIVIDUALS WITH CHARGE**

Presentation Abstract: This presentation will describe useful Person-Centered Planning techniques the presenter has used to assist families and professionals to come together to meet the needs of students with CHARGE syndrome. In both situations, the family members and professionals were at odds prior to the evaluation and Person-Centered Plan. The presentation will be highlighted with photographs and descriptions of the specific plans created, the stories behind the plans, and the positive outcomes for the students. The families of these students may be available to answer questions as well.

Abstracts of Saturday Breakout Sessions *(continued)*

1:00-2:00 BREAKOUT SESSION #6 *(continued)*

IV SE Maryann Girardi: Balance: What is it and how does it work?

Presentation Abstract: The topic of balance is frequently discussed concerning children with CHARGE syndrome. This presentation will explore the many components of the human balance system including what they are, how they work, and how they interact with each other to enable humans to react to the challenges in their environment

2:10-3:10 BREAKOUT SESSION #7

I ED Kasee Stratton: Pain and stress: the case of CHARGE

Presentation Abstract: Interviews about stress were conducted with young adults. These interviews focused on stress experiences with specific attention paid to behavioral reactions. Themes based on education, family, friendships and employment would be presented. Additionally this presentation would also focus on pain experiences in CHARGE Syndrome. The experience of chronic pain would be explained along with research findings showing a lack of appropriate pain management in populations that cannot vocally communicate, such as infants and developmentally disabled. The focus would be to include pain in our understanding of CHARGE behaviors (such as outbursts, unexplainable crying, behavioral disruptions, etc) and ways to identify pain non-vocally that are reliable and valid. Without appropriate identification, pain management and treatment cannot be provided.

II ED Barbara Purvis: The NICU experience: its impact and implications

Presentation Abstract: Preterm infants complete their development in an environment markedly different than their mother's womb. The multi-sensory experiences in an intensive care nursery impact immature systems in ways that interfere with typical prenatal development. Regardless of whether they are born early, children with CHARGE Syndrome often spend extended time in the NICU, encountering experiences difficult for their compromised sensory systems to handle. This session examines implications of the NICU experience for both infants and families.

III ED Martha Majors & Sharon Stelzer: Successful Teaching Strategies and Key Components for use in Educational and Community Settings

Presentation Abstract: This presentation is focused on teaching strategies and other factors that are critical in both educational and community settings for individuals with CHARGE syndrome. Some of the specific topics include sensory loss, communication, curriculum, teaching strategies, and environments.The information for this presentation was gained from the Deafblind Program at Perkins School for the Blind as well as national and international consultation with school districts and families regarding educational strategies and communication.

IV TA Michael Fagbemi & Joe McNulty: Taking charge of your life, developing a positive vision for employment

Presentation Abstract: Young adults with dual sensory impairments are not often given the opportunity to live and work in their communities of choice. This lack of opportunity further limits a young adult's exposure to new experiences and as a result impacts on their ability to socialize effectively live independently and develop work skills beneficial to employers. The focus on educational mandates and the pressures that are associated with it have had a profound effect on youth who are in need of independent living skills. One important component of living independently is work. We will discuss the national picture and briefly explore an employment model based upon a negotiated employment relationship between employer and job seeker that fits the needs of both..

3:30-5:30 BREAKOUT SESSION #8

I ED Five members of the Costello Family, Jennifer Miller & Donna Consacro: **Look at me now: the impact of early inclusion on outcomes**

Presentation Abstract: This panel presentation will showcase a young woman with CHARGE syndrome who has been successfully included in home, school and community throughout her life. The panel will include this young woman, her parents, siblings, and Tennessee Deafblind Project staff. As well as triumphs we will discuss barriers and strategies to overcome them. We plan to offer both practical advice as well as inspiration for families, their children and the people who work with them.

II MG Kim Blake: **Anesthesia management of individuals with CHARGE syndrome**

Presentation Abstract: Why is anesthesia important? How many surgeries is my child likely to have? Why is it important to combine procedures with one anesthesia?

III ED Laurie Denno & Deanna Rothbauer: NANNY 101: **Top 10 strategies for promoting positivie behavior in children with CHARGE**

Presentation Abstract: Learn proactive approaches to prevent behavior difficulties. It is much easier to prevent problem behavior than to change it once the child has learned it. Behavior analysis is first and foremost a teaching strategy that stresses the interaction of the family, the school and the social environment with children and their behavior. Learn the principles from a behavior analyst and how to implement the principles from a parent who has done it.

IV TX David Brown: **vision issues for people with CHARGE syndrome**

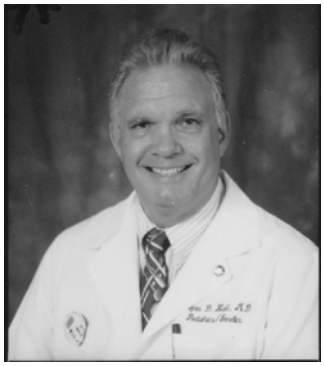
Presentation Abstract: Many of the anomalies found in CHARGE Syndrome carry significant implications for the development of functional vision skills. Some of these anomalies are specifically associated with eye defects, but many are not so are constantly overlooked or misunderstood as contributing to functional visiondifficulties. This session will examine all these anomalies, their behavioral implications, and strategies for intervention.

THE CHARGE SYNDROME FOUNDATION

SPECIAL GUEST PRESENTATION

FRIDAY
GENERAL ASSEMBLY 9:45 – 10:15
Trillium Ballrooms, Conference Center

How the CHARGE Association (Syndrome) Was Identified and Expanded: A Personal Scenario



BRYAN D. HALL, M.D., F.A.A.P.

In 1979, Dr. Hall was the first to recognize the pattern of anomalies that today is called CHARGE syndrome.

Presenter Information: *Dr. Bryan Hall* is Emeritus Professor of Pediatrics and Retired Chief of Genetics/Dysmorphology at the University of Kentucky, School of Medicine in Lexington, KY. His career as a pediatrician and geneticist has spanned 44 years. He is a member of many professional organizations and is the author of 129 articles in medical, genetic, and scientific journals. Though Dr. Hall's official status is "retired", he still is actively practicing in outreach clinics in Kentucky. It was his insightful observations over 30 years ago that led geneticists and physicians to recognize that what we now call CHARGE was not an unrelated collection of congenital anomalies that were treated as individual disorders. The significance of his observations and their value to every parent who has had a child with CHARGE has been very great.

Presentation Abstract: "In 1968 I saw my first child with choanal atresia and multiple anomalies. Between then and 1977 I saw 5 more cases, but was uneasy about the variability of the clinical features. However, the last case stimulated me to review all University of California at San Francisco medical records (1963-1978) that were coded under choanal atresia. This identified 11 additional cases. With this overwhelming evidence of 17 cases with a recurring pattern of anomalies I published my paper (Journal of Pediatrics 95:395-398) in September, 1979. The rest is an evolving history of challenges and triumph.

How the CHARGE Association (Syndrome) was Identified and Expanded: A Personal Scenario

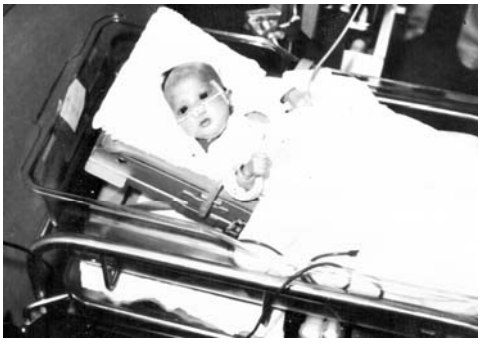
Bryan D. Hall, M.D., F.A.A.P.
Emeritus Professor of Pediatrics
Retired Chief of Genetics/Dysmorphology
University of Kentucky, Lexington, KY, USA

Early Years When I Began to Develop an Interest in Dysmorphology
(clinical genetics #1)

- 1963 – 1964: Pediatric externship each summer at Louisville Children's Hospital
  - I became particularly fascinated with rare/unique conditions.
- 1965 – 1968: Pediatric internship/residency
  - That interest continued to increase.

In 1968, I saw my first patient with choanal atresia, who also had other anomalies. No diagnosis was forthcoming…

…and I wondered why?



First Child I saw with Choanal Atresia and Multiple Anomalies

Early Years When I Began to Develop an Interest in Dysmorphology
(clinical genetics #2)

- 1968 – 1970: Spent two years in US Air Force at David Grant Hospital near Fairfield, California
  - I was allowed to attend UCSF genetic clinic every Wednesday.
    - These experiences initiated my career path toward dysmorphology.

Early Years When I Began to Develop an Interest in Dysmorphology
(clinical genetics #3)

- 1970-1972: Two-year dysmorphology training fellowship with David W. Smith at the University of Washington, Seattle
  - I began to realize the value of taking the rarest feature of an unknown multiple congenital anomaly syndrome and searching out syndromes that had that feature as part of their overall pattern.

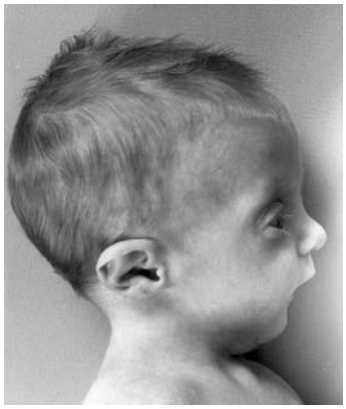
Mentor: David W. Smith (Father of Dysmorphology)

Time Frame when My Interest in Choanal Atresia and its Accompanying Anomalies Peaked

- 1968 – 1977: I came across five children (USAF and UCSF) with choanal atresia and multiple anomalies. Except for the choanal atresia, the additional anomalies were extremely variable in a single child, but repetitive in the group. The fifth patient had all the features found, in part, in the other four.
- **I knew I must study this.**

Time Frame when My Interest in Choanal Atresia and its Accompanying Anomalies Peaked

- 1977 – 1978: I reviewed UCSF medical records (time frame of 1963-1978) using choanal atresia as the ascertainment feature.
- **Eleven similar cases were identified!**



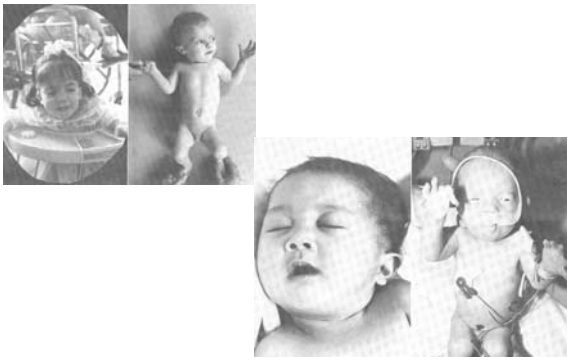
Child That Precipitated My Study

Original Table: Findings of the 17 Patients

Table. Associated abnormalities in 17 patients with choanal atresia*

Features	*Total*	*Percent*
Choanal atresia†	17/17	100
Mental retardation	11/11	100
Short stature (postnatal)	11/11	100
Small ears	13/17	76
Cardiac defects	12/17	71
Micrognathia	10/17	59
Microcephaly (postnatal)	8/14	57
Ocular coloboma	9/17	53
Hypogenitalism (males)	7/9	78
Deafness	6/15	40
Cleft palate	4/17	23
Small mouth	4/17	23
Short philtrum	4/17	23
High nasal bridge	4/17	23
Facial palsy	4/17	23
Short neck	4/17	23
Short fifth fingers	4/17	23
Tracheo-esophageal fistula	2/17	12

Collage of Four of the 17 Published Patients



Early Establishment of the CHARGE Association

- Oct. 1978: Hall presented 17 cases
  - ASHG, Vancouver
- Apr. 1979: Hittner, et al published 10 cases
  - J Pediar Oph Strabismus
- Sept. 1979: Hall published 17 cases
  - J Pediatr 95:395
- Aug. 1981: Pagon et al published 21 cases
  - J Pediatr 99:223
  - Coined the term, "CHARGE association"
- 1978 – current: Plus 1000 cases reported since

FEATURES OF ALL LITERATURE CASES (N62)

FEATURE	NUMBER	%
COLOBOMA	47/59	80%
HEART DISEASE	47/60	78%
ATRESIA CHOANAE	35/62	56%
RETARDED GROWTH	38/43	88%
RETARDED DEVELOPMENT	53/54	98%
GENITAL HYPOPLASIA	28/40 MALES	70%
EAR ANOMALIES	49/58	85%
FACIAL PALSY (UNILAT)	26/50	54%
SWALLOWING DIFFICULTY	11/28	39%
CLEFTS (MOSTLY PALATAL)	9/39	23%

Early Summary of the Most Common Features of CHARGE

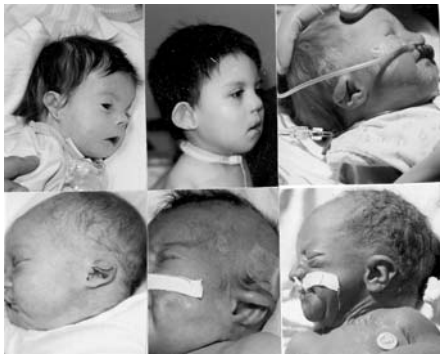
Infant with Bilateral Choanal Atresia and the Same Child as Seen at an Older Age

CHARGE Association Mnemonic Explanation

C = **C**oloboma (iris/retina)
H = **H**eart defects (PDA, conotruncal defects)
A = **A**tresia chonae
R = **R**etarded growth (postnatal) and development
G = **G**enital hypoplasia
E = **E**ar anomalies* (and deafness)

*Can be very distinctive (absent lobule, laterally protruding)

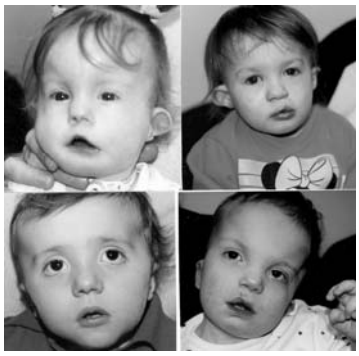
Various Ear Configurations Seen in My Patients with CHARGE



CHARGE Association: Other Major Features Noted in Early Reports

- Swallowing problems
  - Lethality factor
- Facial/cranial nerve palsies
  - Often requires long-term tracheostomy
- Cleft lip/palate
- Esophageal atresia/T-E fistula
- Distinctive face

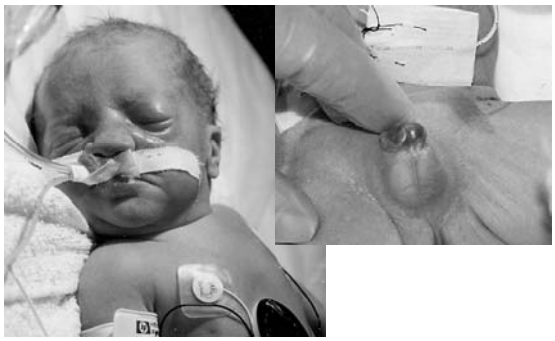
Facial Features in Four Children with CHARGE



CHARGE Association: More Recently Recognized Features

- Very Common Features:
  - Semicircular canal hypoplasia or aplasia
  - Central nervous system anomalies
- Moderately Common Features:
  - Limb defects
  - Renal anomalies
- Uncommon Features:
  - Vertebral defects
  - DiGeorge problems
  - Omphalocele
  - Hypospadias
  - Hernia
  - Web neck
  - Anal atresia

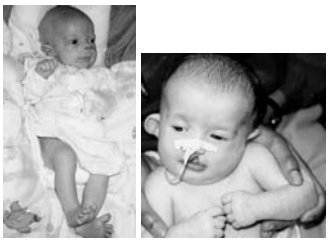
Collage of Newborn with CHARGE Showing Hypospadias



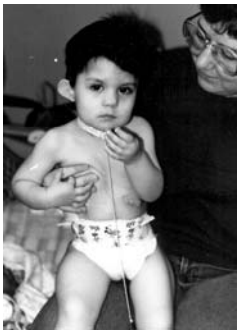
CHARGE Association: Potential Long-term Problems

- Failure-to-thrive (78%)
  - Often requires G-tube
- Postnatal proportionate short stature
  - Improves in some cases in late childhood
- Hearing/visual problems (89%) “deaf-blind”
- Marked delayed mobility
  - Poor coordination
- Seizures (25%)
- MR
  - Multisensory impairments make early testing problematic
  - Many aren’t ultimately retarded
- Delayed/inadequate puberty

Two Children with CHARGE with Cleft Lip and Nasal Stents



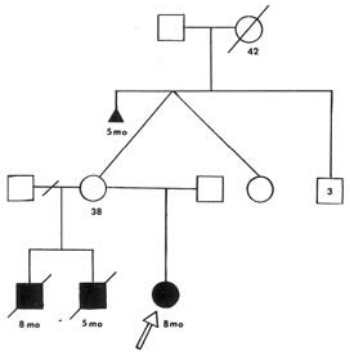
Young Boy with Trach and G-Tube with CHARGE



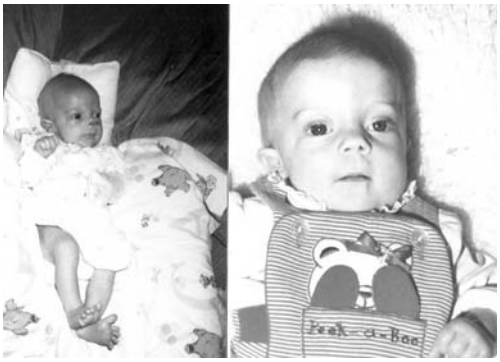
Search for the Etiology of CHARGE Syndrome

- After initial reports by Hall (1978), Hittner et al (1979) and Pagon et al (1981), some authors would occasionally report familial cases of CHARGE syndrome.
- In the late 1990’s, I saw an unaffected mother who had three affected children, by two different fathers. Chromosome studies were negative and no teratogens were identified. Until 2004, no etiology was established.

Pedigree of family with familial CHARGE



Female (Proband) in Family with Familial CHARGE



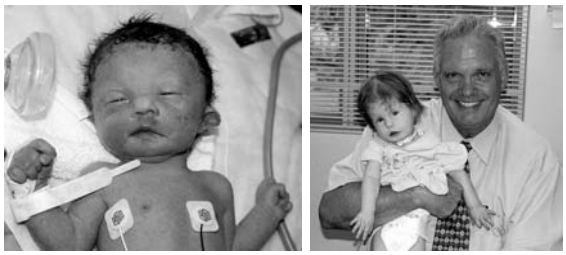
CHD7 Gene Identified as the Etiology of the CHARGE Syndrome in 2004

- Vissers et al. Nat Genet 36:955-957, 2004
- CHD7 gene on chromosome #8 (8q12.1)
  - C = Chromodomain
  - H = Helicase
  - D = DNA binding
  - 7 = ?
- Mutated CHD7 gene found in 66% of patients with CHARGE features

CHD7 Gene: Additional Information

- CHD7 is an autosomal dominant gene which can give extremely variable phenotype
- CHD7 gene mutations have little phenotype/ genotype correlation
- Some unaffected parents may carry the mutated CHD7 gene in their gonads and be at increased risk of having affected children with CHARGE syndrome

Newborn with CHARGE and Follow-up Photo of Same Child



Discovering the CHARGE syndrome has defined my professional life even though I spend less than 1% of my time with CHARGE-related issues.

The children with CHARGE syndrome and their families have taught me so much. I am humbled to be a part of the CHARGE family.

The identification of the CHD7 gene as the cause of the CHARGE syndrome is an important finding with great potential to enhance our diagnosis and treatment of individuals with the CHARGE syndrome.

My First Kentucky Patient with CHARGE.
What a Fellow!!

Dining at the CHARGE Meeting
in St. Louis with Adrian

9th International CHARGE Syndrome Conference

PLATFORM PRESENTATION ABSTRACTS & HANDOUTS

Sensory Information

FRIDAY
Breakout Session #1: 10:45 – 11:45 AM
Trillium Ballroom II, Conference Center

BONE ANCHORED IMPLANTS:
Baha

Lisa Christensen, Au.D.

Audiologist
Arkansas Children's Hospital
Little Rock, AR

Presenter Biography: Lisa Christensen is a pediatric audiologist at Arkansas Children's Hospital in Little Rock, Arkansas. Dr. Christensen serves as the Director of the Bone Anchored Implants Team. She is the current and founding president of the Arkansas Academy of Audiology and a participant in the Inaugural Future Leaders of Audiology Conference sponsored by the American Academy of Audiology. She is the recipient of the 2008 Audiology Award for the Arkansas Speech-Language Hearing Association. Dr. Christensen is also a consultant for Cochlear Americas. You may reach her at christensenlv@archildrens.org.

Presentation Abstract: Bone anchored implants are becoming more popular as a treatment for inoperable conductive and mixed hearing loss in children. This presentation will discuss the Baha system manufactured by Cochlear Americas, which at the present time is the only FDA approved bone anchored implant in the United States. Candidacy and criteria for implanting in children, complications, current available processors, and how a bone anchored implant works.

Bone Anchored Implants
Baha

Lisa Christensen, Au.D.
Arkansas Children's Hospital

The Baha System

The Baha System has been a well recognized hearing treatment for conductive and mixed hearing loss since 1977.

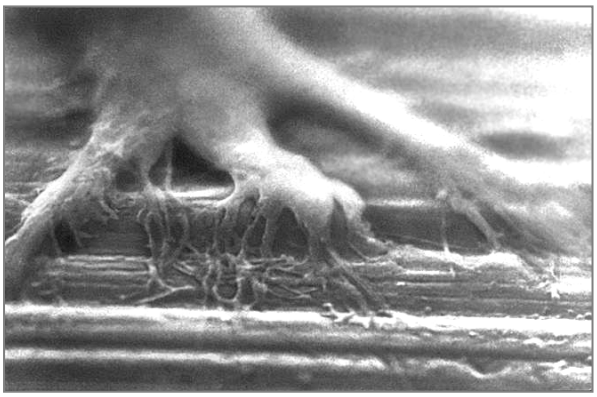
The Baha System is the only implanted hearing treatment that works through direct bone conduction.

Sound is conducted through the skull bone bypassing the outer and middle ear and stimulating the cochlea.

The Baha System is composed of three parts: a titanium implant, an external abutment, and a detachable sound processor.

Baha

Baha System:
Osseointegration



Baha System:
Osseointegration

History:

Work first done by Swedish Professor Per Ingvar Branemark who was looking at regenerative qualities of bone.

He established no adverse surgical reaction to titanium.

He discovered that titanium, if left undisturbed in bone, forms an initial bond which strengthens over time.

Baha System:
Osseointegration

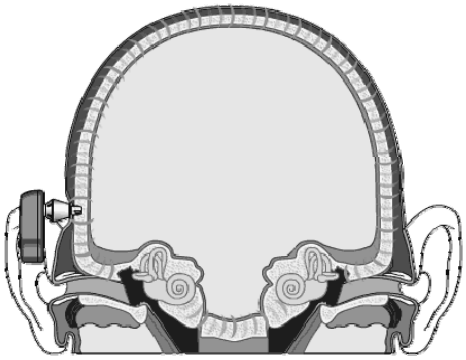
Process:

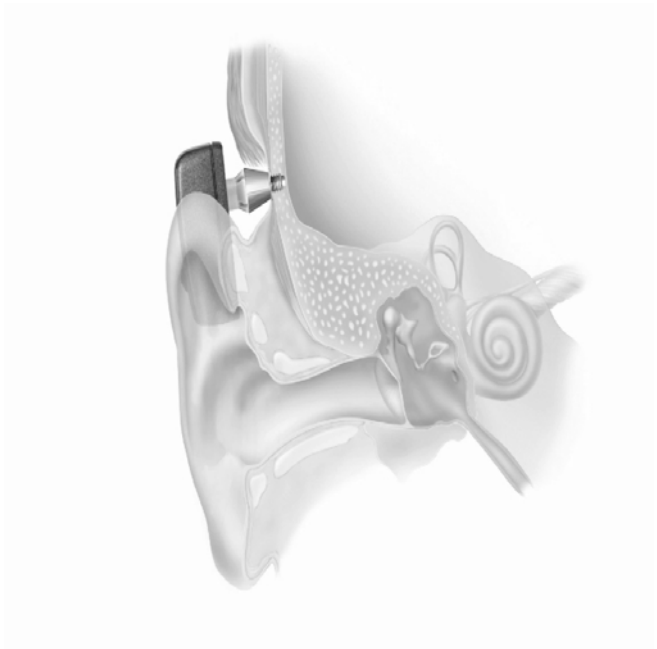
The process by which living bone tissue bonds with titanium

Makes direct bone conduction possible

The long-term predictability and success of both the Baha System and Vistafix™ rehabilitation is based on the fact that an active bond between tissue and implant is created without capsule formation. The implant is not only accepted, but incorporated into the bone.

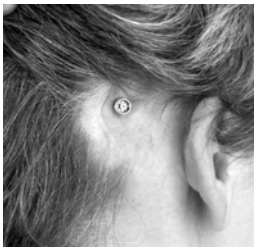
Baha System

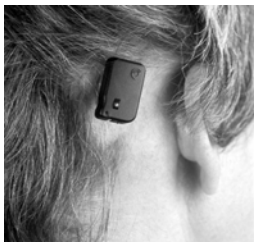




Baha System

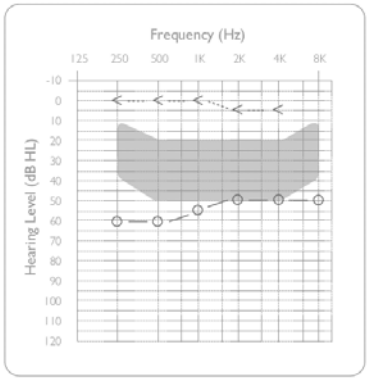
Implanted





Key Indications for Baha

1. **Conductive Loss**
2. **Mixed Loss**
3. **Single Sided Deafness (SSD)**



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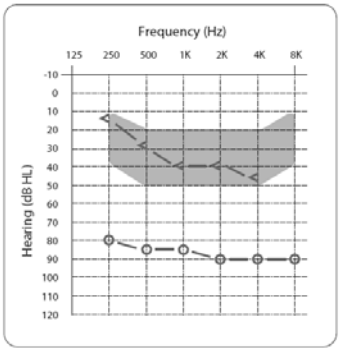

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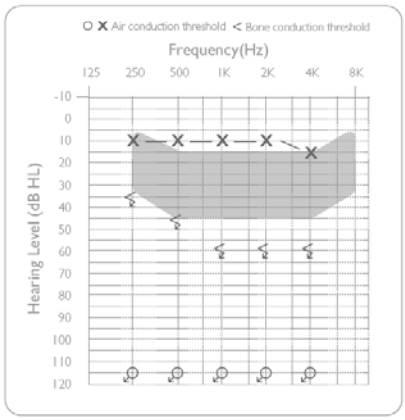


FDA Indication Criteria

≥ 5 years of age for surgical intervention

≤ 45 dB HL BC PTA

For bilateral fitting - Symmetric bone conduction thresholds are defined as less than 10 dB difference on average (0.5, 1, 2, and 3kHz) or less than 15 dB at individual frequencies.

European Implantation Criteria

Skull thickness of 2.5 mm

No age restrictions but this usually happens around 2.5 years of age

The Waiting Game…

5 years of age for implantation in the U.S.

Baha can still be utilized until the child is old enough for FDA approval

Baha Softband

Baha Divino

Digital technology

Two internal microphones
- Omni directional microphone
- Directional microphone

Two trim pots
- Adjustable tone control and automatic gain control output

Colors: black, blonde, silver/grey and brown

Divino

Divino Stickers

Divino

Divino Program Selection

Program 1 - Omni Directional Microphone
Program 2 – Directional Microphone

Sound processor picks up sound regardless of the direction the sound comes from.

Program is suitable for quiet or less noisy surroundings.

Note: program 1 is on when switch is turned up.

Intenso

Mixed Hearing Loss (added gain)

Digital Signal Processing

Active Feedback Cancellation

3 Listening Programs (omni, noise reduction, and DAI)

2 Trim pots for fine tuning – Gain and Low Cut

Colors: black, blonde, silver/grey, brown

Baha Pre-Implant Considerations

Lifestyle
- Swimming?
- Contact Sports?

Cleanliness
- Can the patient or caregiver clean the abutment daily?

Appearance of the device
- It is not fully implanted!

Other concerns
- Frequent hat wearer?
- Scar tissue?
- Hair growth?

Baha Trials

Can demo the processor in office

Let the patient try the Baha in office using a
- metal test band
- Cordelle or Intenso work best

Functional gain in the booth for CHL and MHL

Baha Follow-Up

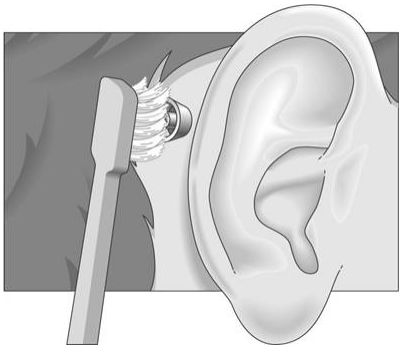
Has a volume control much like a hearing aid

Uses a hearing aid battery
- What is the correct battery size:
  - #13 (Divino)
  - #675 (Intenso)

What is the expected battery life:
- 200 hours or two weeks for Compact or Divino
- 150 hours or 1.5 weeks for Intenso

Baha Follow-Up

Abutment site must be cleaned daily
- Important to avoid infection and should be performed at least 3 ties a week with a soft cleaning brush provided by the manufacturer

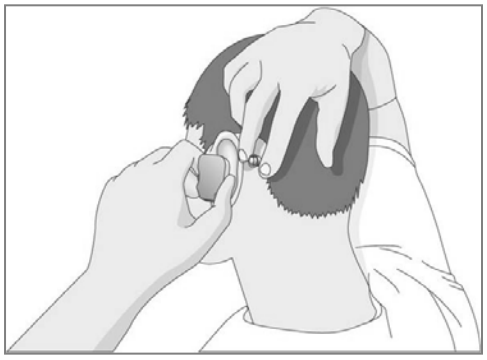


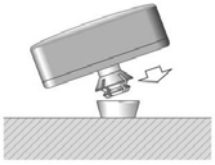
Baha Follow-up

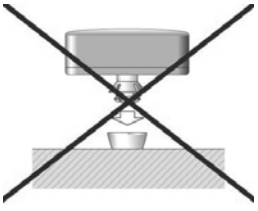
Care must be taken with snapping the sound processor on and off the abutment.

Snap trainers are available for practice.

Push hair from area and carefully push the device into the abutment at a slight, inward angle.



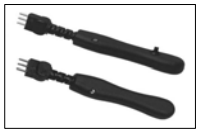




Baha Accessories

Phonak Microlink

Telecoil Units



Audio Adapter

Baha Case Studies

christensenlv@archildrens.org

Family Support

FRIDAY
Breakout Session #1: 10:45 – 11:45 AM
Trillium Ballroom III, Conference Center

MINDING THE GAP

David Brown
Education Specialist
California Deaf-Blind Services
&
Gail Deuce
Teacher of the Deaf
SENSE, United Kingdom

Presenter Information: *David Brown* began his career in the United Kingdom but now provides educational services in California. He has written and spoken widely on CHARGE syndrome and deaf-blindness.. He is an extraordinarily perceptive observer of individuals with deaf-blindness. His descriptions of the challenges faced by specific individuals with multiple anomalies and the effects on individual behavior have provided a model for the study of CHARGE syndrome and have shaped the body of knowledge that has been amassed over the past two decades. ADDRESS: 885 Corbett Avenue, San Francisco CA 94131; TEL: 415-405 7559; EMAIL: davidb@sfsu.edu

Gail Deuce works in the UK and is a qualified teacher of the deaf and has a M.Ed. in Multi-sensory Impairment. She has over twenty years experience in the field of special education, working initially in schools for children with severe learning difficulties and then a school for the deaf before moving into peripatetic work focusing on learners who are deafblind. Gail has worked for the ILEA, Surrey, Bedfordshire and Essex local authorities before joining Sense in December 2001. Gail has a particular interest in CHARGE and is on the committee for the CHARGE Family Support Group. She has recently embarked on a PhD, undertaking research considering the challenges to learning for children with CHARGE Syndrome.

Presentation Abstract: This session will be based on interviews undertaken by David and Gail with parents of young people/ adults to consider: (1)Whether or not parents have talked to their child about their having CHARGE or about them having certain CHARGE anomalies; (2) If they did this what made them decide to do it and when did they do it? If not, why not?; (3) If they did this, how was it done and were any other persons involved (e.g. a professional, another family member, another person with CHARGE); (4) What worked and what didn't work in this process?; (5) Has it been an ongoing process and what determined the timing and nature of the steps in the process?; (6) How has this issue been addressed to meet the communicative and cognitive needs of the individual?

No handout is included in the Conference Program for this presentation.

Information for New Families

FRIDAY
Breakout Session #1: 10:45-11:45 AM
Trillium Ballroom IV, Conference Center

CHARGE 101:
CHARGE SYNDROME:
DIAGNOSIS AND FEATURES

Meg Hefner, M.S.

Genetic Counselor
Clinical Associate Professor of Pediatrics
Saint Louis University, School of Medicine

Presenter Biography: *Meg Hefner* has been a genetic counselor for nearly 30 years, more than 25 of those working on CHARGE syndrome. She wrote the original Booklet for Families with Sandra Davenport and Jim Thelin and co-wrote and co-edited the Management Manual for Parents. Meg was a founding member of the CHARGE Syndrome Foundation Board of Directors and coordinated the first several International conferences. She continues as a Special Advisor to the Board, Chair of the Research Committee and presenter at conferences. She was a guest editor of the American Journal of Medical Genetics special issue on CHARGE syndrome. She has been an invited speaker on CHARGE syndrome to professional and lay audiences around the world. In addition to her work with CHARGE syndrome, she is a prenatal genetic counselor.

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Presentation Abstract: A brief history of CHARGE: where did the name come from? How often does it happen? How often does it happen again? Diagnosis: What else can look like CHARGE? What about the DNA test? What are the features? Which ones are helpful in diagnosis and which ones are most important in management? What are the consequences of the features of CHARGE?

CHARGE Syndrome diagnosis and features

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Where does the name come from?

**1981 Pagon, et al.

- C = coloboma of the eye
- H = heart anomalies
- A = atresia of the choanae
- R = retardation of growth or development
- G = genitourinary anomalies
- E = ear anomalies and/or deafness

****No longer used for diagnosis**

Epidemiology of CHARGE Syndrome

- 1 in 8,500-10,000 births
- Most often a new dominant mutation
  - Advanced paternal age
  - Increased but low recurrence risk
  - Major gene identified 2004 (CHD7)
- Mild end of spectrum is unknown
  - Parents or other family members with "suspicious" findings

Recurrence

- Empiric risk of 1% for parents with one child with CHARGE

- 50% for children of individuals with CHARGE

1998/2003 CHARGE Diagnostic Criteria Major and Minor Features

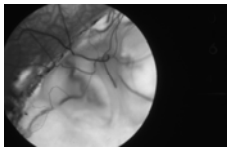
Definite CHARGE Syndrome:

- 3 or 4 Major Features OR
- 2 Major Features + 3 Minor Features OR
- Positive gene test (CHD7)

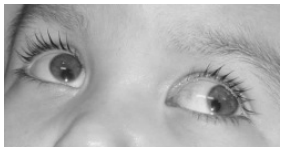
CHARGE Syndrome Major Features (the 4 C's)

- **Coloboma of the globe**
- Cranial nerve anomalies
- Choanal atresia/stenosis
- Characteristic CHARGE ears

Coloboma – cleft of the eye



Retinal Coloboma
Increased risk of retinal detachment with retinal coloboma

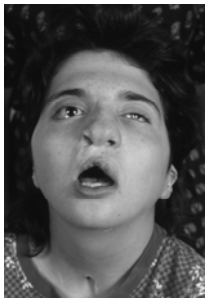


Iris Coloboma

Retinal coloboma causes upper visual field defects

Macular coloboma – central vision loss

- The child will look <u>above</u> the point of interest and not appear to be making eye contact



CHARGE Syndrome Major Features (4 C's)

- Coloboma of the globe
- **Cranial nerve anomalies**
- Choanal atresia/stenosis
- Characteristic CHARGE ears

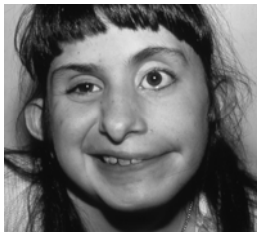
Cranial nerves 9 & 10: Swallowing problems

- Many require G-tube feeding
- May last for years
- Some adults still avoid certain textures

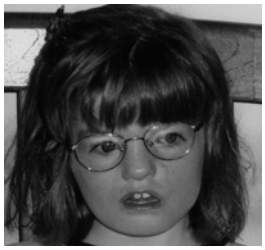
Swallowing complication: Aspiration

- Most common reason for hospitalization in first 2 yr. of life
- GE reflux is common
  - Surgery (Nissen) may help
- Gastrostomy may be needed to reduce aspiration

Cranial Nerve # 7
Facial palsy



Unilateral – lopsided face



Bilateral – no facial expression

Cranial Nerve #1
Sense of smell

Decreased or absent sense of smell

– Difficult to evaluate, but EXTREMELY common (>90%)
– Changes "taste"
– Implications for feeding
– Social implications
  - smelly feet
  - body odor
  - passing gas

CHARGE Syndrome
Major Features (4 C's)

- Coloboma of the globe
- Cranial nerve anomalies
- **Choanal atresia/stenosis**
- Characteristic CHARGE ears

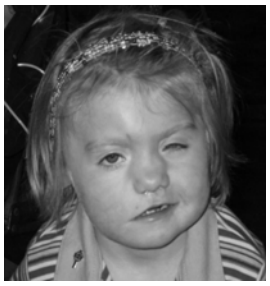
Choanal atresia/stenosis

- Two sides or one side
- Bony or membranous
- Blockage (atresia) or narrowing (stenosis)
  (One-sided stenosis may be difficult to diagnose)
- Often requires several surgeries to remain open

Baby with stents

Choanal stenosis

- Narrowing of nasal passages
- Goopy nose all the time
- Lots of ear infections, fluctuating hearing



CHARGE Syndrome
Major Features (4 C's)

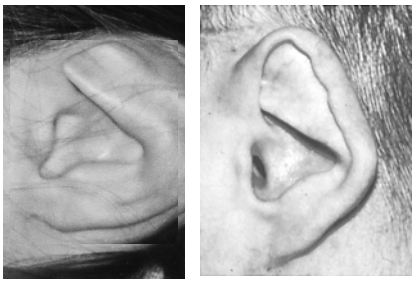
- Coloboma of the globe
- Cranial nerve anomalies
- Choanal atresia/stenosis
- **Characteristic CHARGE ears**

Characteristic CHARGE Ear: OUTER EAR

- Asymmetry between the two ears
- Floppy (deficient cartilage)
- Small/absent earlobe
- Triangular concha
- Clipped off helix (outer fold)

Characteristic CHARGE Ear

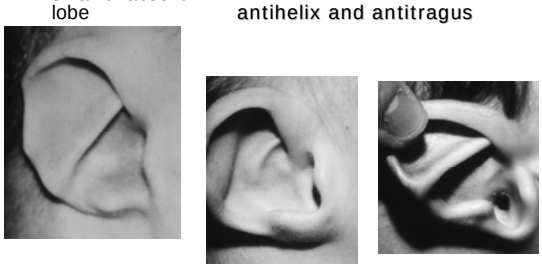
- Short, wide, triangular concha, absent lobe



Characteristic CHARGE Ear

Snipped off helix with small or absent lobe

Discontinuity between the antihelix and antitragus

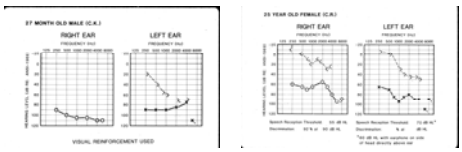


Characteristic CHARGE Ear: MIDDLE AND INNER EAR

- Middle ear bones – ossicular malformations (stapes, incus)
  - Conductive hearing loss
- Inner ear
  - Mondini defect: 1-1/2 turns to the cochlea
  - Small or absent semicircular canals
    - Balance problems

Hearing in CHARGE

- Mixed hearing loss
- Fluctuating with infections
- May respond to cochlear implant
- May be progressive



Inner Ear Balance Problems

- Small or absent semicircular canals
- Characteristic gross motor development
  - Stay close to the ground
  - Creep or combat crawl
  - 5-point crawl
  - "Bottom shuffle"
- Age of walking
  - Walk at 24-32 mo. if no other problems
  - Walk at 3-4 yr. if hypotonia and visual impairment

CHARGE Syndrome Minor Diagnostic Features

- Some are very common to CHARGE but difficult to evaluate consistently
- Some are very common to CHARGE but also very common in other syndromes
- Some are less common in CHARGE

CHARGE Syndrome Minor Features Unique to CHARGE

- Upper body (?truncal) hypotonia
- Characteristic CHARGE face
- Hockey stick palmar crease

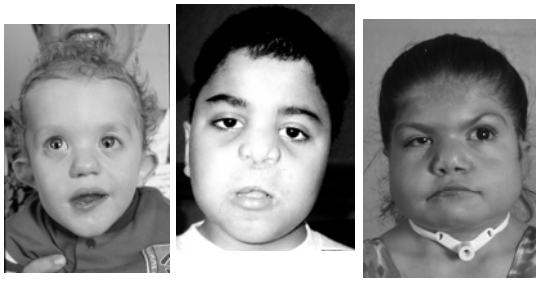
Truncal hypotonia

- Slumping posture
- Crawl on back
- Bottom shuffle
- Delayed walking
- Complicated by poor balance

Characteristic CHARGE Face

- Square face
- Broad forehead
- Round eyes
- Nose
  - Prominent bridge
  - Square root
  - Prominent columella
- Small chin, gets bigger with age

Characteristic CHARGE Face



To see more faces: "About CHARGE" link at www.chargesyndrome.org

CHARGE: 7 to 19 years

Minor feature: Hockey-stick palmar crease



CHARGE Syndrome
Minor Feature: Heart defects

- Tetralogy of Fallot
- Aortic arch anomalies
- Complex heart defects
- Can be any heart defect

- Same spectrum of heart defects as in VCFS (22q deletion syndrome)

Heart surgery at four years old

CHARGE Syndrome
Other Minor Feature: Clefts

- Cleft lip
- Cleft lip and palate
- Cleft palate
- Submucous cleft palate

Repaired cleft lip

CHARGE syndrome minor feature: Esophagus/Trachea

- Esophageal atresia (EA)
  - esophagus not connected to stomach
- Tracheo-esophageal fistula (TEF)
  - connection between trachea and esophagus
- H-shaped TEF
  - can be hard to diagnose, but important
- Trachoemalacia
  - weak, collapsing trachea

CHARGE syndrome minor feature: Renal (kidney) anomalies

- Hydronephrosis
- Reflux
- Horseshoe kidney
- Small or absent kidney

- 40% have renal anomalies

CHARGE Syndrome
Minor Feature: short stature

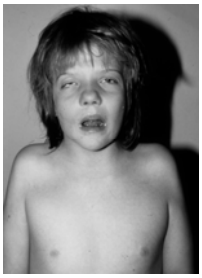
Due to:

- Medical problems
  - Heart
  - Feeding
- Growth hormone deficiency
- Short stature with no known cause

Treated with growth hormone

CHARGE Syndrome
Minor Feature: genital hypoplasia

- Males
  - Micropenis
  - Cryptorchidism (undescended testes)
  - Lack of normal puberty
- Females
  - Small labia
  - Absent uterus
  - Lack of normal puberty



17 year old with no hormone treatment (and facial palsy)

Variability

- EVERY feature can vary from absent to severe in different children
- NO ONE FEATURE is seen in every person with CHARGE
- Definite CHARGE is 3 or 4 Majors or 2 Majors and 3 Minors
- Probable/Possible CHARGE is more difficult
- The new gene test helps, but $$

Beyond diagnosis: Other features (any organ system!)

- Brain anomalies seen on MRI or CT
- Seizures
- Apnea
- Laryngomalacia
- Floppy cartilage
- Nipple anomalies
- Thymic or parathyroid abnormalities

More Other features

- Webbed neck
  - Sloping shoulders
  - Absent trapezius muscle
- Abdominal wall defects
  - Umbilical hernia
  - Omphalocele
- Limb/skeletal anomalies
  - Polydactyly common in Japan
  - Scoliosis

CHARGE *management* issues beyond Major and Minor features

- Constipation - Autonomic nervous system?
- Potty training – nerve abnormalities?
- Sleep disturbances
  - abnormal circadian cycle
- Cyclic vomiting/abdominal migraines
- Sensory integration
- Behavior

Behavior in CHARGE syndrome

- Autistic-like behaviors
  - May be deafblind behavior
  - May be autism
  - May be CHARGE
- Stubborn, perseverative
- OCD
  - A real feature of CHARGE

Intelligence in CHARGE

- Long-term prognosis may be excellent
- Most are “input impaired” due to combined vision & hearing loss
- A few are “output impaired” due to bilateral facial palsy
- Delayed motor milestones due to vestibular dysfunction, upper body hypotonia and impaired vision.

How well do they do?
The typical 2 year old with CHARGE:

- has spent 6 months in the hospital
- has had at least 6 surgeries
- is followed by 10 medical specialists
- is fed by G-tube
- is not walking or talking
- has some unusual behaviors

Looks pretty “retarded,” but probably isn’t

A major gene for CHARGE

Finding a change (mutation) in a single gene is like locating a single person from space

Major CHARGE gene: CHD7 on chromosome 8q12

- Very large gene
- Many, many different unique mutations have been identified (no hot spots)
  - Makes testing very expensive
- CHD7 is a regulatory gene (turns other genes on and off): Thus affects every organ system

Now what - How is this gene helpful?

- Confirm diagnosis in questionable cases
  - Help define the mild end of the spectrum
  - *Not finding a mutation does NOT rule out CHARGE*
- Test other people in the family
  - Can “normal” people be carriers?
- Prenatal diagnosis
  - First must identify the mutation in the family
  - Can identify the gene, not the severity of the features

Information for New Families

FRIDAY
Breakout Session #1: 10:45-11:45 AM
Trillium Ballroom IV, Conference Center

CHARGE 101:
ANESTHESIA COMPLICATIONS IN CHARGE SYNDROME

Kim Blake, M.D.

General Pediatrician
Dalhousie University
Halifax, Nova Scotia

Presenter Biography: *Dr. Kim Blake* began her involvement with CHARGE more than 25 years ago at Great Ormand Street Hospital in the UK, where she lectured on CHARGE and published several of the earliest papers describing the syndrome. She was instrumental in organizing the UK family support group. Since moving to Canada, she has continued to be involved with the CHARGE Syndrome Foundation. She has been an invited speaker at every conference and received funding from the Foundation for several of her research projects. Kim's research career continues to focus on CHARGE, particularly the issues of the adolescent and adult population. She routinely involves students in her research, both educational and clinical. Most of her students have had abstracts and/or papers published, some even with first authorship. Kim has recruited many local medical faculty members in her research and developed a center of excellence for research and knowledge in CHARGE syndrome. She is regularly asked to present on CHARGE syndrome, both nationally and internationally. Her most recent research projects are on the effects of anesthesia in CHARGE syndrome and the feeding difficulties in children with CHARGE syndrome.

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Presentation Abstract: Why is anesthesia important? How many surgeries is my child likely to have? Why is it important to combine procedures with one anesthesia?

ANESTHESIA COMPLICATIONS IN CHARGE SYNDROME

Dr. Kim Blake, MB, MRCP, FRCPC

9TH International CHARGE Syndrome Conference
July 24-26, 2009
Chicago, Illinois

Objectives

- By using case reports, you will understand the risks of anesthesia in CHARGE syndrome.
- You will recognize that Botox injections into the salivary glands can be used to decrease oral secretions.

There are Always Risks of Complications with Anaesthesia

- "…you sign a consent"
- Are you informed?

- Are Individuals with CHARGE Syndrome More at Risk?

If yes, what are the risks and who should know?

APSEA 2006

Growing up With CHARGE Syndrome

Kennedy

Age 0-2 years: 7 surgeries

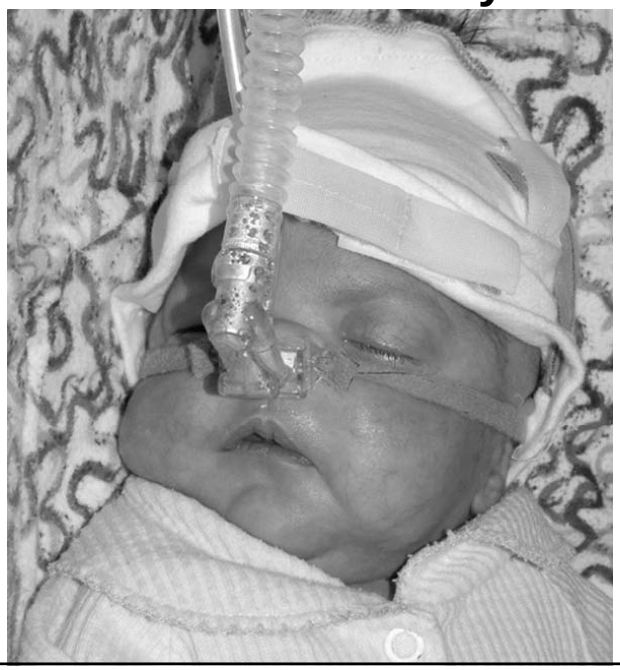
Age 2-4 years: 3 surgeries

Age 4-6 years: 6 surgeries

Kennedy's ICU Admissions
Four in Total

- 5-19 days – open heart surgery and supraventricular tachycardia.
- 6 months – G-tube/fundoplication
- extubation attempted (x 3)
- 18 months – aspiration pneumonia
- 6 yrs – post op heart surgery – 1 day ? pneumonia

Frederick's Story

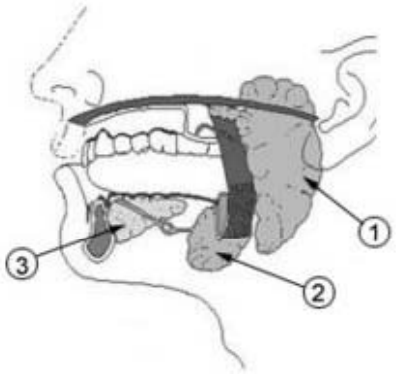


Freddy at 2 Months

- Difficulty with intubation
- ToF repair, vascular ring repair, PDA ligation
- Increased oral secretions
- Multiple attempts at extubation

Site of Botox Injections

1. Parotid glands
2. Submandibular glands
3. Sublingual glands



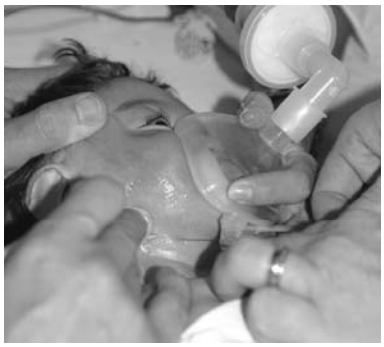
Botox 7.5 units was injected into salivary glands 1 and 2 on each side

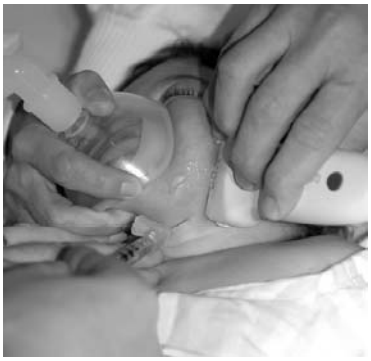
Freddy at 7 Months

- Aspiration pneumonia from oral secretions
- Gastroesophageal reflux
- Required ventilation

Botox Injection

Submandibular Gland Via Ultrasound and Parotid Gland by Palpation





10 Units/gland

Botox Injection

Prophylactic Use to Prevent Increase in Oral Secretions and Aspirations (4-5 monthly)

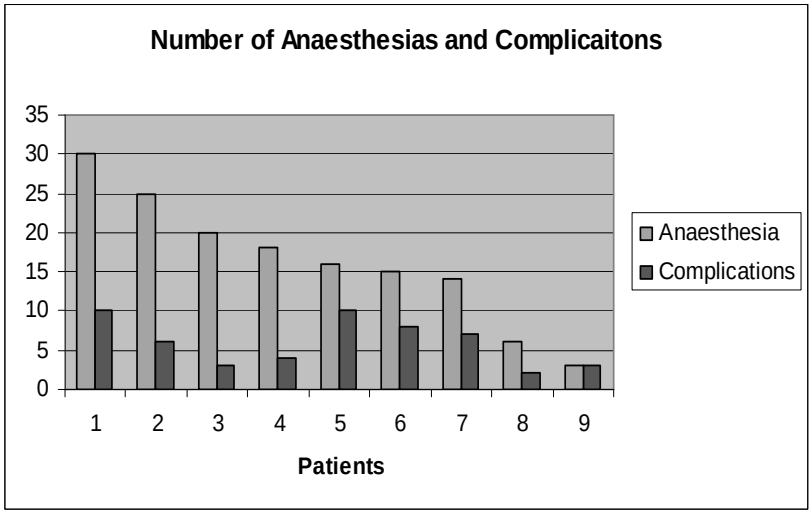
Waiting for picture

Postoperative Airway Events of Individuals with CHARGE Syndrome

- Detailed chart reviews on nine patients
  - Mean age 11.8 years (± 8.0)
  - 215 surgeries (average 22 per child)
  - 147 anesthesias (average 16 per child)
- Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Blake K, MacCuspie J, Hartshorne TS, Roy M, Davenport SLH, Corsten G.*International Journal of Pediatric Otorhinolaryngology*, *Vo. 73*, *February 2009*

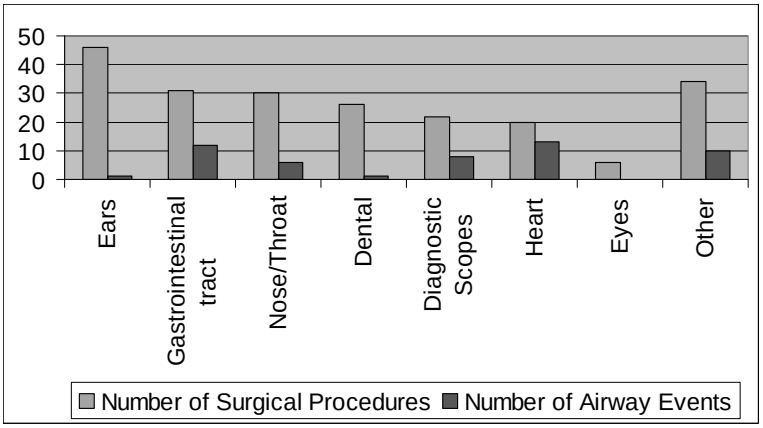
Results



35% (51/147) of anesthesias resulted in complications (>60% were major)

Results

Anesthesia related complications occurred most often with heart, diagnostic scopes (L/B/E) gastrointestinal tract procedures



Results

Number of surgical procedures per anesthetics with resulting postoperative airway events.

Number of surgical procedures	Number	Post-operative Events	Percent resulting in airway events
1	94	37	39% (n= 37/94)
2	36	8	22% (n= 8/36)
3+	15	5	33% (n= 5/15)

P=0.1 Combining multiple procedures under one anesthesia does not lead to an increase in post-operative events.

Results

Feeding procedures and rates of postoperative airway events.

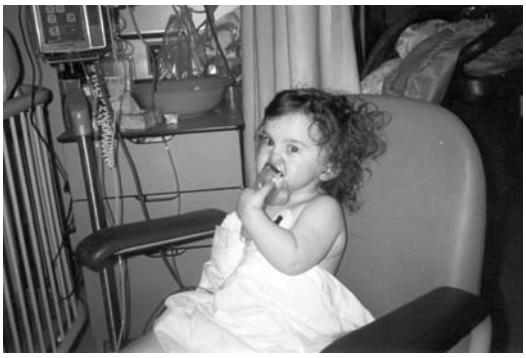
	Number of Anesthesias	Airway Event	No Airway Event	Significance
G/J tube	82	36	46	Yes p=0.0092
No G/J tube	63	15	48	
Nissens fundoplication	79	33	46	Yes p=0.049
No Nissens fundoplication	66	18	48	

Having a G/J tube or Nissens fundoplication increases your child's risk of post-operative airway events

Tonsils and Adenoids

- How many of your children have had a tonsillectomy and/or adenoidectomy?
- How many of your children had anesthetics after that?
- How many of your children had improved recovery after subsequent surgeries?
- 2/3 of our population had improvement after T/A.

MacKenzie's Story



- 27 surgical procedures
- 18 anesthesias
- 4 complications
- Multiple ICU admissions

Discussion

- 35% of anesthesias resulted in post-operative complications
- Heart, diagnostic, and gastrointestinal tract procedures result in the most complications
- At least one complication occurred with every type of surgery except for eyes

Discussion

- High risk of complications in individuals with Nissen fundoplication or gastrotomy/jejunostomy tube
- Low risk with cleft palate
- What about individuals with CHD7 mutations who have mild clinical criteria?
  - Will they be at risk in the future?
  - Have they actually been challenged with surgeries?

Take Home Messages

Your children are at high risk of post-operative anesthesia complications. Combining procedures during one anesthesia does not increase the risk of post-operative airway events.

The anesthesiologist needs to be aware that, even with simple procedures, the individual with CHARGE syndrome is at high risk of complications.


Thank
You!

FAMILY SUPPORT

FRIDAY
Breakout Session #1: 10:45 – 11:45 am
Byrne Room in the Hotel, Atrium Level

GRANDPARENT MEETING

Moderator:
Leonard Stanger

Moderator Information: *Mr.* Stanger is the grandfather of a teenage young adult who has CHARGE syndrome (Megan) and the father of the CHARGE Syndrome Foundation president (Neal Stanger).

EMAIL: Marlboro8@aol.com

Purpose of the Grandparent Meeting: CHARGE syndrome affects every member of the family. This meeting is only for grandparents. It is an opportunity for those who have a grandchild with CHARGE to meet and discuss whatever is of importance to those in attendance. By request of grandparents at earlier CHARGE Conferences, the Grandparent Meeting is held at the beginning of the Conference so that the grandparents can meet each other early and socialize and share information for the next two-and-half days. This informal discussion session will be led by Leonard Stanger who is very familiar with the issues of importance to those who have a grandchild with CHARGE.

FAMILY SUPPORT

FRIDAY

Breakout Session #1: 10:45 – 11:45 am

Jensen Room in the Hotel, Atrium Level

FATHERS' FORUM

Moderators:

John Reiman & Timothy Hartshorne

Moderator Information: *Dr. Reiman* has been in the special education field for 30 years. He is the Associate Director of the OSEP-funded National Consortium on Deaf-Blindness. He also is a Dispute Resolution Specialist with CADRE (The National Center on Dispute Resolution in Special Education) and has mediated hundreds of conflicts addressing family, special education (including IEP's), workplace, social service, and community matters over a 20 year period. He is an Advanced Practitioner member of the Association for Conflict Resolution. John is a trained Crisis/Trauma Intervention team member (with law enforcement) and is a practicing trauma psychotherapist (licensed in Oregon). He is the father of a 21 year old son with learning disabilities and runs groups for fathers.

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Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. His particular interest is in understanding the challenging behavior exhibited by many individuals with CHARGE. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project. His current project is editing a book, along with Sandy Davenport, Meg Hefner, and Jim Thelin, on CHARGE which should be published in 2010.

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TEL: (989)774-6479 office; FAX: (989)774-2553; EMAIL: tim.hartshorne@cmich.edu ;
WEB: www.chsbs.cmich.edu/timothy_hartshorne

Purpose of the Fathers' Forum: CHARGE syndrome affects every member of the family. This meeting is only for fathers who have a child with CHARGE. It is their opportunity to have an open discussion and share experiences with other fathers who have had to deal with the issues associated with CHARGE syndrome. The moderators of the Fathers' Forum are two fathers with extensive personal and professional experience with special needs children and adults.

Young Adults & Adults with CHARGE Syndrome ONLY

FRIDAY
Breakout Session #1: 10:45 – 11:45 am
Cyperus IV, Lower Level in the Conference Center

WELCOME TO FUN FEUD

Wendy M. Bridgeo, M.Ed.
&
Christa Gicklhorn, M.Ed.

Deafblind Program
Perkins School for the Blind
Watertown, MA

Presenter Information: *Wendy Bridgeo* is a Vocational Teacher in the Deafblind Program at Perkins School for the Blind. She has been teaching career education and developing community-based vocational placements for students in the Deafblind Program for over 25 years. Wendy has extensive experience presenting to both national and international audiences on topics related to vocational training and portfolio development for adolescent students with significant disabilities. EMAIL: Wendy.Bridgeo@perkins.org
Christa Gicklhorn holds the position of Vocational Coordinating Teacher in the Deafblind Program at Perkins School for the Blind. As the Vocational Coordinator in the Deafblind Program, Christa has more than 23 years experience developing career education curriculum and community-based vocational training placements for students in the Deafblind and Outreach Summer Programs. Christa has extensive experience presenting to national audiences on topics related to vocational training and portfolio development for adolescent students with significant disabilities. EMAIL: Christa.Gicklhorn@perkins.org

ADDRESS: Perkins School for the Blind, 175 North Beacon Street Watertown, MA 02472; TEL: 1-617-972-7304

Presentation Abstract: This presentation will highlight the importance of providing students with significant disabilities with a preparatory vocational curriculum that starts with the end in mind. Functional and creative vocational education and training enhances vocational exploration, skill development and assessment. Seasoned vocational educators from the Deafblind Program at Perkins School for the Blind will share ideas, activities and strategies that have been implemented and refined in school and community-based vocational settings.

Welcome to Fun Feud

Your Hosts:

Wendy Bridgeo

Christa Gicklhorn

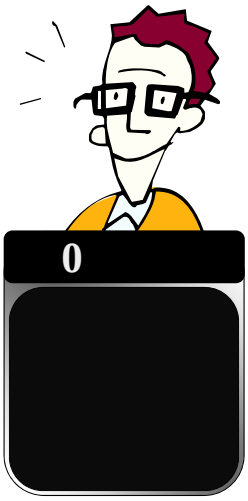
Start

To Reset your game

This game sample is provided compliments of PTT, Inc.

Reset

Family Feud Score Slide



Question 1
Question 2
Question 3
Question 4
Question 5
Question 6
Question 7
Question 8
Question 9
Question 10
Question 11
Question 12
Question 13
Question 14
Question 15

Close

Q1

Name the top 5 answers to this:

Why do people work?

		Points
1	Money (housing, food, clothing, entertainment)	50
2	Pride and satisfaction	20
3	Way to get connected in [illegible] community	20
4	Meet new people and build friendships	10
5	Helps structure your day	10

Wrong

X X X

Team 1

Team 2

Q2

Name the top 5 answers to this:

What are 5 ways to explore your vocational interests?

		Points
1	Summer Jobs	30
2	Community Service	25
3	Network	20
4	Job Shadowing	15
5	Career Fairs	10

Wrong

X X X

Team 1

Team 2

Q3

Name the top 5 answers to this:

What are 5 ways to find a job?

		Points
1	Family Member	30
2	Job Applicatio	20
3	Internet Search	20
4	Guidance Counselor / Teacher	20
5	Newspaper	10

Wrong

X X X

Team 1

Professional Training Technologies, Inc. - www.pttinc.com - info@pttinc.com

Team 2

Q4

Name the top 5 answers to this:

What personal information is asked for on a job application?

		Points
1	Name / Address	25
2	Work Experie e	25
3	Education	20
4	References	20
5	Interests and Special Skills	10

Wrong

X X X

Team 1

Professional Training Technologies, Inc. - www.pttinc.com - info@pttinc.com

Team 2

Q5

Name the top 5 answers to this:

Who should fill out a job application for you?

		Points
1	You	30
2	Family Member	20
3	Job Coach / Job Developer	10
4	Teacher	10
5	Transition Counselor	10

Wrong

X X X

Team 1

Team 2

Q6

Name the top 5 answers to this:

What do you do to prepare for a job interview?

		Points
1	Dress for success / clean neat clothes	30
2	Organize your "personal information" (Résumé, references)	20
3	Know your strengths and weaknesses	20
4	Plan your rides to / from the interview	15
5	Communication Supports (tech, notes)	15

Wrong

X X X

Team 1

Team 2

Q7

Name the top 5 answers to this:

What are some job interview questions?

		Points
1	What is your name?	40
2	Where did you go to school when did you graduate?	20
3	Have you had any work experience?	15
4	What are your strengths and weaknesses?	15
5	Would you prefer to work full or part thime?	10

Wrong

X X X

Team 1

Team 2

Q8

Name the top 5 answers to this:

What are things you should do on a job interview?

		Points
1	Be on time	40
2	Introduce yourself & shake hands	20
3	Bring resume	15
4	Make eye contact	15
5	Ask questions	10

Wrong

X X X

Team 1

Team 2

Q9

Name the top 5 answers to this:

What are things you don't do at an interview?

		Points
1	Arrive late	40
2	Don't be impolite / rude	20
3	Don't ask about pay	20
4	Don't answer your cell / text	10
5	Don't be silly or impatient	10

Wrong

X X X

Team 1

Team 2

Q10

Name the top 5 answers to this:

What work conditions should you consider when looking for a job?

		Points
1	Work hours	30
2	Standing / sitting / moving	25
3	Alone or with others	25
4	Uniform or dressy/casual clothes	10
5	Busy or quiet place	10

Wrong

X X X

Team 1

Team 2

Q11

Name the top 5 answers to this:

What makes a good worker?

		Points
1	Good attendance / on time	40
2	Honest / hard working/ reliable	20
3	Accepts supervision	20
4	Follows workplace rules	15
5	Follows break / lunch schedules	5

Wrong

X X X

Team 1

Team 2

Q12

Name the top 5 answers to this:

What are rules you would find in a workplace?

		Points
1	Come to work on time	25
2	Follow the dress code	25
3	Listen to your supervisor	25
4	Follow safety / sanitary rules	15
5	Return from lunch / breaks on time	10

Wrong

X X X

Team 1

Team 2

Q13

Name the top 5 answers to this:

What do you do at break and lunch time?

		Points
1	Use your cell phone to make calls/text	50
2	Eat	20
3	Socialize	15
4	Use bathroom / wash hands / tidy up	10
5	Leisure activities, books, magazine, ipod	5

Wrong

X X X

Team 1

Team 2

Q14

Name the top 5 answers to this:

What are some things you don't do during work time?

		Points
1	Don't' use your cell phone to call/text	40
2	Don't disrespect others sound, don't make unnecessary noise (whi , bang, tap, sing)	25
3	Don't over share: don't tell ple everything about your personal life	20
4	Don't eat or drink at your work station.	10
5	Don't invite friends to work	5

Wrong

X X X

Team 1

Team 2

Q15

Name the top 5 answers to this:

What are some personal accommodations?

		Points
1	Hearing aids / glasses	35
2	Mobility aids / transportation	30
3	Communication devices	20
4	Technology	10
5	Job coach support / rehab counselor	5

Wrong

X X X

Team 1

Team 2

Sensory Information

FRIDAY
Breakout Session #2: 1:00-2:00 PM
Trillium Ballroom II, Conference Center

COCHLEAR IMPLANTS

Beth Tournis, Au.D.
Children's Memorial Hospital
Chicago, IL

Presenter Information: *Dr. Tournis* is an audiologist at Children's Memorial Hospital in Chicago , IL. She is the coordinator of the hospital's Cochlear Implant Program.

EMAIL: BTournis@childrensmemorial.org

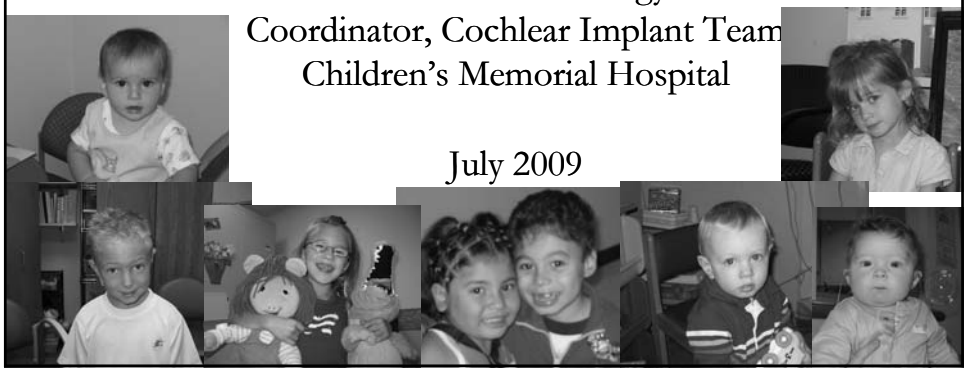
Presentation Abstract: This presentation will describe the procedures associated cochlear implantation: the criteria for candidacy for implantation, the audiologic information needed; expectations and success; and post-surgical services. Case studies will be presented.

Handouts of material not included in the Conference Program may be obtained from Dr. Tournis at BTournis@childrensmemorial.org

Cochlear Implants

Beth Tournis, Au.D.
Doctor of Audiology
Coordinator, Cochlear Implant Team
Children's Memorial Hospital

July 2009



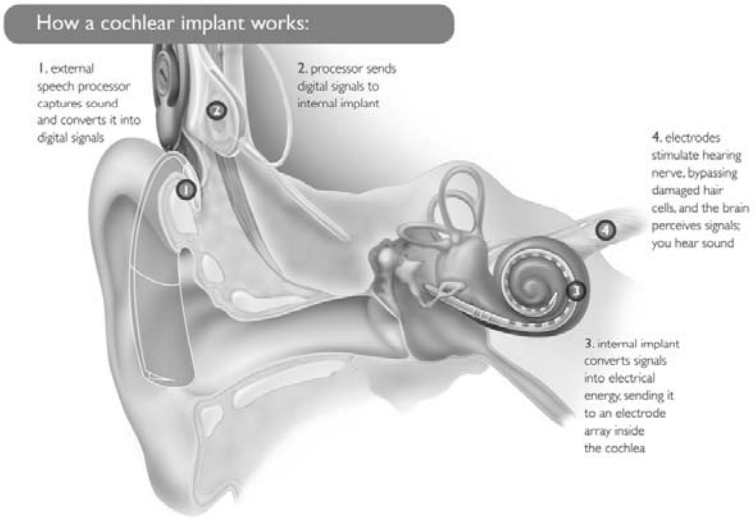
Cochlear Implant Team

- Otologist – Nancy Young, M.D.
- Pediatric Audiologists- Beth Tournis, Au.D., Denise Thomas, Au.D., Meghan Crimmins, Au.D., Stephanie Yaras, Au.D., Lisa Weber, M.S.
- Speech pathologists- Jennifer Still, SLP, Rachael Potter, SLP Megan Rascher, SLP, Avanti Lal, SLP
- Psycholol gist - Connie Weil, SLP
- Hart Family Educational Coordinator- Stacy Simek, M.Ed.
- Social worker - Anne Lyons, M.S.
- Music Therapist - Liz Espinoza, M.A.
- Otology nurse – Mary Ellen Brennan, R.N.
- Cochlear Implant Program Assistant – Krista Geier
- Insurance coordinator- Georgina Simpkin

Goal of a Pediatric Audiologist

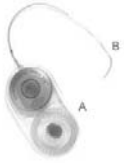
Goal of a Pediatric Audiologist is to deliver an effective signal to the ear that provides enough sound awareness and meaning to contribute positively to the child's language development

How Cochlear Implants Work



Cochlear Implant Devices







Candidacy Criteria

'Who qualifies for an implant?'

Assessing Candidacy

Our role as Audiologists is to determine if the patient would receive more **benefit** from a cochlear implant than they do with hearing aids.

So, to determine implant candidacy, we must first measure how much benefit **patient receives with appropriately fit hearing aids**

What is benefit**?**

- Detection of environmental sounds
- Discrimination of environmental and speech sounds
- Better speech reading skills
- Better comprehension of speech auditory only
- Better communication on the phone
- Hearing music

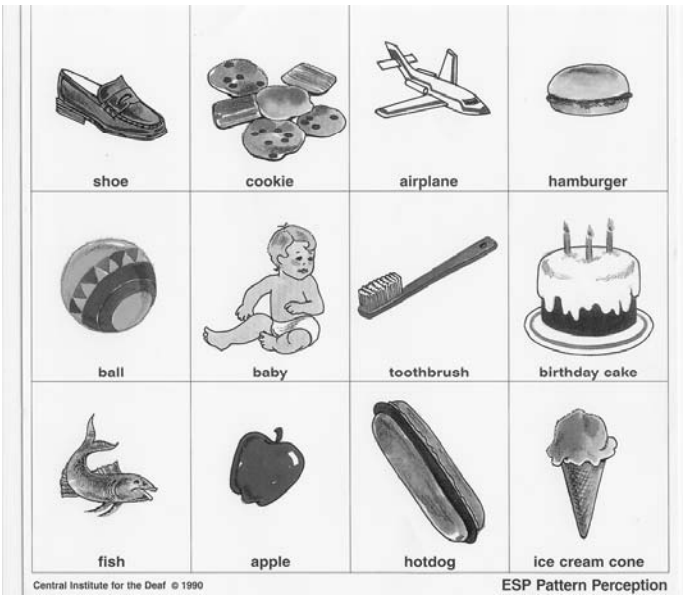
What tests do we use to assess CI candidacy?

- Unaided testing – CPA, VRA, ABR/ASSR
- Aided testing binaural (both aids)
- Aided testing monaural (each ear aided individually)
- Speech perception testing
- Parent/Therapist report

Speech Perception Testing

- Open set tests
- Closed set tests
- Parent surveys

Closed Set Test



IT-MAIS/MAIS

- Designed for parents of young children
- 10 questions in interview format
- each area scored based on frequency of occurrence
- total score=40
- example question: "Is the child's vocal behavior affected while wearing his/her hearing aid?"
  - 0=never, 1=rarely; 2=occasionally; 3=frequently; 4=always

So exactly who qualifies?

- No age limitations- routinely implanting children under 1 year of age
- Children with severe-to-profound SNHL who are not reaching their auditory potential with hearing aids alone
- Children with auditory neuropathy who are not detecting or who are not making appropriate speech/language gains

Minimum Expectations

- Full time use of appropriate amplification
- Enrollment is therapy
- Appropriate school placement, if applicable
- Regular attendance and compliance of necessary appointments
- Realistic Expectations

Additional Steps Include

- Aural hab/language eval
- Meeting with social work
- Therapy/school contact
- Psych eval if necessary

So exactly who qualifies?

- No age limitations- routinely implanting children under 1 year of age
- Children with severe-to-profound SNHL who are not reaching their auditory potential with hearing aids alone
- Children with auditory neuropathy who are not detecting or who are not making appropriate speech/language gains

Minimum Expectations

- Full time use of appropriate amplification
- Enrollment is therapy
- Appropriate school placement, if applicable
- Regular attendance and compliance of necessary appointments
- Realistic Expectations

So how do we predict outcome in general?

What is "Success"?

- Improvement in Spoken Language Skills
- Improvement in Expressive Language
- Improvement in Receptive Language
- Improved Awareness/Connection with Environment

"It is impossible to determine how well a child will perform with a cochlear implant. The duration of deafness, the age at implantation, the degree of auditory experience, training provided after implantation, and family support may all have an effect on a child's post-operative performance with a cochlear implant. In addition, personal attributes such as cognitive abilities, behavioral temperament, communication mode, and overall health and functioning may affect progress."

What about after the surgery?

Post Surgery Timeline

- 1 week after surgery – post-operative check
- 3-4 weeks after surgery-initial stimulation
- 3 weeks after initial stimulation- 3 week check
- 6 weeks after initial stimulation- 6 week check
- 12 weeks after initial stimulation- 3 month check
- 6 months after initial stimulation- 6 month check
- 9 months after initial stimulation- 9 month check
- 1 year after initial stimulation- 1 year check
- Every 6 months after that… interval checks

Initial Stimulation

- Device fitting or 'activation'
- Child is fitted with external equipment
- Perform subjective and/or objective testing to create very first MAP
- Every child reacts differently to sound through the implant
- Familiarize family with equipment

What happens at interval checks?

Interval checks

- History including wearing time and equipment needs
- Audiogram
- Speech perception testing
  - Ling Sound testing
  - Formal Speech Perception testing
- Reprogramming if necessary

Case Studies

Medical & Genetic Information

FRIDAY
Breakout Session #2: 1:00 – 2:00 PM
Trillium Ballroom III, Conference Center

GENETIC DIAGNOSIS OF CHARGE SYNDROME

Conny M. A. van Ravenswaaij-Arts, M.D., Ph.D.
Associate Professor in Clinical Genetics
and
Jorieke Bergman
Clinical Geneticist in Training and Ph.D. Student
Department of Genetics
University Medical Center Gronigen
The Netherlands

Primary Presenter Information: Conny van Ravenswaaij studied medicine at the University of Leiden, Netherlands. A PhD study was completed in 1993 at the Radboud University Nijmegen. In 2002 she was registered as a clinical geneticist. Her main interest has always been children with multiple congenital anomalies and chromosome disorders. Her group discovered the CHD7 gene as major cause of CHARGE syndrome in 2004. In 2006 she changed affiliation to the University Medical Centre Groningen, where she continued her two multi-disciplinary outpatient clinics (for rare chromosome disorders and for CHARGE syndrome) and her studies in these syndromes. Amongst many other activities, she initiated a European project on rare chromosome disorders (www.ECARUCA.net). Beside her work as a clinical geneticist she is involved in a number of parent support groups (CHARGE, Wolf-Hirschhorn syndrome, European 11q, Rare chromosome disorders).

For the studies in CHARGE syndrome she collaborates with many other researchers in Groningen as well as abroad. At this moment the studies focus on clinical variability and phenotype-genotype correlations, puberty development and smell, the role of CHD7 in heart development, and other aspects of CHARGE syndrome.

ADDRESS: University Medical Center Groningen, P.O. Box 30.001, 9700 RB Groningen, the Netherlands; TEL: 0031(0)503617229, FAX: 0031(0)503617231,EMAIL: c.m.a.van.ravenswaaij@medgen.umcg.nl

Presentation Abstract: The gene involved in CHARGE syndrome was identified in 2004. Mutations in this gene, the CHD7 gene, are found in the majority of CHARGE syndrome patients. Different techniques are used to analyze this large gene. Sequence analysis is the method of first choice and will identify mutations in the CHD7 gene. Two other techniques, MLPA and array CGH, can be used to find deletions or duplications of the CHD7 gene.

University Medical Center Groningen

Genetic Diagnosis of CHARGE syndrome

Conny van Ravenswaaij-Arts,
Jorieke Bergman, Lies Hoefsloot
Department of Genetics,
University Medical Center Groningen
The Netherlands

Outline of the presentation

- ✓ **The clinical diagnosis of CHARGE syndrome**
- ✓ **chromosomes and genes**
- ✓ **the *CHD7* gene**
- ✓ **DNA-diagnostics in CHARGE syndrome**
- ✓ **Should parents be tested?**

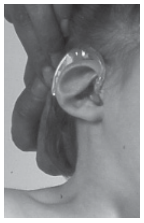
CHARGE Syndrome

Coloboma
Heart malformation
Atresia of choanae
Retardation (growth and development)
Genital hypoplasia
Ear abnormality including deafness

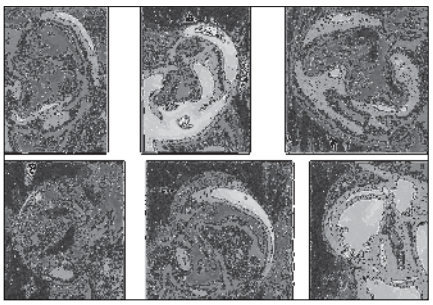
Estimated birth incidence 1:10,000

Associated features

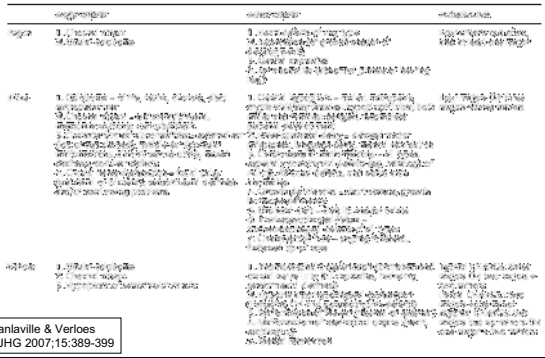
- Agenesis semicircular canals
- Cleft lip/palate
- Tracheo-oesophageal fistula
- Facial nerve palsy
- Renal anomalies
- Malformations of brain



Clinical variability in CHARGE syndrome



Clinical criteria



Sanlaville & Verloes
EJHG 2007;15:389-399

Clinical criteria

Blake:
(4 major or 3 major + 3 minor)

Major criteria

- Coloboma
- Choanal atresia
- Cranial nerve dysfunction
- Characteristic ear abnormalities

Verloes:
(3 major or 2 major + 2 minor)

Major criteria

- Coloboma
- Choanal atresia
- Hypoplastic semicircular canals

Blake *et al* Clin Pediatrics 1998

Verloes AJMG 2005

CHARGE syndrome ?

- Feeding problems / tracheomalacia
- Heart defect: vascular ring
- Mild MR
- Height P3
- Dysmorphic ears
- Bilateral severe hearing loss
- Aplasia of semicircular canals

- No coloboma
- No choanal atresia
- No cleft lip/palate

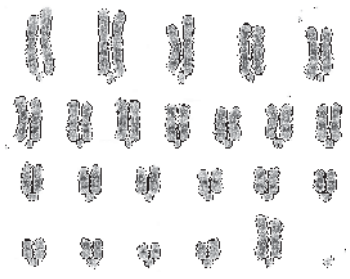
Diagnostic criteria

Diagnosis: Doctor or DNA???

Outline of the presentation

- ✓ **The clinical diagnosis of CHARGE syndrome**
- ✓ **chromosomes and genes**
- ✓ **the *CHD7* gene**
- ✓ **DNA-diagnostics in CHARGE syndrome**
- ✓ **Should parents be tested?**

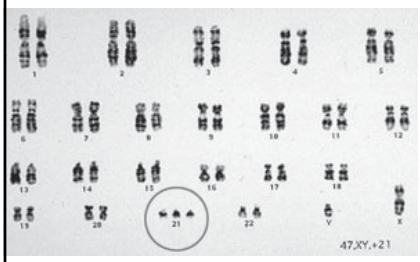
Chromosomes



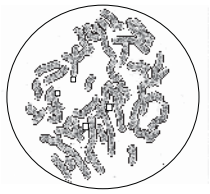
46 chromosomes per cell

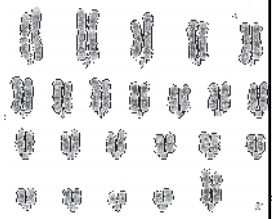
Sex chromosomes
XX = female
XY = male

Down syndrome



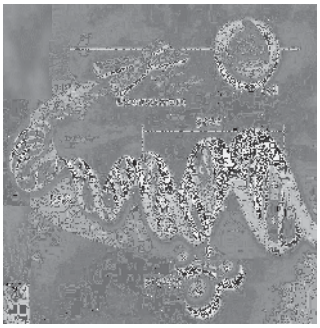
Chromosomes





Chromosomes are normal in CHARGE syndrome

DNA
the human genetic code

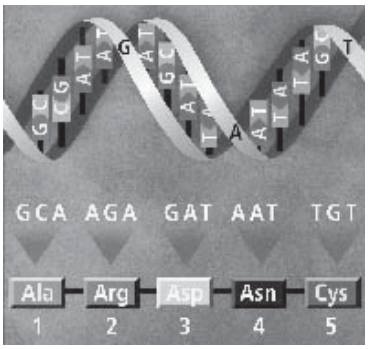


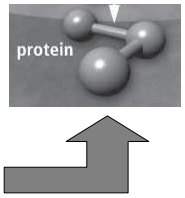
All human cells contain 46 chromosomes

On the chromosomes are 30,000 genes (DNA)

The genes encode for proteins

Genotype





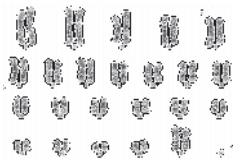
DNA alterations

Large chromosome abnormalties
- whole chromosomes
- parts of chromosomes

Small chromosome abnormalities
- small deletions/duplications
- Aberrations within genes

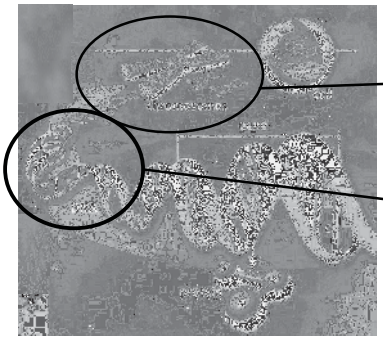
Visible through a Microscope

1000x



Not visible with a 1000x magnification

genetic aberrations



Microscopically visible
→Not in CHARGE syndroom

Microscopically not visible

Other techniques:
- DNA-analysis
- MLPA/array

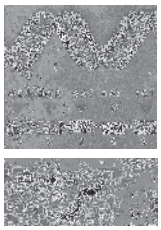
Genotype / Phenotype

Genotype

Phenotype

genes

"... genes code for proteins that are responsible for the correct functioning of our body..."

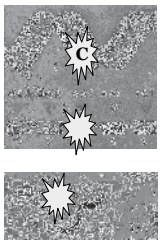


DNA: the code of a gene

Amino acids: the building stones of a protein

The protein

Mutation



Change in **one** of the characters of the gene

Change in **one** of the amino acids of the protein

Protein **not** able to function properly

Mutation – example 1

Tim and his toy

Tim and his toy

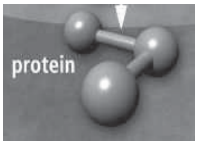
Tim ndh ist oy

Mutation – example 2

Tim and his toy

Tim and his toy

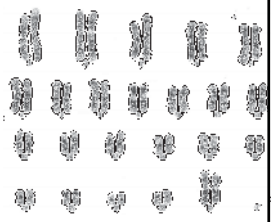
Tim and toy



Outline of the presentation

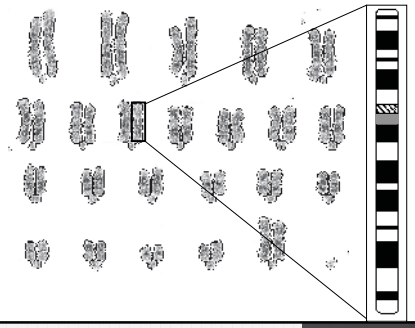
- ✓ **The clinical diagnosis of CHARGE syndrome**
- ✓ **chromosomes and genes**
- ✓ **the *CHD7* gene**
- ✓ **DNA-diagnostics in CHARGE syndrome**
- ✓ **Should parents be studied?**

Chromosomes



Chromosomes are normal in CHARGE syndrome

Genetics of CHARGE syndrome



CHD7 and CHARGE syndrome

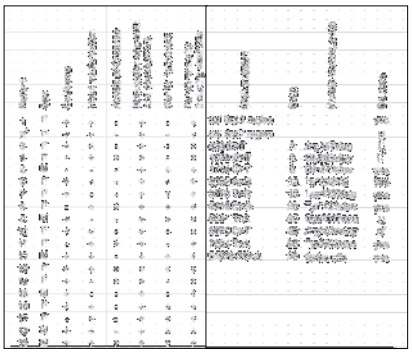
nature genetics

2004

Mutations in a new member of the chromodomain gene family cause CHARGE syndrome

Lisenka E L M Vissers[1], Conny M A van Ravenswaaij[et al]

CHD7 and CHARGE syndrome

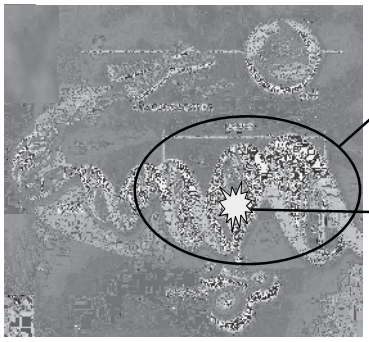


Absent or incorrect *CHD7* in 12/19 patients

↓

18/19

The CHD7 gene

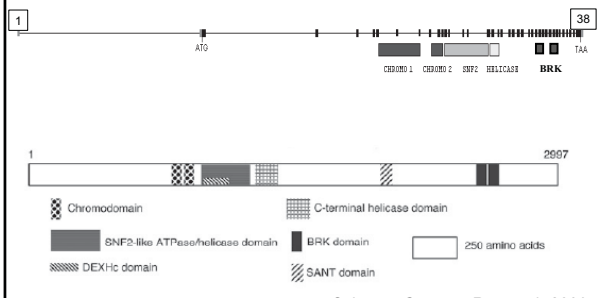


19 patients with CHARGE syndrome

2 patients with missing piece of DNA that contains CHD7

16 patients with an alteration in the CHD7 gene

CHD7



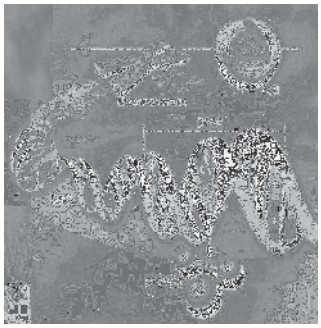
Schnetz, Genome Research 2009

Function of CHD7

- Many different organs show developmental defects

→ CHD7 has its function during the early embryonic development of the child, together with many other developmental genes

DNA: 30,000 genes the human genetic code



What is the function of our 30.000 genes?

Differences per cell type

Differences per developmental stage

→ Something must regulate gene expression

Function of CHD7

Other genes

Body,
first weeks of pregnancy

CHD7

CHD7: helps with the transcription of genes

CHD7

Responsible for the transcription of developmental genes

↓

Insufficient CHD7 may lead to insufficient transcription of developmental genes

↓

This may cause developmental defects in one or more organs

Outline of the presentation

- ✓ **The clinical diagnosis of CHARGE syndrome**
- ✓ **chromosomes and genes**
- ✓ **the *CHD7* gene**
- ✓ **DNA-diagnostics in CHARGE syndrome**
- ✓ **Should parents be studied?**

DNA diagnostics in CHARGE syndrome

Different kinds of alterations in CHD7

- Small changes -> sequencing (checking the CTAG-code)
- Large changes -> MLPA or array
- Chromosomal alteration -> microscopy

Small mutation – example 1

Tim and his toy

Tim ~~a~~nd his toy

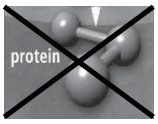
Tim ndh ist oy

Small mutation – example 2

Tim and his toy

Tom and his toy

X and his toy



CHD7 diagnostics

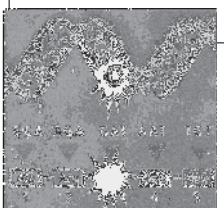
***CHD7* sequencing:**

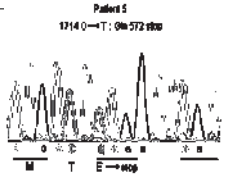
Mutation frequency: 65-75%

In typical CHARGE: > 90%

atcgttaccgtatcgaattg

-Gly-Leu-**Stop**





DNA diagnostics in CHARGE syndrome

Different kinds of alterations in CHD7

- Small changes -> sequencing (checking the CTAG-code)
- Large changes -> MLPA or array
- Chromosomal alteration -> microscopy

Large Mutation – example 1

---- Tim and his toy ---- Luc and his box --- Kim and her son ------ Jan and his dog and his cat ------- Dad and his hat ----

---- Tim and his toy ---- ~~Luc and his box~~ --- Kim and her son ------ Jan and his dog and his cat ------- Dad and his hat ----

Will be missed by sequencing

Large Mutation – example 2

---- Tim and his toy ---- Luc and his box --- Kim and her son ------ Jan and his dog and his cat ------- Dad and his hat ----

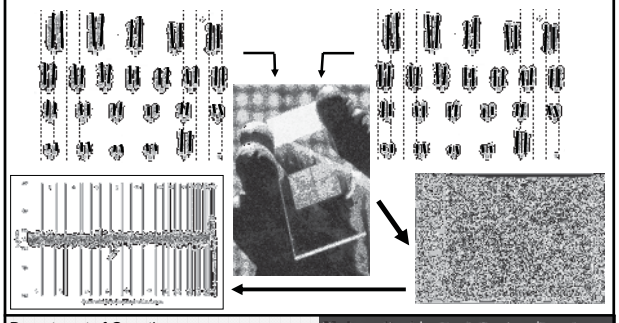
~~---- Tim and his toy ---- Luc and his box --- Kim and her son ------ Jan and his dog and his cat ------- Dad and his hat ----~~

Will be missed by sequencing

Comparative Genomic Hybridisation

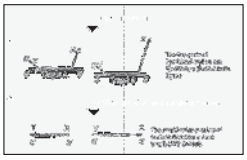
patient

controle



MLPA

(partial) deletions of *CHD7* (MLPA)



Deletions of *CHD7*

Dutch study:

MLPA of 54 CHARGE patients without *CHD7* mutation

Clinical information of patients was obtained through a written questionnaire

Deletions of *CHD7*

Patients:	Diagnostic criteria	
	Blake	Verloes
6	+	+
5	+	atypical
7	-	+
30	-	atypical
6	-	-
54	**18 typical** **30 atypical** **6 suspected**	

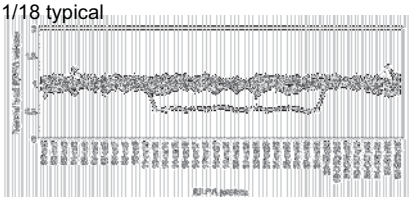
atypical Verloes: 2 majors or 1 major and 2 minors

Deletions of *CHD7*

Results:

One patient with deletion in CHD7

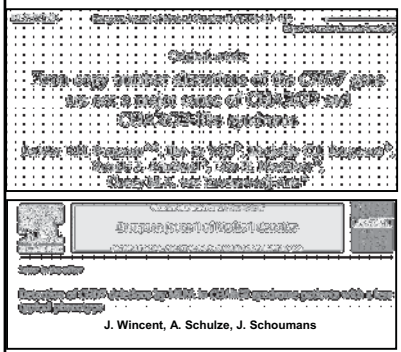
1/54 suspected, 1/18 typical



Conclusion:

small deletions in CHD7 are not a common cause of CHARGE syndrome

CHD7 diagnostics



J. Wincent, A. Schulze, J. Schoumans

Nijmegen:
302 mutation pos.
1 gene deletion
1 intragenic del.

4/18, 3 atypical

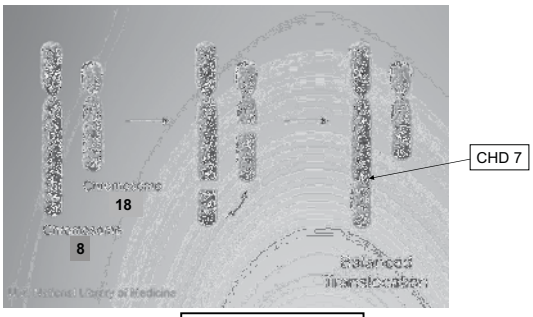
Always perform MLPA

DNA diagnostics in CHARGE syndrome

Different kinds of alterations in CHD7

- Small changes -> sequencing (checking the CTAG-code)
- Large changes -> MLPA or array
- Chromosomal alteration -> microscopy

translocation



Extremely rare !

The Dutch experience

450 patients suspected for CHARGE syndrome:

1 whole gene deletion	**(array CGH)**
1 intragenic deletion	**(MLPA)**
302 mutations	**(sequencing)**

Routine DNA diagnostis will pick up almost all mutations

If no mutation is found, MLPA and microscopy of the chromomes is recommended

CHD7 diagnostics

***CHD7* sequencing:**

Mutation frequency: 65-75%

In typical CHARGE: > 90%

***CHD7* MLPA:**

Whole gene deletions are extremely rare

Whole exon deletions in a few patients

CHD7 diagnostics

***CHD7* sequencing:**

Mutation frequency: 65-75%

In typical CHARGE: > 90%

***CHD7* MLPA:**

Whole gene deletions are extremely rare

Whole exon deletions in a few patients

Cytogenetics:

translocations involving 8q12 are extremely rare

Diagnostic criteria

Diagnosis: Doctor or DNA???

Aspecific CHARGE syndrome

- Feeding problems / tracheomalacia
- Heart defect: vascular ring
- Mild MR
- Height P3
- Dysmorphic ears
- Bilateral severe hearing loss
- Aplasia of semicircular canals

- No coloboma
- No choanal atresia
- No cleft lip/palate

- 5833C>T (R1945X)

Outline of the presentation

- ✓ The clinical diagnosis of CHARGE syndrome
- ✓ chromosomes and genes
- ✓ the *CHD7* gene
- ✓ DNA-diagnostics in CHARGE syndrome
- ✓ Should parents be tested?

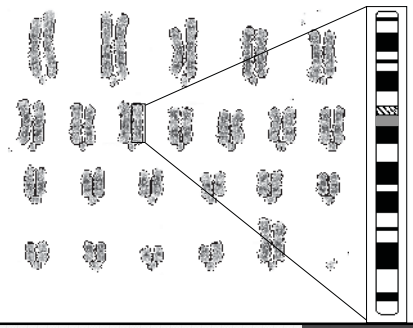
Familial CHARGE ?

How often do we see recurrence ?

- 1 in 10.000 newborns has CHARGE syndroom
- approximately 3% of them has a sib or parent with CHARGE syndrome
- often family members have a milder presentation

Why does recurrence occur ?

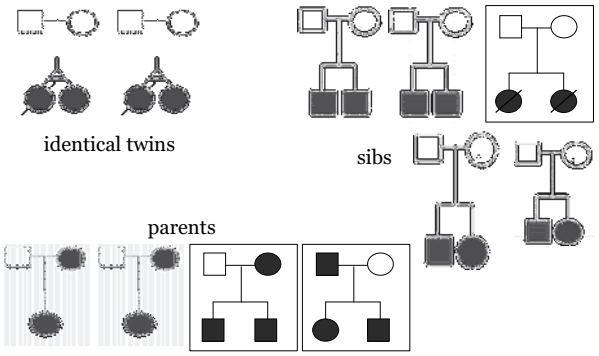
Chromosomes
are passed on from parent to child



Are changes in the CHD7-gene, inherited ?

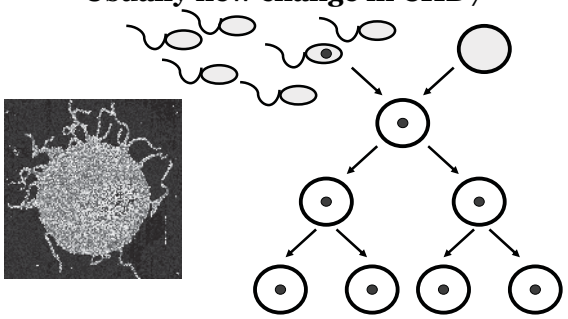
Usually not, however ...

CHARGE in families

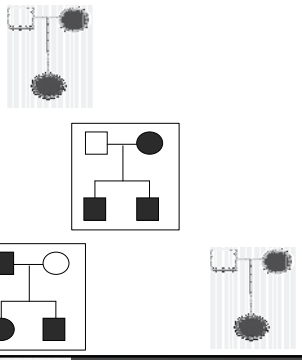


Cause of CHARGE syndrome

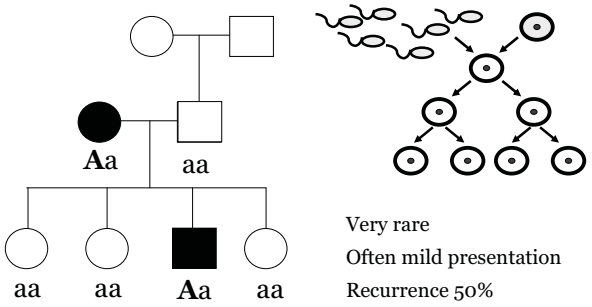
Usually new change in CHD7



Parent and child(ren)

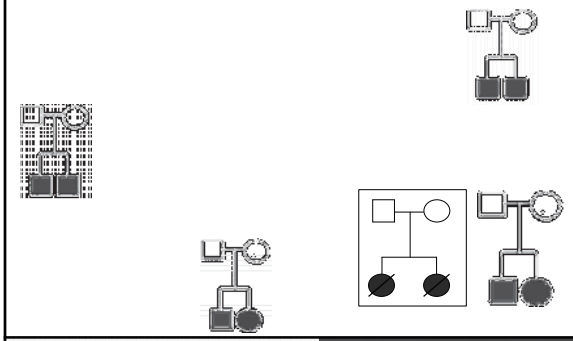


Parent and child(ren)



Very rare
Often mild presentation
Recurrence 50%

Sibs with normal parents



Familial CHARGE

Conclusions

- familial CHARGE syndrome is rare
- familial CHARGE syndrome often has a mild to moderate presentation, however
- variability within families is considerable
- CHD7 analysis is recommended in parents with child wish
- recurrence risk for children of affected parents is high

Conclusions

- *CHD7:* major CHARGE gene (> 65% *CHD7* positive)

  DNA is helpful in confirming the diagnosis

- most mutations are unique

  DNA analysis takes a long time

- Sibs: similar mutation → different features

  DNA does not predict the clinical problems

questions

c.m.a.van.ravenswaaij@medgen.umcg.nl

Familial CHARGE syndrome; is there a recurrence risk?

Since 2004 we know that CHARGE syndrome is caused by a change in the CHD7 gene. Every person has **two** CHD7 genes, one inherited from father and the other one inherited from mother. CHD7 is a regulatory gene. It regulates the expression of developmental genes very early during the development of the unborn child (embryo). If there is insufficient CHD7 the risk for developmental defects in specific organs like the heart, eye, ear, kidney, etcetera, is elevated (figure 1). A change in **one** of the two CHD7-genes is enough to result in CHARGE syndrome. However, CHARGE syndrome is highly variable and it is not possible to predict the clinical consequences for the child from the specific change in CHD7.

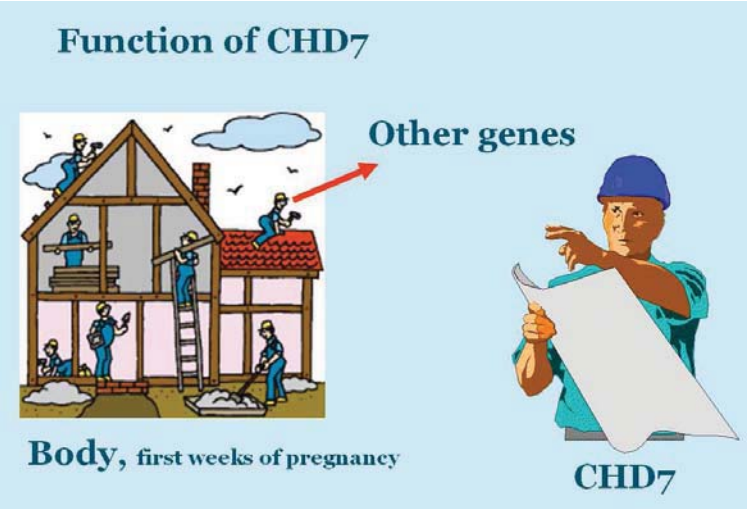


Fig. 1 CHD7 regulates the function of developmental genes during early pregnancy.

Two children with CHARGE syndrome within one family incidentally occur. CHARGE syndrome has a birth prevalence of approximately 1 in 10,000 newborns. About 3% of all persons with CHARGE syndrome have a sib or parent who also has CHARGE syndrome. We have collected as much as possible information of such families because this reveals important information on the variability of CHARGE syndrome. Within families all affected family members have the same change in CHD7, while their clinical problems may differ a lot. Moreover, we discovered that within families often an unexpectedly mild form of CHARGE syndrome can be found. In such families mildly affected persons were identified through a child with typical CHARGE syndrome.

In my presentation I will focus on why CHARGE syndrome sometimes occurs in more than one family member and what this recurrence risk is.

<u>Monozygotic (identical) twins</u>
CHARGE syndrome is almost always caused by a new change in CHD7. This means that the change occurred in the single egg or sperm cell that gave rise to the child. Thus the change is present in the fertilized egg. This fertilized egg will start dividing, billions of times, and in all cells that arise from the fertilized egg the CHD7 change will be present. Thus the CHD7 change will be present in all cells of the embryo, and subsequently in all cells of the child (figure 2).

If the early embryo divides into two embryos (monozygotic or identical twins), both children will have the same CHD7 change and thus they will both have CHARGE syndrome.

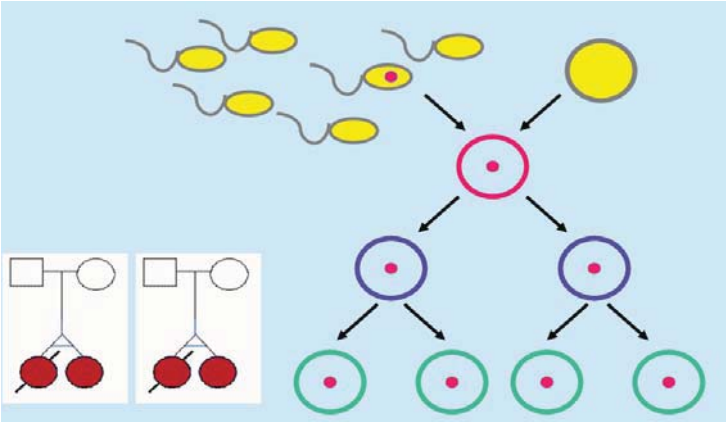


Fig.2 An egg is fertilized by a sperm cell containing a change in the CHD7 gene. All cells that arise from this fertilized egg will carry the CHD7 change. If two children develop from one fertilized egg they will share all genetic information and thus, in this situation, both will have CHARGE syndrome. The pedigrees show two of our families with monozygotic (identical) twins.

A parent with CHARGE syndrome

We collected four families with a mildly affected parent and one or two children with mild to moderate CHARGE syndrome (Figure 3). In all families the diagnosis was made initially in a child with clear symptoms of CHARGE syndrome.

The mildest consequences of a CHD7 change appeared to be abnormal shape of the ears and balance disturbances (abnormality of the semicircular canals). DNA studies of the CHD7 gene could confirm the diagnosis in the parent and, of course, the affected child has the same CHD7 change as the parent.

In these families the recurrence risk is 50% for each pregnancy. Parents always give half of their genetic information to their children. The parent with CHARGE syndrome thus passes on either the normal CHD7 gene or the changed CHD7 gene. This explains the 50% recurrence risk.

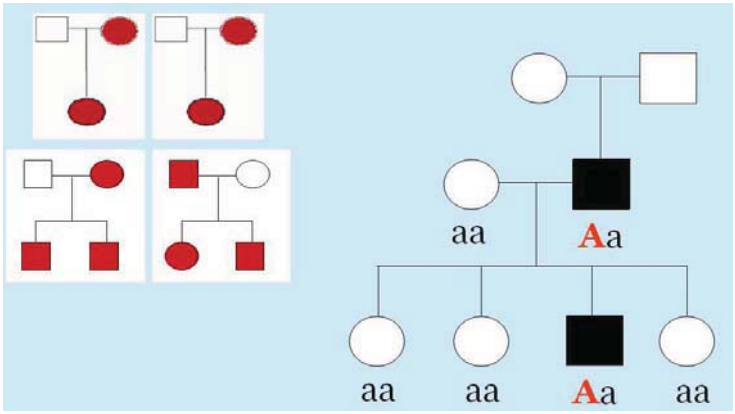


Fig. 3 Four families with parent-to-child transmission of CHARGE syndrome. The parent with (mild) CHARGE syndrome passes on the normal CHD7 gene (a) or the changed CHD7 gene (A). Thus the recurrence risk is 50% for each pregnancy.

Two children with CHARGE syndrome and healthy parents

In five families two sibs of different ages had CHARGE syndrome. Within each family the same CHD7 change was found in both affected children. Thus recurrence had occurred while the parents did not show any symptom of CHARGE syndrome. How is that possible?
In two of these families we showed that one of the parents carried the CHD7genes in part of his/her body cells. This is called a mosaic. A mosaic situation can occur if in the fertilized egg no CHD7 change is present, but this change occurs after a few cell divisions (figure 4). Only the cells that arise from the cell with the altered CHD7 gene will carry this change. If these cells are also present in the ovaries or testes, egg or sperm cells with the CHD7 change can be formed and, if these are involved in a pregnancy, a child with (non-mosaic) CHARGE syndrome will be born.

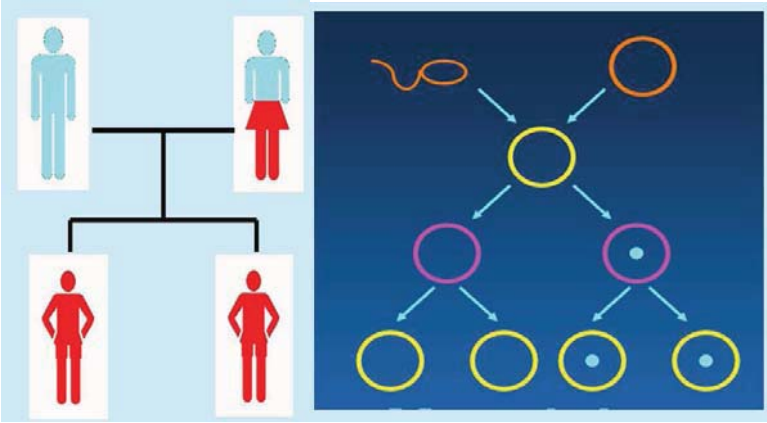


Fig. 4 In this example mother has a CHD7 change in part of her body cells (mosaic). The scheme on the right side explains that this can occur when the CHD7 change arises during a cell division *after* fertilization. Mother will not have CHARGE syndrome herself, but she has an increased risk for children with CHARGE syndrome (she can pass on the CHD7 change via an egg cell, and subsequently the child will have the CHD7 change in all cells).

In the other three families we did not reveal a mosaic in one of the parents. Nevertheless their affected children carried the same CHD7 change. The most likely explanation is that a low-grade mosaic is present in the ovaries or testes, but not in other body cells (or in such a low percentage that this cannot be detected by DNA studies). This is called a germline mosaicism.

What is the recurrence risk in CHARGE syndrome?

Familial CHARGE syndrome is extremely rare. This already demonstrates that in general the recurrence risk will be low. If parents have a child with CHARGE syndrome and want to be informed about future pregnancies we recommend the following:

- Investigate parents for mild symptoms of CHARGE syndrome (hearing, balance, smell, shape of the ears).
- If a CHD7 change has been found in the child, offer DNA analysis to the parents as well.

If the CHD7 change can be found in the parent, either in mosaic or all cells, than the recurrence risk is elevated (maximal 50%). If the parents want to, prenatal diagnosis can be performed in future pregnancies. However one should be aware that the severity of CHARGE syndrome can not be predicted by DNA-analysis.

If the CHD7 change can not be found in one of the parents (the most frequent situation), than a small risk for germline mosaicism remains. Therefore the recurrence risk is not zero, but 1 to 2 %. Also parents without CHD7 change may opt for prenatal diagnosis if they want to.

As mentioned before, the severity of CHARGE syndrome cannot be predicted by DNA-analysis. Fetal ultrasound examination can give extra information, for example on the presence of a heart defect. But not everything can be seen by ultrasound, for instance deafness, developmental delay and behavioural problems will remain undetected.
The choice for prenatal diagnosis will always remain a personal one, and the task and challenge of the clinical geneticist/genetic counsellor is to inform the parents in such a way that they can make the choice that fits them best.

Dr Conny van Ravenswaaij, MD PhD
Clinical geneticist
University Medical Center Groningen
c.m.a.van.ravenswaaij@medgen.umcg.nl

Information for New Families

FRIDAY
Breakout Session #2: 1:00 – 2:00 PM
Trillium Ballroom IV, Conference Center

CHARGE 102:

SENSORY LOSS: HOW DOES THAT CHANGE EARLY DEVELOPMENT?

Sandra L.H. Davenport, M.D.

SensoryGenetic/Neurodevelopment, Bloomington MN

Presenter Biography: Dr. Sandra Davenport is a Pediatric Geneticist and Developmental Pediatrician who specializes in sensory losses. She has a private consulting practice called Sensory Genetics/Neuro-development. She is currently a consultant to the Minnesota DeafBlind Technical Assistance Project and has been on its Advisory Council for 15 years and is also member of the Early Hearing Detection & Intervention and Newborn Screening committees through the Minnesota Department of Health. She has also been the President and Board Member of SENTAC (the Society for Ear, Nose and Throat Advances in Children).

Dr. Davenport was the chairman for two years of the Usher Consortium, a group of geneticists who were searching for the Usher genes and was involved in the research that led up to finding two of these genes. She was also a founding member of the CHARGE Syndrome Foundation board and is the co-author/editor of the original and updated CHARGE Manual for Parents and Professionals. For six years she directed the Vision Screening Project, a federally funded grant which helped identify students with dual sensory loss among those in deaf and hard-of-hearing programs in Minnesota and North Dakota.

Sandra L.H. Davenport, M.D., SensoryGenetic/Neurodevelopment, 5800 Southwood Drive, Bloomington MN 55437-1739; TEL: 952-831-5522; EMAIL: slhdaven@tc.umn.edu

Presentation Abstract: Development: How do all those features of CHARGE influence development? What are sensory deficits? What does DeafBlind mean? What does balance have to do with it? What about weakness? How well can my child do?

CHARGE 102: PHYSICAL INFLUENCES ON DEVELOPMENT IN CHARGE

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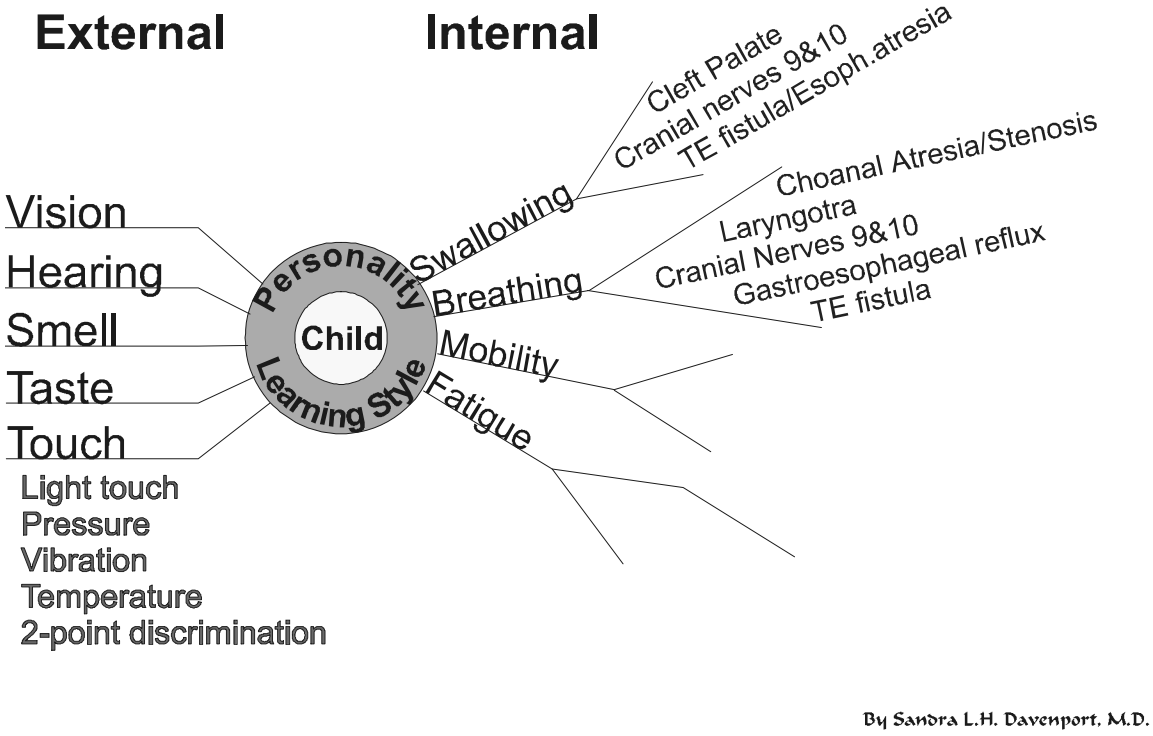


CHARGE is such a complex disorder that many of the physical components have a profound effect on the child's development and understanding of the world. The above diagram breaks these influences down into those that are External and those that are Internal. In one way all are internal since the five 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.

A more complete discussion of this is in the Manual

INFLUENCE OF SENSORY LOSS ON DEVELOPMENT: The Communication Bubble

Are all five major senses working?

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. 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.

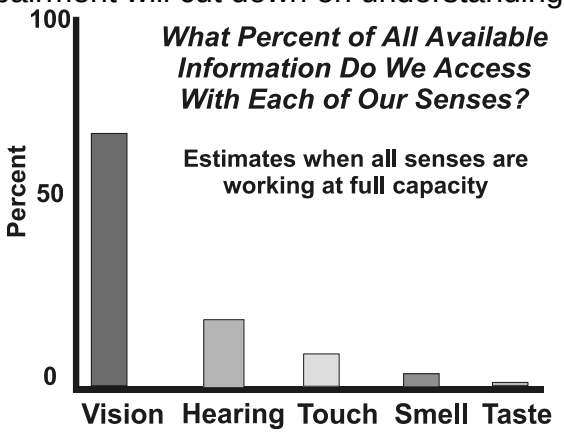
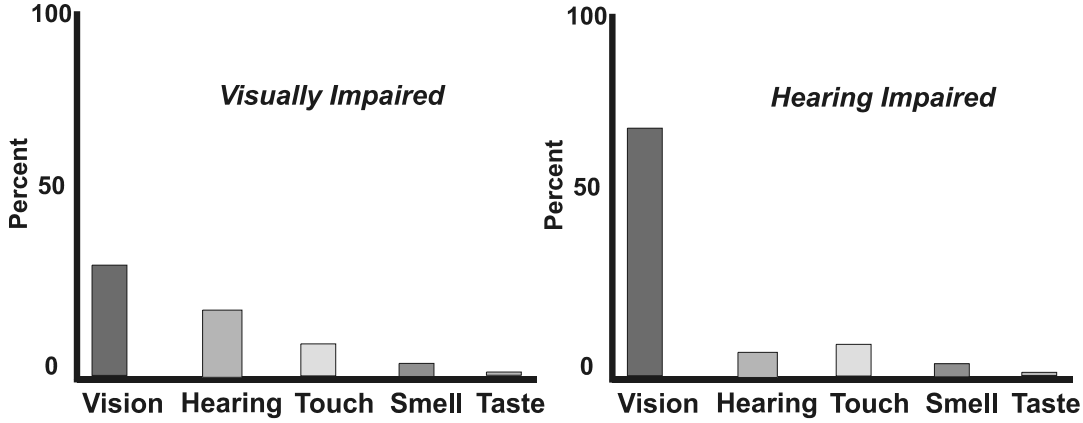


Figure 1: Full access to information from all senses.

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.

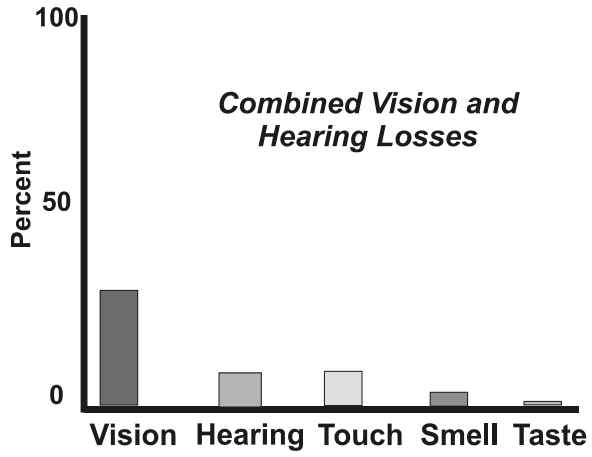


Figure 4: Combined vision/hearing loss with half of each gone.

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. 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 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.

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.

PHYSICAL EDUCATION

FRIDAY
Breakout Session #3: 2:10 – 3:10 PM
Trillium Ballroom I, Conference Center

RESOURCES FOR PHYSICAL EDUCATION, SPORT & FITNESS FOR CHILDREN WITH CHARGE SYNDROME

Lauren J. Lieberman, Ph.D.
State University of New York at Brockport

Presenter Information: *Dr. Lauren Lieberman* is currently a professor of Adapted Physical Education at The College at Brockport in Brockport, NY where she teaches undergraduate and graduate courses. Before teaching at Brockport she taught at The Perkins School for the Blind in the Deafblind Program. Her area of expertise is working with children with visual impairments and deafblindness. She has co-authored several books in this area such as "Games for People with Sensory Impairments" now in its 2nd edition through the American Printing House for the Blind, and Going PLACES: Transition Guidelines for Community Based Physical Activities for Youth with Visual Impairments, Blindness or Deafblindness also through the American Printing House for the Blind. She also co-authored "Strategies for Inclusion" now in its 2nd edition through Human Kinetics Publishers. She works very closely with the American Printing House for the Blind developing products to promote physical activity.
Lauren founded and directs Camp Abilities: A developmental sports camp for children who are visually impaired, blind or deafblind now in its 14th summer. The camp is in Brockport, NY but other camps can be found throughout the country and Guatemala. See the web site for more information at www.campabilitiesbrockport.org.

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EMAIL: Llieberm@brockport.edu

Presentation Abstract: Children with CHARGE Syndrome are often medically fragile, and have ongoing health issues. Often times parents, teachers and physicians do not believe they can be physically active within their schools and communities. The lack of physical involvement can have detrimental effects on their physical, social and emotional growth. This presentation will give a variety of resources so teachers, caregivers and physicians can help them access a physically active and social lifestyle regardless of their medical involvement.

Resources for Physical Activity for Children with CHARGE Syndrome

CHARGE Syndrome Conference

July 2009

Lauren J. Lieberman Ph.D.

SUNY Brockport

Department of Physical Education

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Achievement is the Goal

NOT only Participation

Programming

- Placement is an important decision related to physical education
- It is often necessary to have a combination of general physical education and segregated physical education so the child does not feel behind

Physical Education Regulations

- Children with CHARGE Syndrome should learn the same units as their peers.
- Modifications to equipment, rules, instruction and the environment may be necessary.
- They must be taught by a qualified professional
- The paraeducator should go to PE with the student and should be trained

Include PE, Recreation, & Leisure on IEP of Children with CHARGE Syndrome

- Assessment-Brockport Physical Fitness Test (10-17 years of age)
- PLP-include the child when possible
- Set goals for school and at home
- Make time at school and at home to work on goals and objectives
- Assess whether goals are met

Resources

- Products
- Web sites
- Curriculums/Books
- Camps

Walk-Run for Fitness Kit

- Purpose: to help children with VI or who are deafblind learn to walk and run for fitness
- Included
- Backpack with:
- 2 talking pedometers
- Tether
- Guidewire
- Walk-run for fitness manual
- Produced by The American Printing House for the Blind (APH.org)

Jump roping study

- Ropeless jump rope
- Jump rope with hula hoop

- Beaded rope
- Plastic jump rope

- Foam mat with slanted edges

Jump rope kit

- Purpose: To teach children how to jump rope
- Three types of ropes
- Ropeless rope
- Beaded rope
- Rope with hula hoop
- Mat
- Jump rope manual
- Teaches skills
- Goal setting
- Socialization
- Through AHP.org

Beeping Balls (Sound balls)

- Balls that beep
- Nerf material
- Affordable
- Durable
- Accessible
- (Also have Rib-it balls that have texture and make some sounds)
- See the site APH.org click on physical education

Nutritional Education

- My Pyramid curriculum

- Tactile My Pyramid as a puzzle for elementary age
- My Pyramid as curriculum for secondary students
- APH.org

Dance Dance Revolution

- DDR is a popular psychomotor video game used by children world wide
- Modifications for children with CHARGE Syndrome
- What: Arrows on a screen the child follows and steps on the directional arrow that is presented in front of them
- Modifications:
- Screen plain background with arrows only
- Slow the arrows down
- Make the arrows bigger
- Give verbal cues
- Give auditory cues
- Make into a pattern

Instructional web sites

- Project INSPIRE www.twu.edu/INSPIRE
- Camp Abilities www.campabilities.org
- Video: Teaching children with VI, blindness, or deafblindness in physical education
- USABA www.usaba.org
- Texas School for the Blind
- American Printing House for the Blind
- APH.org
- Other?

Visual Impairments & Deafness
***Includes children with CHARGE Syndrome**

- In J.P. Winnick (2005/10) Adapted Physical Education and Sport Human Kinetics Publishers www.humankinetics.com

  1-800-747-4457

Brockport Physical Fitness Test

- Health Related Criterion Referenced Fitness Test
- Ages 10-17
- Assesses all 5 areas of health related fitness
- Has standards for children and youth with visual impairments
- Sold through Human Kinetics 1-800-747-4457
- www.Hkusa.com

"Everybody Plays": How Children with Visual Impairments, Blindness or Deafblindness Play Sports

- Elementary age children's book
- Profiles of 16 sports and recreation activities with interviews of children who do them
- Forward by Erik Weihenmayer
- Advice from Elite Athletes by:
  - Tricia Zorn-Swimming
  - Jim Mastro-Judo
  - Marla Runyan-Running
  - Jen Armbruster-Goal ball
  - APH.org

Going PLACES: A Transition Guide to Physical Activity for Youth with Visual Impairments or Deafblindness

- Transition guide to physical activity
- Youth 14-22
- Lieberman, Modell,, & Ponchillia (2006)

- American Printing House for the Blind
- APH.org

Games for People with Sensory Impairments

- Games, sports, recreation and aquatics for children with sensory impairments
- Includes modifications to all
- Lieberman & Cowart APH.org

"Strategies for Inclusion" 2nd edition

- Practitioner book on how to include children with disabilities into physical education
- Comes with curriculum modification ideas for children with sensory impairments.
- Comes with peer tutor training curriculum
- By Lieberman & Houston-Wilson (2009)
- Sold through Human Kinetics 1-800-747-4457
- www.Hkusa.com

Camp Abilities

- A developmental sports camp for youth with visual impairments, blindness, and deafblindness
- Track & field, swimming, tandem biking, goal ball, beep baseball, gymnastics, judo, fishing, canoeing, kayaking, rollerblading, horsebackriding, rock climbing, basketball, dancing and more
- Camp Abilities
    - Camps now available in:
    - Brockport, NY
    - Long Island, NY
    - Tucson, AZ
    - Anchorage, AK
    - Baltimore, MD

- Ft. Myers, FL
- West Chester, PA
- Puerto Rico
- Guatemala City, Guatemala
- San Francisco, CA

Sports Camp Start Up Manual

- Perkins School for the Blind
- General manual to help start sports camps
- Applications, forms, fund raising, orientation, etc.

Additional Ideas or Questions?

Sensory Information

FRIDAY
Breakout Session #3: 2:10 – 3:10 PM
Trillium Ballroom I, Conference Center

IMPACT OF COCHLEAR IMPLANTS FOR CHILDREN WITH CHARGE SYSNDROME: PRELIMINARY FINDINGS

Susan M. Bashinski, Ed.D.
Associate Professor – Special Education,
Curriculum & Instruction Department, East Carolina University
and
Kathleen Stremel Thomas, M.A.
Director, National Consortium on Deafblindness (NCDB),
Western Oregon University

Presenter Information:

Dr.Bashinski has been working in the field of special education for more than 35 years, teaching in public school Pre-K through high school programs, as well as at the IHE level. She is currently a member of the faculty at East Carolina University. Susan has written numerous research articles, book chapters, and manuals associated with topics relevant to learners who experience significant support needs, including deaf-blindness. She has directed numerous federal and state grants in low-incidence disabilities. Susan has extensive experience providing professional development and technical assistance across the United States and internationally, particularly in the areas of language and communication development, augmentative communication, and nonsymbolic communication intervention strategies for learners who have low-incidence disabilities, including deaf-blindness.

Kathleen Stremel Thomas (Kat) has worked in the area of communication & language assessment, intervention, generalization and evaluation for students with severe disabilities, including deaf-blindness, for the past 39 years. She has written numerous book chapters and articles and continues to conduct workshops nationally and internationally. Kathleen's primary experience includes working with infants, toddlers, and young children within natural environments. She served as the Director for the National Consortium on Deaf-Blindness for the past 11 years. Kat is now involved in Cochlear Implant research and intervention for children who are deaf-blind with Cochlear Implants.

Presentation Abstract: Presenters will share preliminary findings from their ongoing research study investigating outcomes for children with deaf-blindness, who have received a cochlear implant. Participants in this study are diverse; the second largest group, by etiology, is children with CHARGE syndrome. In addition to sharing research findings they have to date, researchers will present suggestions parents might utilize to promote their children's communication development and listening skills. Sound inventories for home, school, and community environments, developed by the researchers, will be discussed.

IMPACT OF COCHLEAR IMPLANTS FOR CHILDREN WITH CHARGE SYNDROME: PRELIMINARY FINDINGS

International CHARGE Syndrome Conference
July 23- 25, 2009

Presented by:
Susan M. Bashinski
Kathleen Stremel Thomas

Department of Education
Office of Special Education Programs
(Grant # H327A050079)

to The Teaching Research Institute, University of Kansas &
Midwest Ear Institute

Presentation Objectives

Parents should leave this session with:

1. suggestions for interacting with their child in ways that will help to promote communication development
2. ideas regarding how they might help their child *learn to use* auditory input, within natural environments
3. suggestions for establishing a "listening environment"
4. Information that might be helpful when considering whether or not to seek a cochlear implant for their child

Participating States

• **Arizona**	• **Massachusetts**
• California	• Mississippi
• Connecticut	• Missouri
• Delaware	• Nebraska
• Florida	• New York
• Georgia	• North Carolina
• Illinois	• Ohio (CCHMC)
• Kansas	• Oklahoma
• Louisiana	• Oregon
• Maryland	• Pennsylvania
	• Texas

Participants with CHARGE Syndrome

- A total of 93 children are participating in this study (with at least one assessment)
- Of this number, 16 participants have CHARGE (the second largest group, by etiology)
- Of the total 93 participants, the number of implants has been reported for 76 children; 11 of these have bilateral implants (14.5%)
- Of the 16 participants who have CHARGE, 3 have bilateral implants (18.8%)

Participants with CHARGE

Gender	*n*	%
Female	3	19
Male	13	81

Participants with CHARGE

Race	*n*	%
African American	3	19
Caucasian	11	69
Latino	2	13

Participants with CHARGE

Age at first implant	*n* (of 14)	%
≤ 12 mos.	3	21.4
13–24 mos.	2	14.3
25–36 mos.	4	28.6
37–48 mos.	3	21.4
49–60 mos.	2	14.3
> 5 years	0	0

Age at which Participants Received Their First Implants

- The average age at which **ALL study participants received a first implant was 36 months (i.e., 3.0 years)**
- **The average age at which study participants with CHARGE syndrome** received a first implant was **30.7 months** (i.e., ~2 years, 7 months)

Participants with CHARGE (with implant age data, n = 14)

	Age of child (months)	Age at Implant (months)	Time in Sound (months)
Mean	70	30.7	39.1

Auditory Development

- Research regarding auditory brain development *should* guide the way we teach children to listen and to use auditory input
- We hear with our brains, not our ears
- A child's brain must be accessed and stimulated in order to develop (Cole & Flexer, 2007)
- Acoustic accessibility of intelligible speech is essential for brain growth
- We are either "growing" the brain or we are not ..!

Research on CI and Children with Multiple Disabilities

- Edwards (2007) reviewed the limited research currently available regarding cochlear implants and children with multiple disabilities
- General findings include:
  - Cognitive functioning is one of the strongest predictors of progress in developing speech perception and speech production
  - Parents report satisfaction with CI due to increased eye contact, awareness of the environment, and response to requests

Observations, to Date

- Younger participants in study, as a group, received their implants at earlier ages
- Participants in study, who had more additional disabilities, as a group received their implants at older ages
- Population of children with DB is extremely diverse; receiving a cochlear implant is *not* associated with any particular etiology

Observations, to Date

- A number of children with deaf-blindness (including CHARGE), who receive implants, do not have pre-linguistic skills
- A child's early communication skills, auditory and speech perception, speech development, and language development must be assessed so the habilitation program can be individualized
- Though a child may receive diagnostic therapy, she needs to learn to *use auditory skills* in authentic environments
- What do we want as outcomes? Are we willing to do what it takes?

Observations re: Outcomes

- Tremendous variability appears to exist in outcomes for children with CHARGE, who receive a cochlear implant

Optimal Outcomes appear to be associated with:

- Children wearing the implant(s) during all waking hours
- Children having their implant(s) mapped frequently
- Interventions focusing on the *auditory* signal
- Intervention focusing on family-child interactions, with an interventionist as "coach"

Preliminary Finding: Age at Implant

The impact of the AGE at which the child received his / her first cochlear implant **was significant on skill development**, as measured by the *Reynell-Zinkin* assessment…

Preliminary Finding: Age at Implant

- Participants who received the first implant at an earlier age appear to demonstrate increased:
  - response to sound
  - verbal comprehension
  - expressive language
  - social adaptation skills
- That is, the **earlier** a child received his / her first implant appears to be associated with **more rapid, significant progress in these areas.**

Preliminary Finding: Time in Sound

The impact of the amount of **TIME IN SOUND** (that is, the length of time the child has had a cochlear implant) **was significant on skill development**, as measured by the *Reynell-Zinkin* assessment…

Preliminary Finding: Time in Sound

- Participants with longer time in sound appear to demonstrate increased:
  - response to sound
  - verbal comprehension
  - expressive language
  - social adaptation skills
- That is, the **more time** a child has been receiving auditory input via a CI appears to be associated with **more rapid, significant progress in these areas**.

Preliminary Finding: Degree of Vision Loss

Although NOT statistically significant, the assessment **scores* for participants with total blindness were lower** than those for children who were reported to have some functional vision—**when scores WERE controlled for age**

*as measured by the *Reynell-Zinkin* assessment

Case Studies – Children with CHARGE Syndrome

See PowerPoint slides
for detailed data display

Examples of What We Need to Teach

- Pre-linguistic communication (a *necessary, but not sufficient* condition for auditory development)
- Differing responses to familiar speech
- Differing responses to environmental sounds
- Differing responses to music
- Differing responses to unfamiliar speech
- Expanding receptive vocabulary and receptive language comprehension

Examples of What We Need to Teach (con't)

- Use of vocalizations as communication
- Differing levels of vocal imitation
- Use of vocal inflection and intonation
- Expanding expressive vocabulary
- Use of expanded expressive language
- Use of speech to communicate
- Use of intelligible speech

Facts

- Many children with DB, who receive a cochlear implant, do not receive intervention or therapy specific learning to listen / use the implant
- Variability in child outcomes indicates the need for individualized and adaptive approaches across receptive and expressive language, as exemplified by:

Auditory – Verbal Continuum

A…..AV…..AV…..AV…..V

(Nussbaum, Scott, Waddy-Smith, & Koch, 2006)

- Children who are deaf-blind might need more than programming across an Auditory – Verbal continuum to support their communication growth

How Do We Teach?

- Establish a "*listening environment*"
- Coach families
- Use auditory – verbal techniques
- Be conscious of the sign – oral continuum
- Use natural routines and activities to embed opportunities for listening and communicating

"Auditory Sandwich"

<u>Definition</u>:

- During an interaction with a child who has received an implant, lead with auditory stimuli
- WAIT for a response from the child
- Implement visual, tactile, and / or kinesthetic cues the child needs for support
- Include spoken language directly in the interaction with the child, *after* other modality cues (Nussbaum, Scott, Waddy-Smith, & Koch, 2006)

EXAMPLE: "Auditory Sandwich"

1. Say, "Get your bib…"
2. WAIT for a response to this verbal cue
   (IF no response, then…)
3. Say, "Get your bib" while pointing or gesturing in the direction of the bib
4. Say, "You have your bib!"

Establishing a Listening Environment

- Position oneself to best interact with the child, in the specific routine
- Use speech that is rich in melody, intonation, and rhythm
- Speak at typical volume
- Minimize all background noise
- Use speech that is repetitive
- Use acoustic "highlighting" techniques

(Estabrooks, 2001)asc ssss

SAMPLE: Home & Community Environments Inventory

Child's Name: Ashley **Sex:** Female
Birthdate: 11/11/1989 **Today's Date:** 11/5/2009

Directions: Please check the sounds that are in your environment (on the left-hand side). Columns on the right-hand side can be used as an assessment to determine your child's detection and identification of specific sounds.

Sounds in Your Home & Community Environment	Your Child's Responses to the Sounds		Motivating Sounds to Target for Learning
	Detection	Identification	
Home Environment – Kitchen and Utility Room:			
X Microwave bell	☐	☐	A good majority of the sounds are only identified in context and Ashley following who is doing the task.
X Oven door opening/closing	X	☐	
X Oven/egg timer	☐	☐	
X Oven temperature setting (beeps)	X	X	
X Refrigerator opening/closing	X	X	
X Drawer opening/closing	X	X	
X Dishwasher	☐	☐	
X Toast popping up in toaster	X	X	

Inventory of Sounds in Home and Community Environments

See Handouts for copy of this sound inventory

SAMPLE: School & Educational Environments Inventory

Child's Name: Ashley **Sex:** Female
Birthdate: 11/11/1989 **Today's Date:** 11/5/2009

Directions: A child's educational team should work together to identify sounds available in various school environments (left-hand side). Columns on the right-hand side can be used as an assessment to determine the child's detection of, and attention to, specifi sounds.

Sounds in Your Child's School / Educational Environments	Your Child's Responses to the Sounds		Motivating Sounds to Target for Your Child to Learn
	Detection	Identification	
Physical Contexts / Settings:			Attention to sounds in general ed classroom; arrangement of open environments to promote sound detection in limited spaces.
X gymnasium	☐	☐	
X corridor	X	X	
X playground	☐	☐	
X general education classroom	X	☐	
Sound Sources:			
X human - adult	X	☐	
X human – child	☐	☐	
X mechanical (toy, fire / tornado warning)	X	X	
X digital (recorded speech, voice output)	☐	☐	

Inventory of Sounds in School and Educational Environments

See Handouts for copy of this sound inventory

Other Activities for Suggesting to Families

- Maintain a joint focus on objects and activities
- Play ritualized games
- Sing and read nursery rhymes (younger children)
- Name objects in the environment
- Describe the location of objects
- Call the child's attention to environmental sounds
- Read to your child
- Play music and instruments

Key Findings from Family Survey

See PowerPoint slides
for detailed data display

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THANK YOU!

We appreciate your interest and attendance!

If you have questions, or would like additional information, please don't hesitate to contact us:

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Legal Information

FRIDAY
Breakout Session #3: 2:10 – 3:10 PM
Trillium Ballroom III, Conference Center

SPECIAL NEEDS FUTURE PLANNING: TRUSTS, GOVERNMENT BENEFITS, GUARDIANSHIP, ETC.

Brian Rubin

The Law Offices of Brian Rubin & Associates
Buffalo Grove, IL

Presenter Information: See last two pages of Brian Rubin's handout.

Presentation Abstract: Mr. Rubin is an attorney whose practice is devoted to services for individuals with special needs. He will present on the topics listed in the presentation title as well as others.

9th International CHARGE Syndrome Conference, Indian Lakes Resort, Bloomingdale, IL, July 24-26, 2009


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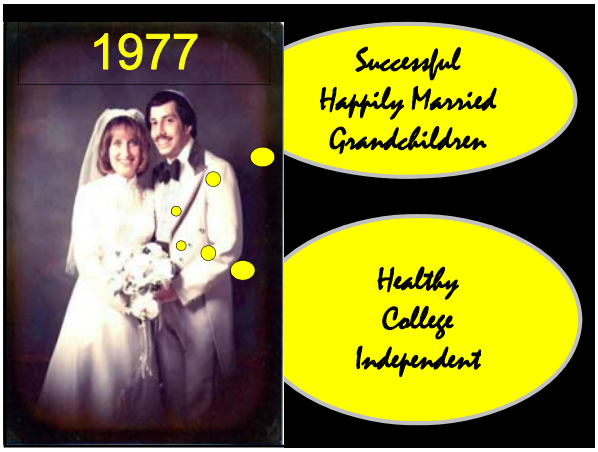
MEMBER
Special needs Alliance
Special needs require special attorneys.
www.specialneedsalliance.org
The 9th International CHARGE Syndrome Conference
Bloomingdale Illinois July 2009
CHARGE
...And All That Jazz


PLANNING FOR CHILDREN & ADULTS WITH SPECIAL NEEDS
for today...
for tomorrow...
& for the future.
Brian Rubin
Attorney & Father of a son with Autism


1977
Successful
Happily Married
Grandchildren
Healthy
College
Independent


4
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PERSPECTIVE
What's important...
PRIORITIES
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The Parents' Prayer
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What YOU
or OTHERS
do NOW...
impacts future
government
benefits...

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WHEN / WHY?

- When are you going to die?
- Gifts and/or inheritances from others?
  - intentional?
  - unintentional?

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How much ?

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WHERE FROM...?

Who will pay?

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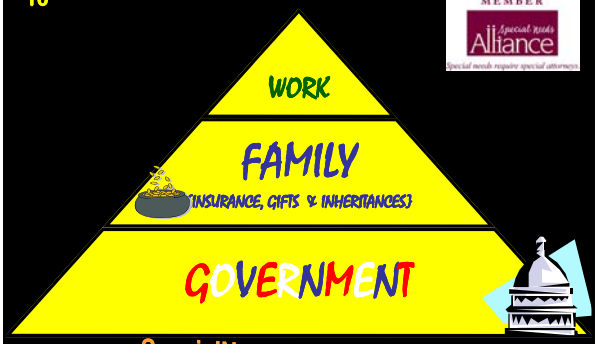

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Government Benefits

$ SSI	Medicaid THE TICKET!
$ DAC/SSDI	Medicare

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How to lose these benefits?

1) 10 day rule on changes such as:
   - Living Arrangement
   - Working
   - Inheritance/Gift
2) Redeterminations
   - Disability
   - Assets/Income
3) Inheritances/Gifts

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215 ILCS 5/367b
PARENTS' GROUP
HEALTH INSURANCE!

- Dependent children NOT subject to "limiting age" provisions!
- Even if living "residentially"!
- Since 1969... revised 1997
- NOT Self Insured Plans!

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WRONG WAY

"Directly"

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WRONG WAY

If no Will..

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WRONG WAY

"Moral Obligation"

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WRONG WAY

"Traditional"
SPENDTHRIFT
SUPPORT TRUST

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The Answer is...

SPECIAL NEEDS TRUST(S)



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☑ 2 Versions...

- ☑ Reserve
- ☑ Supplemental Fund
- ☑ Over & Above Benefits
- ☑ No $ Limit
- ☑ Federal
- ☑ State

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3rd Party

INHERITANCE

GIFT

INSURANCE
IRA
401K

SPECIAL NEEDS TRUST

THEN UPON PASSING... YOU DECIDE:
FAMILY
CHARITY
GUARDIANS

Beneficiary's own assets

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WRONG WAY

No! No! Not!

"inside"

your Will or Living Trust!

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Why ?

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Won't exist until...

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So NO

"Grand Fathering"

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So NO "Piggy Backing"

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Just what is a Trust?

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Just another way of holding "title" to, or "ownership" of, assets, investments or accounts...

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3rd PARTY SPECIAL NEEDS TRUST

So what's all the paper for?

- ✓ Instruction Manual
- ✓ "Rules of the Road"

Who can use it?

- ✓ Parents
- ✓ Siblings
- ✓ Grandparents
- Anyone...
- ✓ EXCEPT BENEFICIARY

Remember - 3rd Party

- ✓ Parents & Family as Trustees...

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EXTENDED FAMILY

- Instruction Letter
- Form Codicil
- Form Trust Amendment
- Trust Certification
- Consult with their Attorneys/Advisors

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But... Beware of absentee parents & grandparents

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EX- SPOUSE LETTER

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So...

- 1st set up SNT
- 2nd reference in Wills/Trusts
- 3rd extended family too

But... But...

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Change Beneficiaries

Life Insurance, IRA,
401k, Profit Sharing...

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NURSING HOME EXCEPTION ?

BUT BEWARE...

Power of Attorney
&/or Living Trust
MUST specify!

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Beneficiary HAS assets

UTMA Accounts
Settlement
Large Gift
Inheritance
Savings Bonds

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If "not that much"

...consider spending it down on allowed expenditures, such as:

- ☑ COMPUTER, TV, DVD PLAYER...
- ☑ VACATION
- ☑ PRE-PAID FUNERAL
- ☑ PAY OFF DEBTS... EVEN TO PARENTS...
- ☑ CLOTHES
- ↘ BUT MUST BE FOR HIS/HER SOLE BENEFIT...

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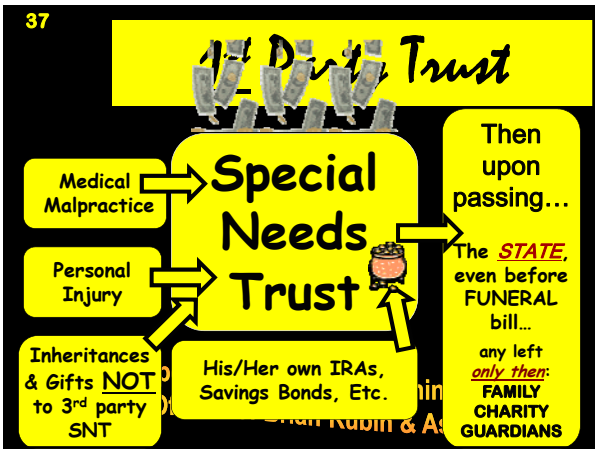

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1st Party SNT

MEMBER Alliance Special needs Special needs require special attorneys

Called… "Self Settled"… or

- ☑ "OBRA 93 Trust"… or
- ☑ "(d)(4)(A) Trust"… or
- ☑ "PAY BACK Trust"

because…

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1st Party SNT "NOT" SO GOOD…

- Only if under 65
- Only by Parents, Grandparents, Courts & Guardians…
- SSA Position…
- Courts often require…
- Pay Back…

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1st Party SNT

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- Irrevocable
- Sole Benefit
- Careful Funding
- Child Support →

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Divorce…

CHILD SUPPORT

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COURT ORDER MUST PROVIDE THAT POST 18 CHILD SUPPORT PAYMENTS PAYABLE FOR THE CHILD WITH SPECIAL NEEDS, SHALL BE PAID TO, & SHALL BE SUBJECT TO THE PROVISIONS OF, A 1st PARTY, SELF SETTLED, SOLE BENEFIT, IRREVOCABLE, PAY BACK SPECIAL NEEDS TRUST!

1st Party SNT

2nd Version... POOLED TRUST

ALSO for Beneficiary's OWN Assets...

Why... when needed ?

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POOLED TRUST

called "(d)(4)(C) Trust"

- IF age 65 {some states}
- IF NO Parent, Grandparent nor Guardian..., and don't desire to have Judges in control.
- Smaller sum of money.

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"Issues" with (d)(4)(C) Pooled Trust

- Not for Profit MUST...
- Establish & manage...
- Not free...
- Still "Pay Back" unless...

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What about GIFTS?

☒ NOT Directly

☒ NOT UTMA

YES 3rd Party SNT

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COMMON ATTORNEY MISTAKES!

- ☒ No "Reformation" Clause
- ☒ No "Parachute" Clause
- ☒ "Food and Shelter"
- ☒ "To" and not just for "the benefit of"
- ☒ "Next Generation Trustee"
- ☒ "Contributions" & "Gifts" in 3rd party SNT
- ☒ "Boilerplate" provisions in other documents
- ☒ Extended Family "OUTREACH"... NOT JUST SIBS!

➢ "Irrevocable" for 1st party SNTs & in some cases required in 3rd party SNTs.

➢ Sole Benefit for 1st party SNTs & in some case required in 3rd party SNTs: such as for Nursing Home exceptions; QDT; Public Pensions.

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You say that you already have a special needs trust?

1. Does it qualify?

2. Was it approved by SSI & your State Medicaid Agency)?

3. Will it be approved when it is submitted (It must be submitted)?

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Find an experienced special needs planning attorney...
Try, in order:

1. www.specialneedsalliance.org, then
2. www.specialneedsplanners.com, then
3. www.naela.org, then
4. Advocacy Groups, Support Groups, other parents, etc.

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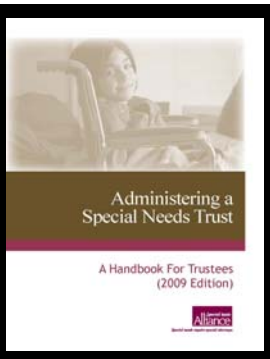

Email Brian for a Copy (PDF file).

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OK...
But I have other children?

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So how do I
DIVIDE the pot?
SLICE the pie?
Head vs. Heart

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Choosing Guardians...

Let me live... one moment longer...

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Choosing Guardians...

Who will make all of the necessary decisions?
- Medical?
- Government Benefits?
- School Issues?
- Recreation & Social?
- Employment?
- Day Programs?
- Residential?

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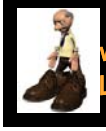
Choosing Guardians...

Who is going to fill our shoes?

- Can they understand?
- Job will outlast them!
- Other Children? Siblings? Grandparents? Friends?
- School District of Guardian?
- Married Couples?
- Same as Trustees?



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Guardians are nominated in parents Wills!

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What happens after my child is eighteen?

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Adult Guardianship & the alternatives...

Health Care Surrogate Act	Power(s) of Attorney
Limited Guardianship	Plenary (Full) Guardianship

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POWER of ATTORNEY

- Transfer of money/property to a 3rd Party OBRA Pay Back Trust...
- Represent:
  - IRS
  - Illinois - School – NEW – 8/07 LAW!
  - Residential
  - Employment or "DT"
  - HFS (DPA), DHS, DPH, SSA - Their OWN...

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GUARDIANSHIP

- Independence vs. Protection?
- Court... Law Suit...
- M.D. Report

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GUARDIANSHIP

- Guardian ad Litem
- Parents as "Co"
- Siblings, others, as "Co"
- Agencies' positions
- Voting... Driving...

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GUARDIANSHIP

- When can do it ?
- Person vs. Estate ?
- Annual Reports ?
- Residential Placement ?

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GUARDIANSHIP

- Move out of State
- Move out of County?
- Why some parents/siblings decline?
- Then what?

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WRITE IT DOWN!

Providing information, guidance & instructions…

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WRITE IT DOWN!

Family Information & History, including Medical History of Immediate Family...

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WRITE IT DOWN!

Diagnoses, Medical History, Medications, Issues & Care of your Child...

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WRITE IT DOWN!

Daily Care, Needs, Activities, Behavior, Habits & Routines...

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WRITE IT DOWN!

Night Time Habits & Routines...

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WRITE IT DOWN!

Diet\Food... Likes and Dislikes...

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"Friends", Social Activities, & Skills...

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WRITE IT DOWN!

Behavior... "Dos" & the "Do Nots", Techniques, "Buzz" Words or Phrases...

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Employment ?
Day Program ?
Residential ?

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Government Benefit Information

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Emergency Information, including Short Term & St... By Guardian Declarations...

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WRITE IT DOWN!

Wills, Trusts, Life Insurance, P...ers of Attorney, SNT, funeral desires & related info...

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WR... DOWN!

How the ... with Ma...

One to two pages... BULLET POINTS!

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IF WE HAVE TIME...

ABLE ACCOUNTS

(BEWARE: Not as advertised…)

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ABLE – SAVINGS ACCOUNT- PENDING IN D.C.

COMPARE 3RD PARTY MONEY

	3RD PARTY $ SNT	3RD PARTY $ ABLE
REQUIRED REPORTS	NO	YES
PAY BACK REQUIRED	NO	YES
USES	UN-LIMITED	LIMITED
MAXIMUM	NONE	$500,000
INCOME TAXED	WHEN EARNED	NO IF!

ABLE – SAVINGS ACCOUNT- PENDING IN D.C.

COMPARE 1ST PARTY MONEY

	1ST PARTY $ SNT	1ST PARTY $ ABLE
REQUIRED REPORTS	YES STATE & COURT	YES PLUS ADD U.S.
PAY BACK REQUIRED	YES NO DIFFERENCE	YES NO DIFFERENCE
USES	UN-LIMITED	LIMITED
MAXIMUM	NONE	$500,000
INCOME TAXED	WHEN EARNED	NO IF!

Special Needs Future Planning

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email@specialneedsfutureplanning.com or email@brianrubin.com

Attorneys: Brian Rubin ● Judith Smith
Staff: Nicole Rosenthal ● Karen Muschkat ● Linda Rubin

Who is Brian Rubin?

On January 27, 1981, Brian Rubin's second of his three children, Mitchell, was born. Mitch has Autism among other diagnosed special needs. The birth of Mitch changed the lives of Brian and his wife, Linda. For Brian, an attorney, his professional life, as well as his personal life took a "fork in the road".

Prior to Mitch's birth, since 1976, Brian had been in private practice, concentrating in Estate Planning, Estate Administration, Tax Planning and Business Law. Since 1981, with Mitchell's birth, Brian's practice became dedicated to appropriate "Special Needs Future Planning" for his fellow Illinois parents and families of children and adults with special needs. This includes Estate Planning, Estate & Trust Administration, Special Needs Trusts, OBRA 93 Trusts, Adult Guardianship Proceedings, Government Benefits' Planning, and Guidance on navigating the "maze" of Illinois Residential and Day Services, Transition Planning and related matters.

Brian is, by invitation, a member of the Special Needs Alliance (SNA), the national organization of attorneys who concentrate their practices in Special Needs Planning. Brian also serves on SNA's Board of Directors.

Brian's personal time, likewise, has been dedicated to assisting and serving his fellow families of children and adults with special needs. Brian serves on The Arc of Illinois Board of Directors. Brian served as Chairman of the State of Illinois Department of Human Services Statewide Advisory Council on Developmental Disabilities, by appointment of the Director of the Illinois Department of Human Services' Division of Developmental Disabilities. Brian serves by appointment of the Governor as a Member of the State of Illinois Department of Human Services' Quality Care Board. The Quality Care Board monitors and oversees operations, policies, and procedures of the Department of Human Services' Office of the Inspector General to assure prompt and thorough investigations of neglect and abuse allegations. Brian serves as Co-Chairman of the State of Illinois Department of Human Services Task Force on Autism, by appointment of the Secretary of the Illinois Department of Human Services. Brian serves on the State of Illinois Department of Human Services, Division of Developmental Disabilities, Strategic Planning Team, by appointment of the Director of the Illinois Department of Human Services' Division of Developmental Disabilities. Brian serves on the State of Illinois Disabilities Services Advisory Committee (The State of Illinois "Olmstead" Committee), by appointment of the Governor of Illinois (First by Governor Ryan, and then by Governor Blagoyevich. Brian also serves and has served on other State of Illinois Department of Human Services Task Forces and Committees.

Continued on the other side…

Brian serves on the Board of Directors of Clearbrook, an agency that serves more than 3,000 children and adults with disabilities (He has served on the Clearbrook Board since 1989). Brian serves on the Board of Directors of PACT, Inc., the PAS/ISC Agent for DuPage County, which also is available to serve as Trustee of Special Needs Trusts and as Guardians of adults with special needs. Brian serves on the Board of Directors of SLSF (Special Leisure Services Foundation) which supports NWSRA (Northwest Special Recreation Association).

Over the years, Brian has served on various Parent and Parent/Teacher Boards, Councils and Committees in and for the Special Education District of Lake County, Illinois (SEDOL), and served as the Founding Chairman of the Board of Trustees of the SEDOL Foundation. Brian has served as an officer and as a member of the Board of Directors of KESHET (Jewish Parents of Children with Special Needs). Brian has served on the Board of Directors of the Northpointe Resources, formerly known as Northpointe Achievement Center. Brian has also served as General Counsel for Countryside Association.

Brian is a former IRS Agent and a former IRS Estate Tax Attorney. Brian served by appointment on the American Bar Association's Sub-Committee on "Special Needs Trusts and Other Protective Vehicles". Brian has served by appointment of the President of the Illinois State Bar Association on that Association's "Section Council" on Estate Planning, Probate and Trust Law. Brian also has served, by appointment, on that Association's "Standing Committee" on the Mentally Disabled. Brian has been admitted as a member of the National Academy of Elder Law Attorneys (NAELA).

Brian serves on the Faculty of the Illinois Institute for Continuing Legal Education (IICLE), specifically on the topic of Special Needs Planning and Special Needs Trusts, and also is a contributing author for the IICLE Special Needs Trusts Practice Handbook. Brian has served on the Board of Governors of the Northwest Suburban Bar Association, as Chairman of that Association's Estate Planning Probate and Tax Law Committee, as Co-Chairman of that Association's Continuing Legal Education Committee, and was the co-founder and served as Chairman of that Association's Committee on Planning for Individuals with Disabilities and the Elderly.

Brian served three, four year terms, twelve years, as an elected Village Trustee in Buffalo Grove, Illinois (population 45,000) and was the founder of and served as Chairman of the Buffalo Grove Commission for Residents with Disabilities. Brian is a Past President of the Rotary Club of Buffalo Grove and has also served as an Assistant District Governor for Rotary District 6440.

Brian sincerely feels that Mitchell has allowed him, his wife, Linda, as well as "big sister/assistant mom" Nicole and "little/big brother" Benjamin, to better appreciate what is truly important in life... and what is... maybe... not so important. Brian feels the tremendous responsibility of not only being the parent of a child with special needs, but also as an attorney with the knowledge and ability to assist others in special needs future planning... needed to secure the future of children and adults with special needs.

While Brian wishes for Mitchell to have a long, healthy and enjoyable life, he often admits that he prays that he lives at least one moment longer than Mitchell... for that ultimately is the "Parent's Prayer"... the desire not to ask anyone else to, or to have anyone else have to "take over" the enormous responsibility of caring for their child with special needs... as only a fellow parent could understand.

Brian is a frequent author and lecturer on the topic of appropriate future planning for families of individuals with special needs, and has been interviewed on television and radio, and has been often quoted in newspapers (including the Wall Street Journal, Chicago Tribune and Chicago Sun Times), books and magazines.

Information for New Families

FRIDAY
Breakout Session #3: 2:10 – 3:10 PM
Trillium Ballroom IV, Conference Center

CHARGE 101:
SETTING THE SCENE – COMMUNICATION WITH BABIES AND TODDLERS

Robert Last

Early Childhood Educator
Vision Australia

Presenter Biography: *Rob Last* is an Early Childhood Educator with Vision Australia working with young children and their families. He has a particular interest in the development of communication systems for children with sensory disabilities in particular in hearing and vision. Rob has been an invited presenter at every International CHARGE Syndrome Conference and the Australian and New Zealand Conferences, which are held in the 'in-between' years. He has done a longitudinal study of the progress made by people who have CHARGE syndrome, beginning by recording their progress in 1993 with follow up interviews in 2000 and 2007.

ADDRESS: PO Box 404, Somers Vic, 3927 Australia; TEL: (03)59832675; EMAIL: roblast@iinet.net.au

Presentation Abstract: "For children who have CHARGE syndrome there is strong evidence that diagnosis and intervention before six months of age significantly improves outcomes in communication, behaviour and learning" Jan van Dijk and Arno de Kort
This presentation explores what communication means, the early considerations in communication with babies and toddlers, the strategies in acquiring communication skills and exploring outcomes.

Setting The Scene: Babies and Toddlers

- Rob Last
- Vision Australia
- 38 Hartnett Drive
- Seaford Vic 3198
- Australia
- email: roblast@iinet.net.au

•"For children who have CHARGE syndrome, there is strong evidence that diagnosis and intervention before six months of age significantly improves outcomes in communication, behavior and learning."

•**Jan van Dijk, Arno de Kort**

The Early Months

- Major medical issues take precedence
- A large team of specialists become involved
- Life and death issues dominate
- Outcomes are unknown
- Major surgery's take place
- Bonding between child and parents is interrupted
- Medical procedures take place without warning or preparation

Early Considerations

•Educators and therapists may assist in:

- Positioning of the crib and the people who surround the baby
- Contrast
- Lighting
- Provision of communicative cues
- Alerting to events
- Early literacy

Early Communication

- Contentment
- Distress
- Wakeful
- Sleepy
- Pain
- Comfort
- Hunger
- The first smile

Speech/Communication

- Communication includes:
  - body language
  - facial expression
  - touch cues
  - gestures
  - situational clues
  - *AND*
- Speech

Why the emphasis on early communication needs?

- Most children with CHARGE are deaf or hard of hearing
- All senses may be affected
- In addition:
  - Breathing difficulties
  - Excessive secretions
  - Facial palsy
  - Cleft palate
  - Eating issues

Communication Embraces

- Sign language - the sign language of the country where the child lives
- Speech with hearing aids
- Speech with a cochlear implant
- Speech assisted by BAHA (Bone Anchored Hearing Aid)
- Visual programs
- Signed English
- Reading and writing

•"For children with CHARGE, the aim is a collaborative approach that brings together a Teacher of the Deaf (preferably fluent in Sign Language), a Pediatric Audiologist (familiar with CHARGE), Speech Pathologist (also experienced in eating issues) and an Occupational Therapist (trained in Sensory Integration)"

•- David Brown

What needs to happen

- Early cues & prompts offered
- Talking up close into each ear
- Gestures and sign language introduced
- Hearing aids introduced
- Cochlear implant discussed and explored
- More advanced sign language continues
- Talking continues and amplification needs closely monitored
- Literacy rich environment provided

Strategies for learning a new language:

- from Teachers of the Deaf
- from signing dictionaries
- from DVD's
- from members of the deaf community
- from CODA's (Children of Deaf Adults)

Strategies for learning a new language:

- from attending classes/courses
- from 'voice off' times
- from child care people who are fluent in sign
- from family week-ends with members from the deaf community
- from exploring multiple opportunities to be immersed in sign

Communication and Behavior

- Behavior and Communication are inseparable
- Poor communication / high risk for behavior issues
- Acquiring symbolic language reduces the risk

Communication and Gross Motor Development

- **"**Through parent surveys it was found that children with CHARGE can achieve gross motor development milestones but do so at a slower rate than normal children. A correlation between gross motor development and communication abilities was also found."

- - James Thelin and Lori Travis

What then is the aim?

- To provide a range of communications
- To ensure the communication is meaningful
- To provide an environment of immersion in a meaningful language
- To aim for communication competence which provides the opportunity and ability to express thoughts, ideas, emotions and humor i.e. symbolic language

"All who are in contact with persons with CHARGE need to **adapt to** each child's communicative development. They need to help establish the most favorable social contexts for making the most of their communication competencies to enrich their social lives."

- Jacques Souriau, et al

•What is considered best practice:

- for parents, extended family and professionals to *embrace bilingualism*
- for the *best amplification* to be provided
- to provide an *environment* of visual language and amplified speech

Outcomes

•**"**We have become a bilingual family. Sean is able to access both oral and signed communication. Both modes of communication are essential to his overall acquisition and use of language."

•- Madelene Rich, Mother of Sean who has CHARGE

‘Children with CHARGE Seven Years Later’

The DVD

- First filmed in 1993
- Follow up in 2000
- Further follow up in 2007
- Available from Rob Last
- email: roblast@iinet.net.au

General Interest Demonstration

FRIDAY
Breakout Session #4: 3:30 – 5:30 PM
Interact at INFORMATION CENTRAL
Trillium Balrooms, Conference Center

FUN-Chi

DE-STRESS FOR KIDS

Sharon Barrey Grassick

Deafblind Education Team Leader
WA Institute for Deaf Education - WAIDE
Western Australia
Adjunct Lecturer – Renwick Centre, RIDBC NSW

Presenter Information: *Sharon Barrey Grassick* is affiliated with the following organizations: WA Institute for Deaf Education, Department of Education and Training, Western Australia; WA Director - CHARGE Syndrome Association of Australasia; Secretary - Australian DeafBlind Council (ADBC); Adjunct Lecturer, Sensory and Multiple Disabilities, Renwick Centre RIDBC and Tai Chi Assistant Instructor – Tai Chi @ The Beach, WA

ADDRESS: 10 Belford Road, CITY BEACH, Western Australia 6015
TEL: +618 9385 8843 mobile: 0410 543 014; EMAIL: sbgrassick@gmail.com

Presentation Abstract: Stress! We all have it at times; individuals with CHARGE often have too much of it. So, how to de-stress? Challenging behaviours, to varying degrees, can emerge in children with CHARGE; stress is a probable key cause. There are no magical answers to reduce stress and self-regulate behaviours. Let's think outside the square - to the circle…Yin Yang or Tai Chi. This presentation will demonstrate a fun approach to exercise and stress management - called *FUN Chi*!

Fun Chi

De-Stress for Kids

Sharon Barrey Grassick
Deafblind Education Team Leader
WA Institute for Deaf Education - WAIDE
Western Australia
Adjunct Lecturer – Renwick Centre, RIDBC NSW

Fun Chi

- Thinking outside the square - to the circle!
  - Tai Chi / Yin and Yang



- A ***Fun*** approach to exercise
- A ***Fun*** way to de-stress

From stressed…

To de-stressed!

What is ***Fun Chi***?

- ***Fun Chi*** is modified Chi Kung/Tai Chi
  - adapted for children
  - particularly children with balance issues
  - E.g. CHARGE Syndrome

What is Chi Kung?

- Chi Kung, or Qigong, is an ancient form of Chinese healthcare
  - 5,000 years old
- Exercise of our internal energy
  - Enhancing fitness of mind and body
  - Developing vitality
  - Helping in cure of stress and disease.

What is Tai Chi?

- Internal martial arts
- 'Chi' is life energy
- Has been described as 'yoga in motion'
  - Moving meditation
- Least amount of injuries of any sport
- Evolved from Chi Kung 800 years ago

Documented Health Benefits

- Improves general health and well being
- Increases energy, agility and flexibility
- Loosens and strengthens joints and muscles
- Rejuvenates body, mind and spirit
- Improves concentration and memory
- Helps in control of emotions
- **Reduces stress**

And there's more…

- **Improves balance!**
- And we all know that balance can be a big issue for most children and adults with CHARGE
- Tai Chi yin yang symbolizes balance

Balance

- Visualize your feet have grown roots into the ground

Balance

- More than just vestibular!
- Need for balance of academics, therapies and life skills
- Holistic approach
- Yin and yang

Stress Management

- Reaching and maintaining a calm but alert state

Just imagine…

- Improved balance!
- And reduced stress!
- Is it possible?
- Of course it is!

Why ***Fun Chi***?

- Holistic approach
- Focus on the 'forgotten senses'
  - Proprioceptive
  - Vestibular
- Documented benefits of Tai Chi reflect areas of specific need for children who are deafblind, including children with CHARGE

What are these areas?

- Reduction of **stress**
- Relaxation
- **Balance**
- Focus/concentration
- Energy
- Sleep
- Immune system

So where do we start?

- With a positive attitude!
  - School
  - Teachers
  - Students
  - Parents

Who?

- Primary Grade 2 Class
  - including child with CHARGE
- Total of 28 children
  - aged 7 and 8 years old

When?

- Twice weekly, following lunch/recess
- 20 - 25 minutes per session
- Commenced March 2009 – to present

Where?

- Undercroft assembly area of school

How?

- Moves chosen and adapted for repetition, weight shift, balance benefits and fun factor:
  - ***Fun Chi*** Bow
  - Warm up exercises
  - Shibashi
  - Lotus (Cool down)
  - Close off
  - ***Fun Chi*** Bow

Fun Chi access for child with CHARGE

Access - Hearing

- FM hearing aid and microphone used

Access - Vision

- Child chose front row position
- Educational Interpreter interpreted/demonstrated moves
- ***Fun Chi*** assistants positioned in front and to side of child's better eye

Access – Communication

- Educational Interpreter using Auslan (Australian Sign Language)

Fun Chi Signs

- New sign or phrase related to ***Fun Chi*** taught to class at end of each session
  - Signs taught chosen by child with CHARGE
  - All signs had positive connotations

Access - environment

- A solid support behind child (stage edge or large wooden box) seemed to aid balance

Warm-ups

- ***Fun Chi*** bow to start
- Repetitive exercises designed to warm-up all joints
  - Starting with head
  - Moving down through hips and legs
  - Ending with feet

The White Crane – Warm up

Shibashi

- Means 18 movements
- Each movement repeated 3 or more times
- Repetition improves memory, skills and confidence
- Diaphragmatic breathing demonstrated and encouraged

The Lotus

- Cool down exercises
- Tells a story using nature and animals
- From the sea to the earth to the sky and back
- Children love this!

Chi = Energy

The Energy Ball

Now put the energy ball on your head!

Move energy down through body…

Practical Benefits

- Can be done anywhere, anytime
- People of all ages and needs
  - Children
  - Arthritis
  - Cancer
- No special equipment
- No special clothing
  - Loose and comfortable best
- Easy to learn

Fun Chi FUNdamentals

- Children invited to participate at all levels
  - Renaming moves
  - Suggestions presented
  - Class votes
    - E.g. Swinging arms now 'Octopus Dance'

Fun Chi Assistants

- Each session 2 or 3 children chosen as assistants
- Children stood at front of class as leaders
- Chosen for any of following:
  - Concentration
  - Helpfulness
  - Attitude
  - Birthday
  - Etc.

Kids love ***Fun Chi***!

- Each child made own ***Fun Chi*** badge
- ***Fun Chi*** presented at whole school assembly
  - All wore ***Fun Chi*** badges
  - Allowed to take badges home
  - Video and photos of assembly
- Video shown to class
  - (**see video here at conference**!)
- Fabulous feedback

Extension into curriculum

- ***Fun Chi*** used as catalyst for student interviews (public speaking)
- Students encouraged to use imagination to rename moves and write them down
  - Using correct grammar, punctuation
- Encouraged to draw ***Fun Chi*** moves
- Building leadership (***Fun Chi*** assistants)
- Students currently rewriting *Lotus* into story that relates to their school.

How has ***Fun Chi*** been evaluated?

- Questionnaires – pre and post ***Fun Chi***
  - Parents
  - Teacher
  - Students

- Video documentation

- Student interviews

Has ***Fun Chi*** been successful?

- Questionnaires indicate YES!
- Video documentation shows YES!
- Student interviews say YES!

Research Needed

- Easy to establish that everyone had **fun**!
- Difficult to establish a measurable reduction in stress;
- Research needed.

Where to from here?

- Class now progressed to Tai Chi Level 1
- Tai Chi is more complex than Shibashi, requiring more weight shift, balance and concentration than basic Chi Kung moves
- Progress being documented

Above all…

- Put ***FUN*** into ***FUN***ctional!
- Provide unique and interesting opportunities for skills development
- Offer incentives to improve skills
- Invite participation in delivery
- Reward efforts
- Think outside the square…
- Have ***FUN***!

Websites and References

- www.worldtaichiday.org
- *www.**taichi**at**thebeach**.com.au*

Fun Chi!

SENSORY AND DEVELOPMENTAL INFORMATION

SATURDAY
Breakout Session #5: 10:45 – 11:45 am
Trillium Ballroom II, Conference Center

SMELL AND PUBERTY IN CHARGE SYNDROME

Jorieke Bergman, M.D.
&
Conny van Ravenswaaij-Arts, M.D., Ph.D.

University Medical Center
Groningen, The Netherlands

Primary Presenter Information: *Dr. Bergman* is a clinical geneticist in training and PhD-student
She studied medicine at the Utrecht University in the Netherlands and started specializing in clinical genetics in Groningen in 2006. In 2007 she received a grant that allowed her to start a PhD project on CHARGE syndrome that is currently still ongoing. As part of this PhD project she studied smell and pubertal development and causes of post neonatal death in patients with CHARGE syndrome In addition, mouse studies were performed in the Wellcome Trust Sanger Institute in Cambridge, Great Britain in order to gain insight into the underlying mechanisms of smell deficit and pubertal delay in CHARGE syndrome. She is also coordinator of the multidisciplinary CHARGE outpatient clinic in the Netherlands and is actively involved in the Dutch CHARGE parent support group.

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TEL: 0031(0)503617229, FAX: 0031(0)503617231, EMAIL: j.e.h.bergman@medgen.umcg.nl

Presentation Abstract: Smell deficiency and delayed/absent puberty often occur in CHARGE syndrome, but few studies have looked at these features in adolescent patients. Therefore, we studied smell and puberty development in 22 adolescent CHARGE patients and showed that puberty and smell problems always co-occur. Therefore, a smell test can possibly predict whether puberty will occur spontaneously or not. This will prevent delay of hormonal pubertal induction, resulting in an age-appropriate puberty in smell deficient CHARGE patients.

University Medical Center Groningen

Smell & puberty in CHARGE syndrome

Jorieke Bergman
MD-medical research trainee
Department of Genetics
UMC Groningen, the Netherlands
CHARGE conference 2009 (Chicago)

Questions / objectives

- How often do smell and puberty problems occur in patients with CHARGE syndrome?
- Can a smell test predict whether spontaneous puberty will occur?
- Why is it important to know from a young age that puberty will not occur spontaneously?

Outline of the presentation

Introduction
- Normal smell and puberty
- Link smell and puberty

Objective of the study

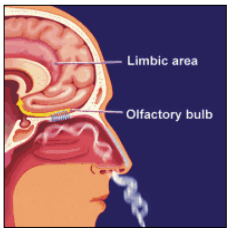
Methods
- Smell test
- Evaluation of puberty

Results of our study

Conclusions and recommendations

Normal smell

Odor
↓
Olfactory nerve
↓
Brain (olfactory cortex)
↓
Perception of smell

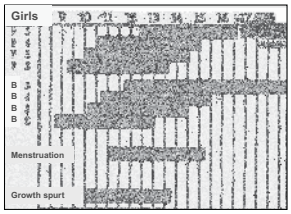


CHARGE: olfactory bulb small or absent
→ inability to smell (anosmia)

Normal puberty - girls

Onset of puberty at approximately 10.5 years
- Breast development (9 – 13 years) (B)
- Pubic hair (9.5 – 13.5 years) (P)
- Growth spurt (10 – 13.5 years)
- Menstruation (11 – 15 years)

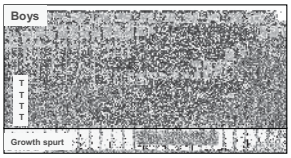
WIDE VARIATION!



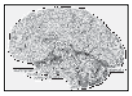
Normal puberty - boys

Onset of puberty at approximately 11.5 years
- Testes enlargement (9.5 – 14.5 years) (T)
- Pubic hair (10.5 – 14.5 years) (P)
- Growth spurt (10 – 14 years)

WIDE VARIATION!



Normal puberty



Hypothalamus: GnRH

CHARGE: no GnRH

→ delayed puberty (hypogonadotropic hypogonadism)

Pituitary gland: LH and FSH

Ovaries: estrogens

Testes: testosterone

Start of puberty

Link smell & puberty

Olfactory bulb (sense of smell) develops together with hypothalamus (regulator of puberty) during embryogenesis

Smell and puberty problems are often seen together in one patient

Smell & puberty

Kallmann syndrome

Anosmia
Delayed puberty
Renal abnormalities
Cleft lip/palate
Mirror movements
Tooth abnormalities

Anosmia: inability to smell

CHARGE syndrome

Anosmia
Delayed puberty
Renal abnormalities
Cleft lip/palate
Coloboma
Heart defects
Deafness
Choanal atresia
Retardation of growth and development
Balance disturbance

Methods

Patients

• 26 *CHD7*-positive CHARGE patients aged 10 years or older that were seen at the CHARGE outpatient clinic in the UMCG

Assessment of pubertal development

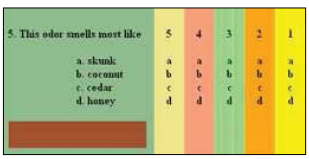
• Evaluation by a pediatric endocrinologist (Tanner stadia, anthropometry, biochemical evaluation)

Smell test

• UPSIT

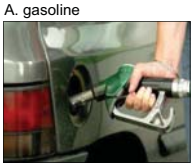
Smell test

UPSIT (**U**niversity of **P**ennsylvania **S**mell **I**dentification **T**est)
4 booklets with 10 “scratch&sniff” odorants each
(for children of 5 years and older)



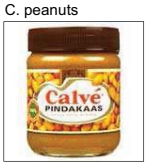
UPSIT picture book

A. gasoline



B. pizza

C. peanuts



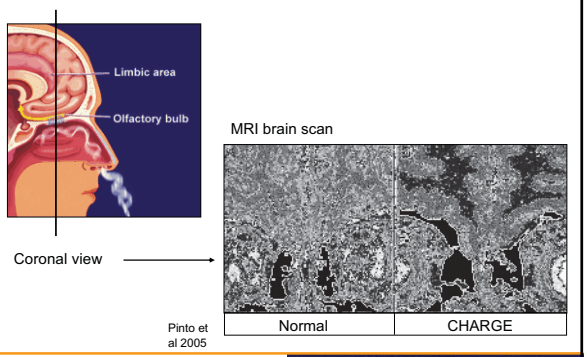
D. lilac

UPSIT problems

Problems (CHARGE related)
- Communication difficulties
- Mental retardation
- Tube feeding
- Bilateral atresia of choanae

→ Evaluation olfactory bulbs on MRI brain scans

Background: smell



Pinto et al 2005

Evaluation of puberty

Physical examination (pubertal stages) by pediatric endocrinologist

Measurement of hormones in blood

What is TOO late?
- For boys > 14 years (95% testes enlargement)
- For girls > 13 years (95% breast development)

Results

Smell
- 13/18 patients could not smell (72%)
- Parents often thought their child could smell, when the child could not

Puberty
- 13/19 patients had delayed/absent puberty (68%)
- 8 patients have started hormone treatment

Conclusion

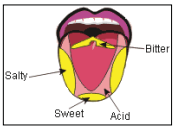
Smell and puberty problems were 100% correlated in our study

A smell test can possibly predict whether spontaneous puberty will occur or not

Significance - smell

The importance of smell

- Alarm function (spoiled food, leaking gas, fires)
- Amplification of our sense of taste
- Important in social interactions (body odors)
- Smell is intimately linked to memory

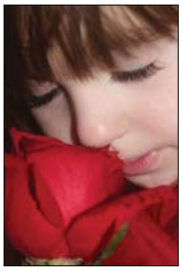


Treatment of anosmia

Anosmia cannot be cured

Preventive measures:

- Smoke alarms
- Natural gas detectors or change from natural gas to electric
- Mark expiration dates on food
- Deodorant / help of friends

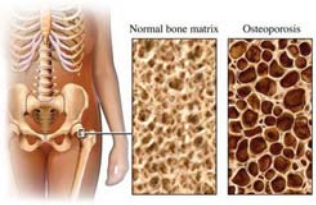


www.anosmiafoundation.org

Significance - puberty

The importance of puberty onset at a normal age

- Less socio-emotional problems (teasing by peers)
- Reduced risk of osteoporosis



Treatment of delayed puberty

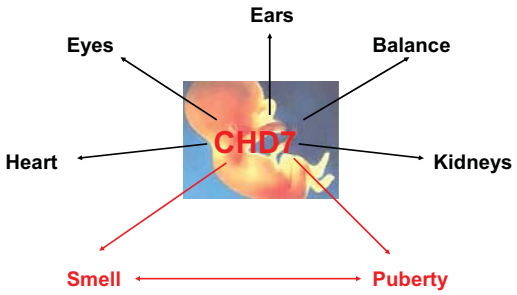
Treatment of delayed puberty:

- Hormones (testosterone, estrogen/progesterone)
- To restore fertility other hormones are necessary (gonadotropins, GnRH)

Disadvantages of hormone treatment:

- Possible increase in behavioral problems (especially in boys: testosterone – aggression, increase in muscle strength)
- Menstruation can be a burden

CHARGE: smell & puberty



University Medical Center Groningen

UMC Groningen
Conny van Ravenswaaij-Arts, clinical geneticist
Gianni Bocca, pediatric endocrinologist
Linda Meiners, neuroradiologist

ZonMw GUIDE

UMC St Radboud Nijmegen
Marjolijn Jongmans, clinical geneticist
Lies Hoefsloot, molecular geneticist
Hedi Claahsen-vd Grinten, pediatric endocrinologist

Viataal Sint Michielsgestel
Eline Tiems & Anneke Schoenmaker, physicians for people with intellectual disabilities

THANKS TO ALL THE PATIENTS WHO COOPERATED IN THIS STUDY

Questions?

FAMILY SUPPORT

SATURDAY
Breakout Session #5: 10:45 – 11:45 am
Trillium Ballroom III, Conference Center

PARENTING CHILDREN WITH CHARGE SYNDROME: PERILS AND PROMISE

Timothy S. Hartshorne, Ph.D.

Department of Psychology
Central Michigan University
Mt. Pleasant, MI

Presenter Information: *Tim Hartshorne* is a professor of psychology, specialized in school psychology, at Central Michigan University. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. His particular interest is in understanding the challenging behavior exhibited by many individuals with CHARGE. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project. His current project is editing a book, along with Sandy Davenport, Meg Hefner, and Jim Thelin, on CHARGE which should be published in 2010.

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TEL: (989)774-6479 office; FAX: (989)774-2553; EMAIL: tim.hartshorne@cmich.edu ;
WEB: www.chsbs.cmich.edu/timothy_hartshorne

Presentation Abstract: Parenting children with CHARGE is a huge challenge, and parenting methods for "regular" kids don't seem to apply. This presentation suggests overall strategies for parenting children with CHARGE. The presentation topics include typical characteristics, parent experiences, courage, executive functions, family meetings, encouragement, behavior & misbehavior, self-regulation, and punishment & rewards.

9th International CHARGE Syndrome Conference, Indian Lakes Resort, Bloomingdale, IL, July 24-26, 2009

Parenting Children with CHARGE Syndrome: Perils and Promise

Timothy S. Hartshorne
Central Michigan University

The World of Disabilities

- Difference – leading to a felt minus
- Work
  - Limitations due to time and resources
  - An escape
- Friendship
  - Lack of social support
  - Change in participation
- Love
  - Expectations
  - Commitment

Typical Characteristics of Child

- Communication difficulties
- Sensory and motor impairments
- Intellectual disability
- Fragile health,

pain, & limitations

Parent experiences

- Parental Guilt: The Courage to be Imperfect

"How do you know if you have the right doctors and therapists doing the right things and enough of them? How do you know if you're doing all the right things and enough of them? I have been avoiding phone calls and insurance stuff, etc, because I just want us to be normal."

Parent experiences

- Need to defend and protect the child

"Our first battle was over getting deafblind consultative services. We fought and fought. Now we are going through it again. Maybe it's just the looming battle, but I feel like weeping. Yes, it can be necessary to fight like the devil, but that doesn't make it right."

Parent experiences

- Capability of the child

"When she was an infant the hospital sent out evaluators. They sat me down and told me she would never walk, talk, or be independent. After he left I cried and had such terror my world spun. Then I thought "Baloney." She ended up learning lots. Her performance always goes above and beyond potential. Always."

Parent Experiences

- Can I cope?

"Tonight I am so very tired of being the person figuring out what's going on. It's been awhile since I've felt so isolated, scared, on and on. Right now it's as though nothing is enough to really help. I'm damned tired of this. I would like some help, too. I need it now. My daughter needs it now."

The problem is how to love your child with no need for your child to be any different, AND without giving up hope that your child may progress with time and resources.

COURAGE IS NOT DENIAL

BUT SOMETIMES COURAGE LOOKS LIKE DENIAL TO OTHER PEOPLE

Hartshorne, T. S. (2002). Mistaking courage for denial: Family resilience after the birth of a child with severe disabilities. *Journal of Individual Psychology, 58*, 263-278.

Genetic Component of Behavior

- Behavioral Phenotype
- Executive Dysfunction
- Self-Regulation

"A pattern of behavior that is reliably identified in groups of children with known genetic disorders and is not learned."
(Harris, 1995)

If I behave like this, I probably have CHARGE syndrome

Executive Functions

- Inhibit
- Shift
- Emotional Control
- Initiate
- Working Memory
- Plan/Organize
- Organization of Materials
- Self Monitor

Self-Regulation

- Poor self-calming with irritability
- Sleep problems
- Feeding problems
- Inattention
- Mood regulation problems
- Sensory processing problems

The problem is how to parent when…

- You exist in the world of disability
- You struggle with problems of acceptance and hope
- Your child exhibits behavior which is related to a syndrome
- You are exhausted by it all

It is hard enough to cope with raising a child who does not have obvious disabilities. Why in the world would you think you should know how to raise a child with tremendous needs?

Parenting Skill #1

- Have the courage to be imperfect
  - Because you are!
  - Because everyone else is too!
  - Even the presenter! [just ask his other kids]

Parenting Skill #2

- Understanding behavior
  - All behavior has a purpose
  - All behavior is communication
  - Behavior reflects solutions that are effective for the child even though they may be a problem for the parents
  - So don't personalize it

Goals of Misbehavior

Attention
Power
Revenge
Inadequacy

Parenting Skill #3

- En**courage**ment
  - We learn by making mistakes
  - We learn by the consequences of our actions
  - Avoid dis**courage**ment

Parenting Skill #4

- Avoid over reliance on rewards and punishments
  - Invite cooperation
  - Teach other methods to solve problems
  - Use natural and logical consequences
  - Be authoritative, not dictatorial or permissive
  - Be sympathetic but set limits

Parenting Skill #5

- Build Communication
  - Read behavior as communication
  - Take turns when interacting
  - Let the child know that you have heard and understood, even if your answer is “no”

Parenting Skill #6

- Family Meetings
  - Raising a child is a family challenge
  - Cooperation rather than competition
  - Getting on the same page
  - Sharing and working together

"I have a question. I struggle with this. The OCD obsessions are our biggest battles. One is water. She has to get her clothing wet before she can take them off. And she needs to change her clothes often. I have tried making her stop both but she just goes over the edge. I have finally come to the approach that I can't stop her, but I need to just try to control the amount of water she gets when she soaks her clothes. If we take the water away, she will pee on them so we have our choice of water supply there. It's going to happen one way or another, from the well or from her. Is it wrong to help her with the water? I am saying it is ok and feeding the obsession. But I have little hope of getting her to stop, and so I don't know what to do and what harm I am doing by allowing it. Jessica's obsessions seem to have come from routine. Then they turned into habits, rituals and then true obsessions. If I had noticed that earlier in life, do you think that I could have prevented some of this?"

Where to Begin?

- Anxiety is a basis for OCD, except perhaps in autism
- Genetics – does not mean it happens without a reason
- What does it solve?
- What is it communicating?

All Behavior is Communication

- Look what I know how to do
- I really enjoy the feel of wet clothes
- If I change my clothes I can avoid something
- I am in pain, and this really helps
- I am anxious, and this really helps
- I like having you engage with me

Parent Issues

- Is it OK to participate in the ritual?
- How do you prevent a power struggle?
- Guilt about not having prevented this.
- Remember, you have an exhausted mom.
  - Guilt
  - Need to defend and protect
  - My child is capable of not doing this
  - Can I cope?

Parenting Objectives

- Communication skills development
- Choice making
- Self-regulation
- Independence

In Crisis

- Read behavior as communication
- Consider pain and health
- Look for sensory overload
- Look for any changes in life space
- Respect choices
- Provide reasonable limits

In General

- Medication should not be the first choice
- Work on optimal eating, sleeping, health
- Provide choices & RESPECT COMMUNICATION
- Provide powerful encouragement
- Reduce stress
  - Routine/consistency
  - Provide choices
  - Sensory breaks

Contact information

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INFORMATION ON COMMUNICATION

SATURDAY
Breakout Session #5: 10:45 – 11:45 am
Trillium Ballroom IV, Conference Center

DEVELOPING MEANINGFUL CONVERSATIONS

Diane Haynes, M.Ed.

State Coordinator, Kentucky Deaf-Blind Project
University of Kentucky

Presenter Information: *Diane Haynes* is has been in the field of education for more than 30 years. She belongs to many professional organizations, has made numerous professional presentations, and has published educational materials. At present, she is the Coordinator of the Deaf-Blind Project in Kentucky. At this Conference, she will become a member of the Board of Directors of the CHARGE Syndrome Foundation, Inc.

ADDRESS: KENTUCKY Deaf-Blind Project, 229 Taylor Education Bldg. Lexington, KY 40506-0001; TEL: 502-777-6235; EMAIL: Diane.Haynes@ksb.kyschools.us

Presentation Abstract: The basic premise of this presentation is that any interaction between human beings is the basis for a conversation. Successful interactions / conversations depend on our ability as communication partners to perform the steps necessary to complete a basic dyadic interaction. Steps encompass accurate identification of receptive functions and receptive forms that reflect knowledge of a partner's functioning within all seven sensory systems. The presenter will share strategies for developing an intervention plan.

"Developing Meaningful Conversations"

Diane Haynes, M.Ed.
State Coordinator
University of Kentucky
Kentucky Deaf-Blind Project

CHARGE Syndrome National Conference
July 2009

Every interaction with another human being is a conversation

One partner sends the information

The other partner receives the information

BASIC DYADIC INTERACTION

- Attract a person's attention to begin the communication exchange
- Direct a person's attention to the topic of communication
- Provide the partner with opportunity to take their turn
- Fill one's own turn in the exchange

RECEPTIVE Communication

INPUT **that is processed**

by the person *RECEIVING*

the information

in order to understand their environment

Receptive Communication is the Key …..

WHY ????????

Think of a computer

- Computer programs don't run well if the command information is not entered completely and accurately
- Computer input does not happen automatically – people enter the information very purposefully
- Like computer processing, if input is appropriate, adequate and consistent– the "output" or response will be accurate

Every conversation / interaction has a different purpose or FUNCTION

RECEPTIVE FUNCTIONS

FUNCTION	What Does the Receiver Need to Process?
1. Greeting	Someone is acknowledging me
2. Getting Someone's Attention	I Need to ATTEND to Someone
3. Tell person about UPCOMING EVENT	Something is going to happen
4. Tell person to BEGIN something	I need to START _____

RECEPTIVE FUNCTIONS

FUNCTION	What Does the Receiver Need to Process?
5. Tell person to CONTINUE something	I need to CONTINUE _____
6. Tell person something is OVER or FINISHED	This is finished / no more
7. Ask "yes/no" question	The answer to the question
8. Ask "wh" question	The answer to the question

RECEPTIVE FUNCTIONS

FUNCTION	What Does the Receiver Need to Process?
9. Ask person to MAKE a CHOICE	I need to choose something
10. Give a COMMAND or DIRECTIVE	I need to _____
11. Tell person you are LEAVING	_____ is going away

RECEPTIVE FORMS

- Speech
- Tactile/Touch Cues
- Object Cues
- Picture Cues
- Gesture Cues
- Tactile Signing
- Platform Signing
- Natural/Contextual Cues

RECEPTIVE FORMS

RECEPTIVE FORMS	Which Sensory System has to be ALERT, FIRING & READY TO GO to Receive & Process the Information Accurately?
SPEECH	AUDITORY
TACTILE / TOUCH	TACTILE / PROPRIOCEPTION
OBJECTS	VISUAL / TOUCH / AUDITORY / SMELL / TASTE / PROPRIOCEPTION VESTIBULAR
PICTURES	VISUAL

RECEPTIVE FORMS

RECEPTIVE FORMS	Which Sensory System has to be ALERT, FIRING & READY TO GO to Receive & Process the Information Accurately?
GESTURES	VISUAL
SIGN LANGUAGE (Platform)	VISUAL
SIGN LANGUAGE (Tactile or Co-Active)	TOUCH & PROPRIOCEPTION
NATURAL or CONTEXTUAL CUES	VISUAL / AUDITORY / SMELL / TOUCH / VESTIBULAR

QUESTIONS to ASK ????

1. What does the child/student understand?
2. Is my function/purpose of communication clear to the child/student?
3. Am I addressing all critical FUNCTIONS of receptive communication?
4. Do I need to modify the form(s) I am using to send information to the child/student?

Young Adults & Adults with CHARGE Syndrome ONLY

SATURDAY

Breakout Session #5: 10:45 – 11:45 am

Cyperus IV, Garden Level in the Conference Center

GOING TO COLLEGE: WHAT DO I NEED TO KNOW?

Beth Jordan, M.Ed.

Helen Keller National Center Regional Representative and National Consortium on Deaf-Blindness. Shawnee Mission, KS

and

Belinda Arnell

Adult with CHARGE syndrome

Australia

Presenter Information: *Beth Jordan* is the Great Plains Regional Representative for the Helen Keller National Center, serving deaf-blind consumers and their families. She also provides training and technical assistance to agencies/organizations who work with deaf-blind people. The Great Plains Region includes Iowa, Kansas, Missouri and Nebraska. Ms. Jordan has worked in this capacity since January, 1992.

Ms. Jordan received her Master of Education degree from the University of Arkansas in 1988, majoring in rehabilitation counseling and independent living with an emphasis in deafness. Prior to her work at HKNC, she worked four years as a VR counselor with a deaf caseload in Pasadena, Texas. Ms. Jordan is a certified rehabilitation counselor (CRC). She resides in Olathe, Kansas with her husband and two sons.

ADDRESS: Shawnee Mission, KS 66205; TEL: 913-677-4562; EMAIL: Beth.Jordan@hknc.org.

Belinda Arnell is a 26 year old young adult with CHARGE syndrome who resides in Australia. Her main obstacles are vision and hearing loss. Belinda has been at college for quite a few years and has completed an Advanced Diploma of Justice and most of a degree majoring in Psychology and Legal Studies. Currently, she works full time and studies part time and hopes to finish her degree in a couple of years.

Presentation Abstract: Applying to a college or training program after high school can be an overwhelming task. This session is designed to help a young adult consider what supports he or she might need during the college application process or when in a classroom on a campus. Participants will receive specially designed tool that provides a simple and easy way to determine the level and types of accommodations that a college offers a student with a vision and hearing impairment (e.g. interpreter services, notetakers, readers etc.).

N o handout available. Contact Beth Jordan for an electronic copy.

Medical Information

SATURDAY
Breakout Session #6: 1:00 – 2:00 PM
Trillium Ballroom II, Conference Center

CHARGE SYNDROME: THE "R" AND THE "G"

Jeremy Kirk, M.D.

Diana, Princess of Wales Children's Hospital
Birmingham, UK

Presenter Information: *Dr. Kirk* is a paediatrician and a long-time contributor to the knowledge base on CHARGE syndrome in the area of endocrinology and growth.

By his own description: he is London born and bred, now Head of pediatric endocrinology at the Diana Princess of Wales Children's Hospital in Birmingham, Britain's own "Second City". .Last remaining member of the UK CHARGE "Brit Pack" still living in the UK.! Associate Professor (Reader) in the University of Birmingham, Council Member of the Royal College of Paediatrics and Child Health, and past secretary of British Society for Paediatric Endocrinology and Diabetes. Has a special in interest in growth and gonadal problems. Medical Advisor to UK CHARGE Family Support Group.

ADDRESS: Diana, Princess of Wales Children's Hospital, Steelhouse Lane, Birmingham B4 6NH, United Kingdom; TEL: +44 121 333 8188; EMAIL: Jeremy.Kirk@bch.nhs.uk

Presentation Abstract: Both growth problems (failure to thrive, short stature, delayed/absent puberty) and also genital problems (micropenis/undescended testicles) are part of the original acronym of CHARGE. Whilst these problems are commonly seen in CHARGE, other more pressing medical and surgical problems often mean that they are not always recognized or treated appropriately. We have been collecting data on these problems from within our local clinic, and also national/international groups, in order to provide best practice.

CHARGE Syndrome: the “R” and the “G”

Dr. Jeremy Kirk
Consultant Paediatric Endocrinologist
Diana, Princess of Wales Children’s Hospital, Birmingham
United Kingdom

CHARGE: the “R” and the “G”

C

H

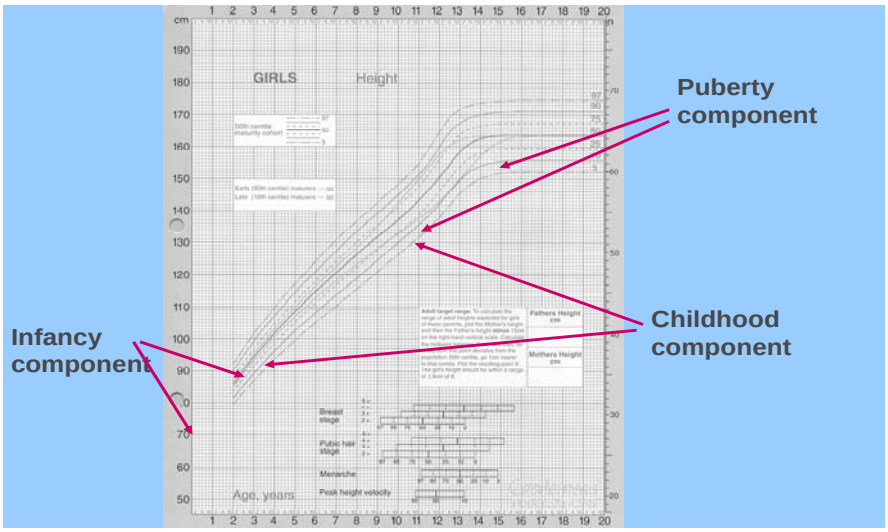
A

R etarded Growth

G enital Hypoplasia

E

Different growth components: all affected in CHARGE



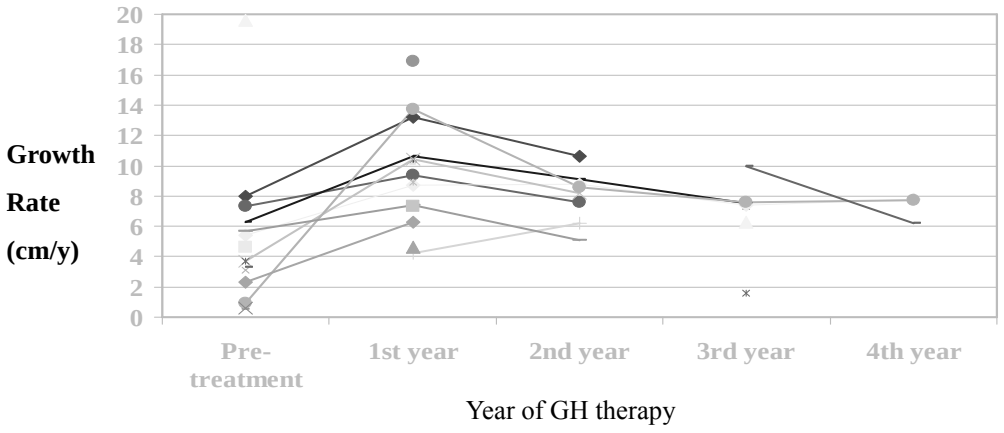
GH-deficiency and underactive pituitary (hypopituitarism)

- Incidence unknown in CHARGE syndrome.
- Probably more common than general population, especially in children with clefting.
- Data on growth hormone (GH) therapy in CHARGE obtained from 2 large international GH databases (NCGS (USA) & KIGS (Europe)).

GH therapy in CHARGE (USA)

- National Cooperative Growth Study (NCGS).
- 26 patients (19 female) aged 0.5-16.7 years. All but one patient were prepubertal.
- Most patients were short (23 had heights below 2nd centile)
- 22 had anterior pituitary function testing; 17 were GH-deficient.

GH therapy in CHARGE (USA)

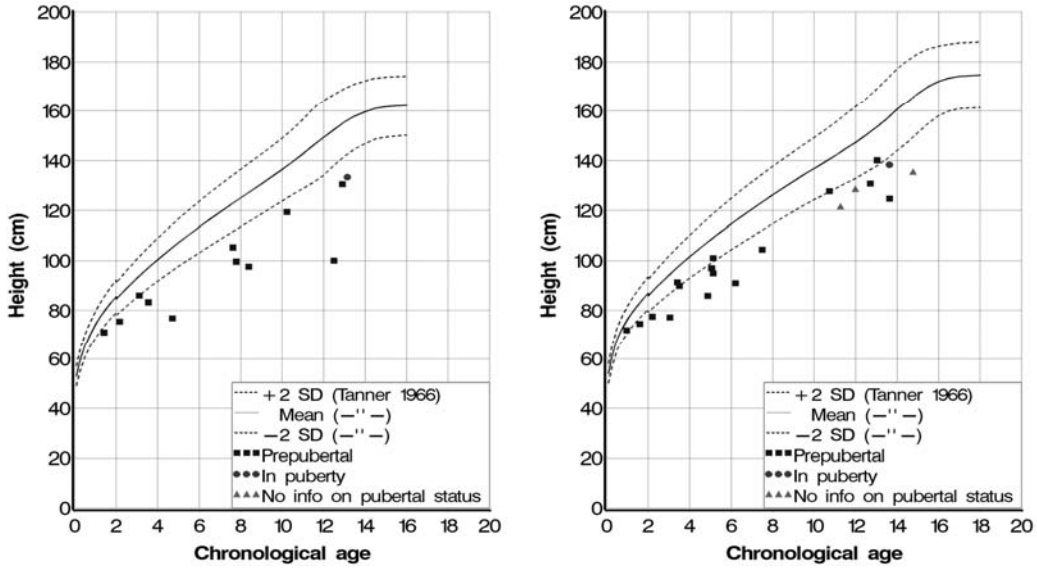


GH therapy in CHARGE (KIGS; European)

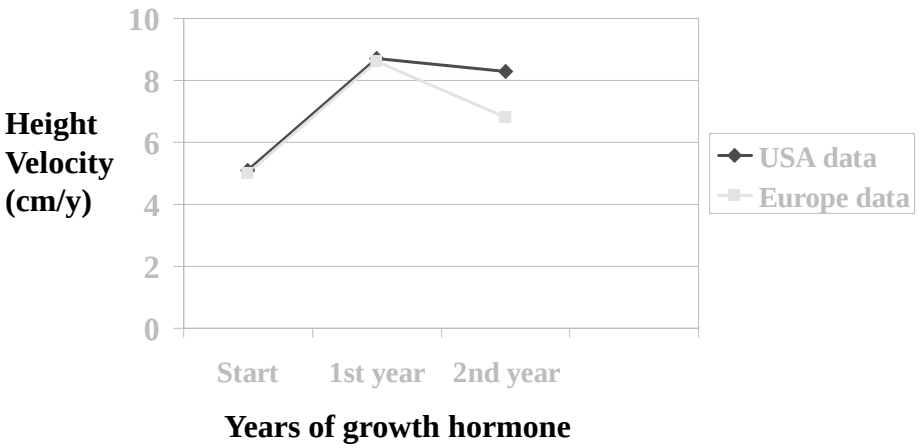
- 32 patients (12 female) average age 7.44 years. All but two (1 male, 1 female) were pre-pubertal.
- Most patients were short (27 (84%) had heights below 2nd centile)
- 26 have had anterior pituitary function performed. Mean GH level was 9.38 μg/L (normal > 10), with 19 (73%) GH-deficient.

CHARGE syndrome (KIGS; Europe): Baseline data

Height at GH start. ***Girls (left) & Boys (right)***



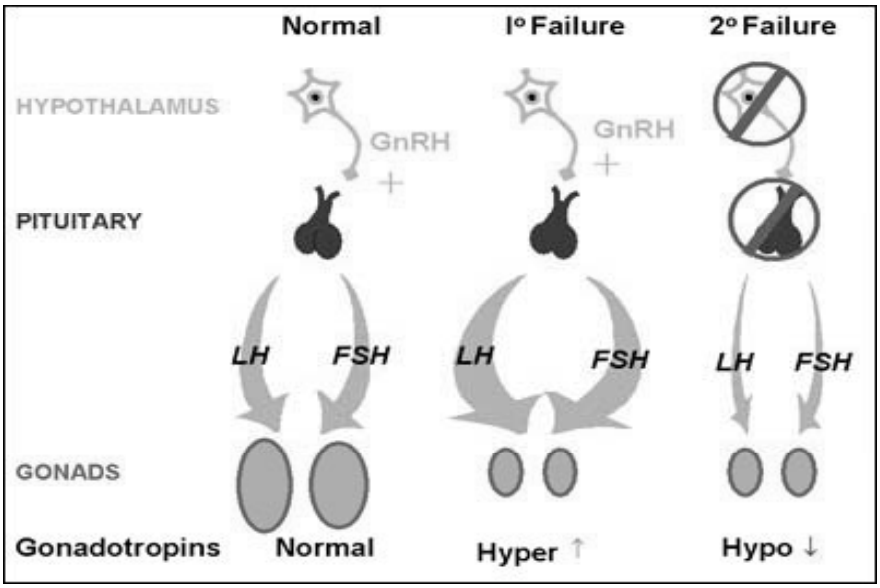
Comparison of US and European data



GENITAL AND PUBERTAL ABNORMALITIES

GENITAL PROBLEMS IN CHARGE

INFANCY	*Frequency*
Males:	
• Micropenis	85%
• Undescended testicles	60%
Females:	
• Underdeveloped labia	Very common
ADOLESCENCE	
• Delayed/arrested puberty	Very common
ADULTHOOD	
• Infertility	Unknown



Forms of gonadal failure: central vs. peripheral

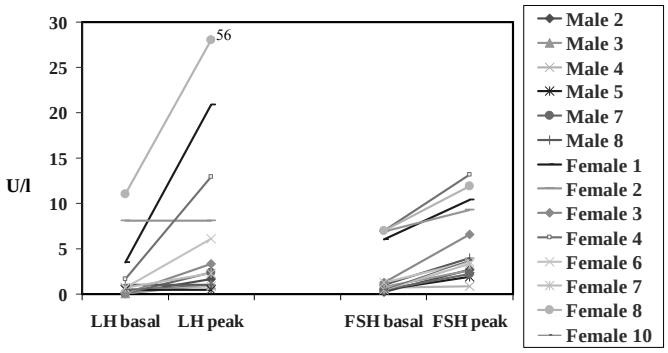
BCH experience: adolescents

- 19 patients (9 male) investigated at average age of 14.7 years (range 10.6-19.4.
- Of the boys 5 had micropenis, and 4 undescended testes requiring surgery (orchidopexy).
- 6 patients (all female) have spontaneous signs of puberty (Breast Stage 2 or Testicular volume >4ml), and 2 have had spontaneous menstruation.
- 9 have had pelvic USS scan, with pubertal uterus in 4.

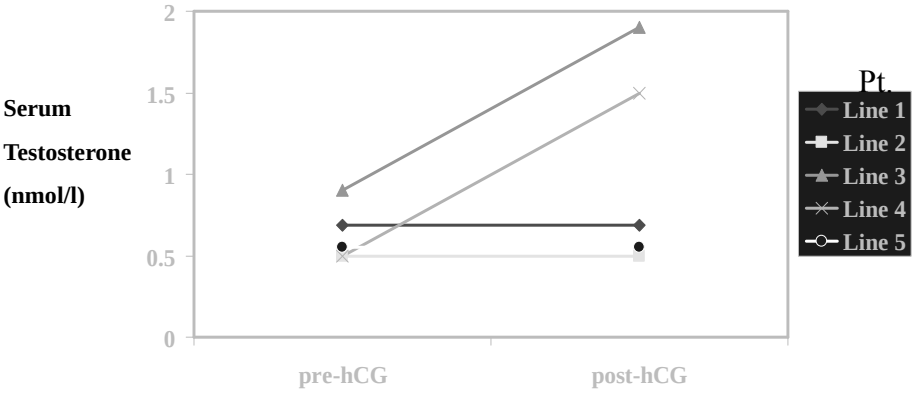
BCH experience: adolescents

- Four patients have been treated with GH: none are growth hormone deficient.
- 10 (4 males) have achieved final height, which is in the normal range ≥-2SDS (2^{nd} centile) in 5 (range -7.5 to -0.7).
- 14 patients have had genetic testing for CHD7, and 8 patients (57%) have been identified as having a mutation.

LHRH testing in CHARGE (N=13)



hCG testing in males with CHARGE (N=5)



Progress

- The seven oldest patients in Birmingham have required sex hormone replacement (oral ethinyl oestradiol in the girls, and testosterone (intramuscular depot in 3, oral in 2) in the boys.
- This has been reflected in the other patients in the UK.

HRT: Concerns in CHARGE

- Worsening behaviour.
- Inappropriate sexual behaviour.
- Menstrual bleeding (in girls).
- Persistent erections (priapism) in boys.

These concerns must be balanced against the long-term risk of osteoporosis, as much bone strength is laid down in late teens under the influence of sex hormones.

Adults with CHARGE (*LaRosa et al., 2009)*

- 8 subjects, 4 males and 4 females (aged 20-28 years), attending adult endocrine clinic.
- 3 had received GH therapy; 2 were GH deficient.
- All had received sex steroid therapy; none had attempted fertility.

- Outcomes on height and bone density compared with age matched subjects affected by primary (Klinefelter Syndrome-Premature Ovarian Failure) and secondary hypogonadism (Hypogonadotrophic Hypogonadism).

Data on patients with CHARGE syndrome and matched hypogonadal groups (*LaRosa et al., 2009*)

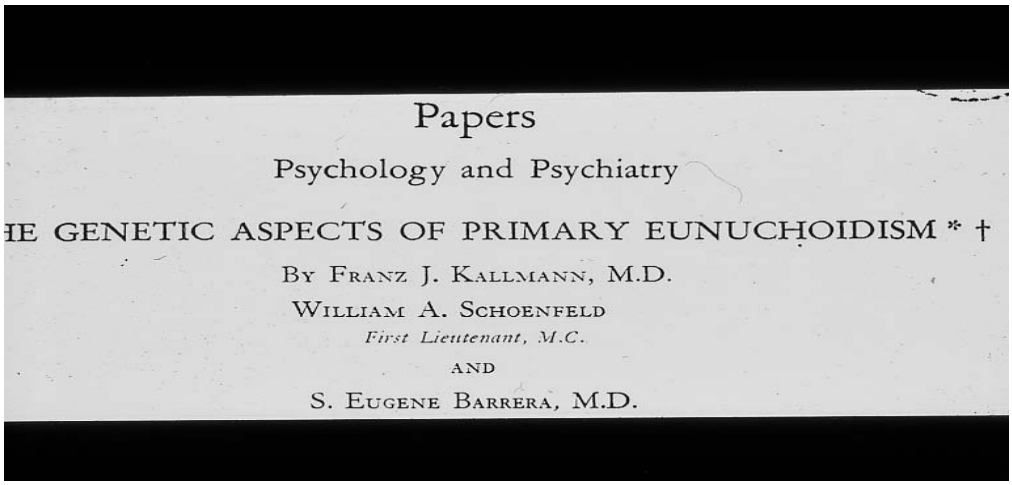
	CHARGE	Hypo-hypo	Klinefelter/ POF
Age	21.5	22	21.7
Height (cm)	158.6	165.4	172.5
Height SDS	-1.6	-1	-0.3
BMI	21.5	25.4	22.9
Spine T-score	-2.4	-1	-1.1
Hip T-score	-0.6	-0.2	-0.3

Adult data (N=11 (*UCLH & BCH*)

- All had low bone mineral density BMD., 8/11 (73%) had osteopenia and 5/11 (46%) had osteoporosis.
- Vitamin D was measured in 6 subjects and was low in 1.

CHARGE and Kallmann Syndrome overlap

Kallmann syndrome



Papers

Psychology and Psychiatry

HE GENETIC ASPECTS OF PRIMARY EUNUCHOIDISM * †

By Franz J. Kallmann, M.D.

William A. Schoenfeld

First Lieutenant, M.C.

and

S. Eugene Barrera, M.D.

Kallmann syndrome (KS)

An association of:

- Central hypogonadism (hypogonadotrophic hypogonadism).
- Absent sense of smell (anosmia).

- Other clinical features eg. absent kidney, mirror movement (synkinesia) are also variably described.

Kallmann syndrome (KS)

- May occur sporadically, or be inherited in an autosomal dominant, recessive or X-linked recessive form.
- A number of different genes (~5) have now been identified, and account for ~30% of all cases.

Common features of CHARGE syndrome and KS (FGFR1 type)

- Anosmia.
- Hypogonadotrophic hypogonadism.
- Cleft lip and palate.
- Hearing impairment.
- External ear abnormalities.
- Iris coloboma.

KS and CHARGE syndrome

- 36 patients with KS and 20 patients with normosmic idiopathic hypogonadotropic hypogonadism (nIHH) in whom mutations in 4 Kallmann had been excluded were screened for mutations in CHD7.
- Three of 56 KS/nIHH patients had mutations in CHD7.

Jongmans et al. 2008

Hypogonadotrophic hypogonadism and anosmia in CHARGE

- Retrospective review of 32 patients with CHARGE.
- 19/20 boys had micropenis and/or cryptorchidism.
- 7/9 nine boys tested < 5 months had v. low testosterone levels. LH response to GnRH stimulation was variable during the first year of life and didn't correlate with clinical abnormalities.
- No girls >12 yr (n = 7) were in spontaneous puberty, and 5 had a decreased gonadotrophin response to GnRH stimulation.
- Olfactory evaluation (n = 10) and MRI (n = 18) of the forebrain revealed defective sense of smell and abnormal olfactory bulbs in all cases.
- Mean height of 25 children >5 yr of age was –2 ± 0.2 SD score.

Pinto et al., 2005

Educational Information

SATURDAY
Breakout Session #6: 1:00 – 2:00 PM
Trillium Ballroom III, Conference Center

PERSON CENTERED PLANNING AS AN ADJUNCT TO PSYCHOEDUCATIONAL EVALUATION FOR INDIVIDUALS WITH CHARGE

Nancy Salem-Hartshorne, Ph.D.
Department of Psychology
Central Michigan University

Presenter Information: *Nancy Salem-Hartshorne* is a professor at Central Michigan University, School Psychologist, Professional Grant Writer, and Evaluation Consultant. She is co-author of numerous articles and book chapters pertaining to developmental outcomes for individuals with CHARGE syndrome during childhood and adolescence, inclusion strategies and quality educational planning tools. Her son, Jacob, age 20, was fully included throughout his elementary school years. He has experienced successful transition into a progressive secondary school experience, including integrated employment, and looks forward to supported living in his own home. Jacob is deafblind and has significant developmental delays as a result of CHARGE syndrome. Along with her expertise in psycho-educational assessment of students who are deafblind and person-centered planning, Nancy is an advocate for individuals with disabilities, teamwork, thorough planning, and forward thinking for quality life outcomes for all individuals.
918 S. Brown St., Mount Pleasant, MI 48858;TEL: (989) 773-6303 (H) (989) 289-7372 (Cell);EMAIL: harts1ns@cmich.edu

Presentation Abstract: This presentation will describe useful Person-Centered Planning techniques the presenter has used to assist families and professionals to come together to meet the needs of students with CHARGE syndrome. In both situations, the family members and professionals were at odds prior to the evaluation and Person-Centered Plan. The presentation will be highlighted with photographs and descriptions of the specific plans created, the stories behind the plans, and the positive outcomes for the students. The families of these students may be available to answer questions as well.

Person Centered Planning as an adjunct to Psychoeducational Evaluation for Individuals with CHARGE

Nancy Salem-Hartshorne, Ph.D.
Central Michigan University

Traditional Purpose of Assessment

- Diagnosis
- Eligibility
- Programming Needs
- Establishing Baselines
- Determine Appropriate Goals

How is this information typically gathered?

- Observation
- Formal testing
- Consult with teacher and sometimes parents
- Collaborative discussion during formal meetings (IEP)

What evaluation questions do we want to answer for our kids with CHARGE?

What can be done to help this student achieve Independence and self-determination in the world?

What information do we need?

- Determine strengths
- Determine needs
- Determine communication skills/preferences
- Determine preferences for people, activities, and settings
- All with an eye to the future, with natural supports in the community

Can these questions be adequately addressed through traditional means?

- Observation
- Formal testing
- Consult with teacher and sometimes parents
- Collaborative discussion during formal meetings (IEP)

No, because….

- Individuals with CHARGE have unique and complex needs that are not easily met through traditional assessment techniques.

Unique and Complex Needs

Mobility

Sensory

Health/Medical

Communication

Self-Help

Choice making

Socialization

There is so much “between the lines of life” stuff that cannot be measured through traditional means.

How can we get that missing information?

Person Centered Planning

GOALS:

- Being present and participating in community life
- Gaining and maintaining satisfying relationships
- Expressing preferences and making choices
- Continuing to develop personal competencies
- Fulfilling respected roles and living with dignity

Traditional Plan vs. Person Centered Plan

• Formal assessment	vs.	Discovering and learning who the person really is
• Deficiencies and needs	vs.	Finding capacities and gifts in the other person
• Professional teams, role and reports	vs.	Using family and friends and other connections

A bit more on PCPs

- Created by Canadian Advocates Forest, Pearpoint, Snow, and O'Brien
- Adopted by service delivery systems
- Advocates successfully lobby for inclusion in state Mental Health Codes
- Sets a different tone
- Sets a positive direction
- Changes the culture
- Contributes to quality of life

One Type of PCP

- **MAKING ACTION PLANS (MAPS)**
- What is this person's **history** and your history with this person?
- What are this person's **dreams**?
- What are this person's **nightmares/fears**?
- **Who is** this person (one or two words/phrases)
- What are this person's **strengths**?
- What are this person's **needs**?
- What can we do to **plan for the future** this person wants? (Action Planning)

CHARGE Eval #1

- Mr. B. Presenting Situation
- Mr. B. Traditional Assessment
- Mr. B. Person Centered Plan
- Mr. B. Outcomes
- What things were "between the lines" of Mr. B.'s life?

CHARGE Eval #2

- Mr. R. Presenting Situation
- Mr. R. Traditional Assessment
- Mr. R. PCP
- Mr. R. Outcomes
- What things were “between the lines” of Mr. R.’s life?

CHARGE Eval #3

- Mr. H. Presenting Situation
- Mr. H. Traditional Assessment
- Mr. H. PCP
- Mr. H. Outcomes
- What things were “between the lines” of Mr. H.’s life?

Considerations for the Individualized Education Plan

- PCP Action Plan
- What would the perfect day look like?
- Building social supports
- Using positive behavioral supports
- Goals and Objectives: What skills are required to fulfill respected roles in life and to live with dignity?

Take Home Message

If an evaluation doesn't

1) answer evaluation questions
2) result in recommendations that help "in reality" to improve the person's life in some way

Why waste your time?

SENSORY AND PHYSICAL DEVELOPMENT

SATURDAY
Breakout Session #6: 1:00 – 2:00 PM
Trillium Ballroom IV, Conference Center

BALANCE: WHAT IS IT AND HOW DOES IT WORK?

Maryann M. Girardi, PT, DPT, ATP
Physical Therapist
Deafblind Program, Perkins School for the Blind
Watertown, MA

Presenter Information: *Ms. Girardi* is a physical therapist at Perkins School for the Blind. She has worked with children with deafblindness for over 20 years. She received her Doctorate of Physical Therapy from Massachusetts General Hospital Institute for Health Professions in 2006. She has done work in modifying a standardize gross motor test to provide a more accurate measurement of skills and developed effective treatment strategies for children with CHARGE Syndrome. She continues to study the skeletal foot anomalies seen in children with CHARGE Syndrome

ADDRESS: Perkins School for the Blind, Deafblind Program, 175 North Beacon St, Watertown, MA 02472;
TEL: 617-972-7526; EMAIL: maryann.girardi@perkins.org

Presentation Abstract: The topic of balance is frequently discussed concerning children with CHARGE syndrome. This presentation will explore the many components of the human balance system including what they are, how they work, and how they interact with each other to enable humans to react to the challenges in their environment

Balance:
What is it and How Does it Work?

July 25, 2009

Maryann M. Girardi, PT, DPT, ATP
Perkins School for the Blind
Deafblind Program

Components of Balance

Sensory Systems

Somatosensory –the system that detects experiences labeled as touch or pressure, temperature, pain (including itch and tickle) and those that belong to proprioceptions.

Skin receptors
- Touch
- Pressure

Proprioceptive Receptors- muscle movement and joint positioning
- Muscle Spindles - located in the muscle belly
  detect changes in muscle length
- Golgi Tendon Organs- located in the tendon
  detect muscle tension
- Joint Receptors- located in the joint capsule
  detect joint pressure and position

Visual
- Detect orientation in space
- Provides environmental information

Vestibular System

Three semicircular canals- positioned in 3 different planes to detect up/down, side to side, and tilting movement of the head through the activation of hair cells by the movement of inner ear fluid (endolymph)

Otoliths (utricle and saccule)-detect linear acceleration

Vestibular Reflexes

Vestibulo-ocular reflex- ensures best vision during head motion by moving the eyes contrary to the head to stabilize the line of sight in space

Vestibulo-spinal reflex- stabilizes the body

Vestibulo-collic reflex- stabilizes the head

Motor Systems

Range of Motion- motion available in each joint
Motor Function- how the body moves against gravity

Central Nervous System Integration

Brain Stem- visual reflex center, pathway for motor and sensory from the brain to the body
Cerebellum- processes information from sensory and motor systems to provide coordinated muscle movement

Balance Reactions

Strategies

Hip
Ankle
Step

Treatment Approaches for Balance Deficits

Computer based feedback systems

Balance Master- uses visual feedback in conjunction with pressure sensitive footplates to assist in the development of static and dynamic balance

Wii Fit™- also uses pressure sensitive foot plate with visual and auditory feedback to develop static and dynamic balance skills through games

Non-feedback based systems

Exercise to increase muscle strength
Assistive devices to increase base of support

Practice in situations where balance is challenged to establish successful motor plans

Characteristics of CHARGE Syndrome that can affect balance

Colobomas- limit visual information
Semicircular canal anomalies- limit vestibular (angular) information
Low muscle tone- slower to contract and cause joint movement
-decreased ability to maintain contraction
Proprioceptive system impairments- limit information on joint and muscle position
Joint laxity- provides an unstable skeletal base
Skeletal alignment
flat feet – provide an unstable base of support foot is not working as designed altering body alignment
scoliosis- alters body alignment
cervical fusions- limits neck/head movement which provides information about the head's position through the proprioceptive system

Behavioral Information

SATURDAY
Breakout Session #7: 2:10 – 3:10 PM
Trillium Ballroom I, Conference Center

WHAT'S THERE TO STRESS ABOUT? THE CASE OF CHARGE SYNDROME

Kasee K. Stratton

Doctoral Student in School Psychology
Central Michigan University

Presenter Information: Kasee Stratton is a doctoral student studying to become a school psychologist at Central Michigan University. She is investigating how stress and pain impact challenging behavior in CHARGE. She has been researching CHARGE and participating in family consultations for four years and has presented at several CHARGE conferences.

ADDRESS: 1637 E. Broomfield Apt 5D, Mount Pleasant, MI 48858;
TEL: (989) 621-2303; EMAIL: strat1kk@cmich.edu

Presentation Abstract: Interviews about stress were conducted with young adults. These interviews focused on stress experiences with specific attention paid to behavioral reactions. Themes based on education, family, friendships and employment would be presented. Additionally this presentation would also focus on pain experiences in CHARGE Syndrome. The experience of chronic pain would be explained along with research findings showing a lack of appropriate pain management in populations that cannot vocally communicate, such as infants and developmentally disabled. The focus would be to include pain in our understanding of CHARGE behaviors (such as outbursts, unexplainable crying, behavioral disruptions, etc) and ways to identify pain non-vocally that are reliable and valid. Without appropriate identification, pain management and treatment cannot be provided.

What's there to stress about?

The case of CHARGE Syndrome

9th International CHARGE Syndrome Conference 2009
Bloomingdale/Chicago, IL
Kasee K. Stratton
Central Michigan University
Strat1kk@cmich.edu

Where it all began?

- Children, even high-functioning children, suddenly lose control
- Strike out
- Express remorse
- Understanding such challenging behaviors

Stress and Early Pain Experiences

- Stress Experiences:
  - Increased academic difficulty around grade 3—cognitive to concrete operational thinking
  - Concentration with sensory deficits
  - Regulatory disorder and maintaining emotional control

Stress and Early Pain Experiences

- Pain Experiences:
  - Multiple medical procedures/surgeries
  - Common CHARGE characteristics, i.e., low muscle tone, jaw discomfort, cranial nerve anomalies, migraines, reflux, gastrointestinal problems, ear infections, etc.
  - Suspect early pain experiences may impact neurodevelopment which may also lead to challenging behavior

CHARGE and High Pain Threshold

- Reported to have a high pain threshold- based on behavior
- Currently, no study has been conducted to identify the pain experiences of individuals with CHARGE
- Suspect they display pain experiences differently than typically developing children

Research on Pain

- Difficult to measure—subjective experience
- "Golden Rule"
  - Identify and treat pain through patient self-report
  - Communication difficulties

Assessment of Pain and Developmental Disabilities

- Studies in the past have largely excluded this population due to:
  - Inability to vocally self-report
  - Pain experiences may differ from the general population (Neurological/Developmental)
  - Advanced medical conditions could complicate the assessment

Developing Appropriate Pain Assessment Tools

Facial Reactions/Expressions

Body Movement

Eating Patterns

Sleeping Patterns

SIB

My child on a good day vs. My child on a bad day

Some interesting findings…

- In 2000, found that children with developmental delays displayed a less intense distress response to everyday pain experiences than non-delayed children
  - More likely to display no reaction and seek help less often
- In 2004, a study analyzing infant expressions during routine immunizations found infants with DD were likely to show pain in facial expression less clearly than typically developing infants

Why do we miss the pain signs?

- Most common reason for lack of treatment is lack of detection
  - Need for appropriate pain assessment tools and caregivers understanding of behavior
- Individuals may not report pain or underreport pain to avoid further hospitalizations or testing—potential signs of Post Traumatic Stress Disorder (PTSD)

PTSD

- Documented in children with a variety of life-threatening medical illnesses
- Children with liver transplants and cancer survivors have significantly greater PTSD symptoms than those compared to routine surgical procedures

PTSD

- Individuals with CHARGE could have some potential sources of PTSD:
  - Life-threatening surgeries beginning very early in life
  - Long-term hospital stays
  - Long recovery periods from medical procedures
- Trauma exposure can lead to loss of self-regulation, aggression toward others and self-destructive behaviors

Hypothesize…

- …that individuals with CHARGE do not have a high pain threshold; rather they express their pain experiences differently through behavior and other such cues
- Experience more chronic pain than the general population and not report it as often— “learn to deal with it”

Other Stressful Experiences

- Need to look to other stressful experiences for challenging behavior
- Remember those children who lashed out at their best friends…what else is causing such behavior?

Stress and Disability

- Research on parenting stress and family stress, but no research on the individual with the disability's stress outside of research on PTSD

Stress and CHARGE

- Interviews conducted with high-functioning young adults
  - Specific attention was given to behavioral reactions to stressful situations
  - School experiences, social and family relationships, and work experience

School Themes

- Lack of appropriate communication
  - "I get tired easily at school and I find it hard to concentrate. Sometimes my behavior is bad. I don't know why I do it and I hate it when it happens."
  - FM system for class only

School Themes

- Lack of understanding from school personnel on student's hearing and vision loss and multi-sensory impairments
  - "I had an FM system to help with my hearing but I found that I didn't have the adequate services for my vision…for whatever reason, I found it hard to communicate what I was being taught."

School Themes

- Emotional Immaturity
  - "In H.S. I would cry a lot, I would just get so upset."
- Rapidly tiring from concentration/focusing on environment with sensory deficits
  - "I often came home and I just wanted to lay down on the couch after school… I needed more breaks"
- Bullying

Friendship Themes

- Recognized need for social interaction
  - "I hate being sick and missing out on school. I want to be at school everyday. I want more people like me around."
  - "Friendships were always hard to make and they still are..I don't know how to strike one up."
- Communication difficulties
  - "I get frustrated especially when friends won't play with me. I don't know how to tell them what I mean."
  - "It's hard to be at the mall and hear and see my friend at the same time. I can't follow our conversations well, it just makes it hard to communicate.

Friendship Themes

- Concerns about the "social rules" of friendships
  - "Am I bothering you?"
- Need for friends who will remain friends despite unexplainable behavior at times

Family Themes

- Need for positive relationship between the parents and the school
- Need for acceptance of physical characteristics
  - "…then I wouldn't have been as self-conscious about my looks."
- Supportive "…they fought a lot for me."
- Allow for more independence

Other Themes

- Physical Abuse
- Sexual Abuse
- Children with communication and learning disabilities are as much as 2 to 3 times more likely to be victims of child abuse than typically-developing children
- Association with conduct disorder

Dealing with Anxiety

- Obsessive-Compulsive Disorder
  - Anxiety disorder; individual experiences obsessions (involuntary thoughts/impulses), which lead to a repetitive behavior called a compulsion. The compulsion is done to rid the unwanted obsessions or thoughts
- Changing clothes, gelling hair, dialing a pattern on the telephone, picking at skin, etc.

How do we help?

- Realize early pain experiences are likely to play a role in a child's challenging behavior
  - Recognition of what my child is like on a good day and what is different on a bad day…could pain be a factor?
- Communication! Need appropriate ways to communicate with others
  - Use of technology
  - Staff trained in various modes of communication

How do we help?

- An understanding of CHARGE syndrome and it's many facets
  - DB, multi-sensory impairments, pain experiences
  - Educate staff and caregivers about CHARGE
- Support from OT, PT, O&M, and speech therapists in the classroom for adapted classroom furniture, elimination of stressors and to assist with communication

How do we help?

- One-on-one support for access to information, serve as an intervener for communication and safety
- Avoid over-demanding workloads
- Allow for breaks, positive motivation
- Development of a circle of friends

Where do we go from here?

- Unknowns:
  - Understand the function of OCD—pleasure or anxiety?
  - Improve our attention to pain and developing proper ways to assess pain
- My thesis:
  - Investigation of multi-dimensional pain assessments appropriate for children/adults with CHARGE
  - To provide parents/caregivers/educators with the behavior and communication signs that may signify pain within this population
  - Contact: Kasee Stratton strat1kk@cmich.edu

Sensory & Educational Information

SATURDAY
Breakout Session #7: 2:10 – 3:10 PM
Trillium Ballroom II, Conference Center

THE NICU EXPERIENCE: ITS IMPACT AND IMPLICATIONS

Barbara Purvis, M.Ed.
Technical Assistance Provider
National Consortium on Deaf-Blindness
Shawnee Mission, KS

Presenter Information: Barbara has worked in both general and special early childhood education, including specialized training to provide developmental care to preterm and medically fragile infants in intensive care nurseries. She first met children with CHARGE Syndrome when working as an early intervention provider and, with NCDB, has continued her work on behalf of children with combined vision and hearing loss. Having "transitioned" three daughters, including one who received special education supports, Barbara is an experienced advocate for individuals with disabilities.

4330 Shawnee Mission Pkwy, Suite 108, Shawnee Mission, KS 66205; EMAIL: barbara.purvis@hknc.org : TEL: 913-677-4562

Presentation Abstract: Preterm infants complete their development in an environment markedly different than their mother's womb. The multi-sensory experiences in an intensive care nursery impact immature systems in ways that interfere with typical prenatal development. Regardless of whether they are born early, children with CHARGE Syndrome often spend extended time in the NICU, encountering experiences difficult for their compromised sensory systems to handle. This session examines implications of the NICU experience for both infants and families.

The NICU Experience: Its Impact and Implications

Barbara Purvis, M.Ed.

CHARGE Conference for Professionals

June 23, 2009 – Bloomington, IL

Overview

1. Impact on Babies and Families

Sensory and developmental implications

Providing supportive care

2. Implications for Service Providers

Increased level of awareness and sensitivity

Importance of early collaboration

IMPACT on BABIES

- Increased survival rate of younger, lower birth weight and medically fragile infants
- NICU is a very unnatural environment
- Babies with (or suspected of having) CHARGE already have altered sensory systems
- Immediate medical concerns take priority over developmental and educational concerns

IMPACT on FAMILIES

- Emotional roller coaster
- Information overload
- Bonding is difficult
- Job stress
- Financial stress
- Strained relationships
- Early challenges can have long-term implications

PRENATAL SENSORY DEVELOPMENT

- **Typical sensory development follows a sequential maturation process**

Tactile ➔ Vestibular ➔ Gustatory ➔ Olfactory ➔ Auditory ➔Visual

- **Each system interacts with and impacts every other system**
- **Any compromise to one system affects much more than just the compromised system**
- **Infant outcomes can be improved through intervention that supports the developing infant and its vulnerable sensory systems**

What is DEVELOPMENTALLY SUPPORTIVE CARE?

- Based on NIDCAP principles (*Newborn Individualized Developmental Care and Assessment Program*)
- Assesses overall nursery environment
- Assesses individual infant's environment
- Provides individualized care to babies
- Provides recommendations to family and medical staff to enhance infant development

NIDCAP ASSESSMENT

- Observe and record a caregiving procedure
  - Heart and respiration rates
  - Oxygen saturation levels
  - Color
  - State
  - Responses to handling/changes in environment
  - Stress signals
  - Self-calming behaviors
  - Motor activity
- Use information to report infant progress and make recommendations for individualized care

Supportive Care Practices

- **Attention to environment**
  - ➔ Overall nursery environment
    - Arrangement of equipment and supplies
    - Ideas for decreasing light, sound, activity levels
  - ➔ Individual infant's bedspace
    - Type and configuration of bedding/clothing
    - Appropriateness of pacifier
    - Ideas to assure bedspace is appropriate based on baby's current gestational age

Supportive Care Practices

- **Positioning**
  - ➔ Encourage hands-to-mouth, midline alignment
  - ➔ Arms and legs flexed and tucked
  - ➔ Nests to provide security, boundaries to facilitate self-regulation/provide proprioceptive input
  - ➔ Kangaroo holding
- **Feeding**
  - ➔ Determine readiness
  - ➔ Choose appropriate nipple
  - ➔ Model appropriate strategies for staff, families

Supportive Care Practices

- **Care giving strategies**
  - ➔ Education regarding infant cues
  - ➔ Planning to minimize handling, over-stimulation
- **Education and support for families**
  - ➔ Principles of developmentally supportive care and recommendations individualized for their baby
  - ➔ Reading infant stress signals
  - ➔ Strategies for being involved in their baby's care
  - ➔ Resources and referral to early intervention programs/support groups/social service agencies

Implications for Service Providers

Increased awareness and knowledege of developmental implications

- Altered development often results in
  - Challenges with state regulation
  - Challenges with attention
  - Challenges with sensory integration
  - Challenges with sensory defensiveness

Implications for Service Providers

Increased awareness and sensitivity

The…infant is in various stages of development to which we place unrealistic demands. The infant is at the mercy of its care providers . . . How we provide care and what we do or don't do can have a lasting effect on the infant and family.

Linda M. Lutes, M.Ed., Infant Development Specialist

Implications for Service Providers

Increased awareness and sensitivity

- Give families of young children space and time – they need and deserve it
- Realize that family behaviors viewed as barriers may have deep-rooted origins resulting from the family's NICU experiences
- Be careful how you "use your words"

Implications for Service Providers

Increased collaboration

- Develop a team for coordinated care – medical, developmental and educational
- Start as early as possible
- Find out about NICU and PICU experiences of children you work with
- Education early intervention providers about CHARGE
- Use what you've learned to inform your intervention and instruction

Selected Resources

Als H (1999) *Reading the premature infant*. In Goldson E (ed.) Nurturing the Premature Infant: Developmental Interventions in the Neonatal Intensive Care Nursery. New York: Oxford University Press, 18-85.

Als H (1992) *Individualized,family-focused developmental care for the very low birth weight preterm infant in the NICU.* In Friedman SL, Sigman MD (eds.) *The Psychological Development of Low Birth Weight Children*. Norwood, NJ: Ablex Publishing, 341-388.

Als H (1988) *Toward a synactive theory of development; Promise for the assessment and support of infant individuality.* Infant Mental Health Jo*urnal* 3:229-243.

Als H (1986) *A synactive model of neonatal behavioral organization: Framework for the assessment and support of the neurobehavioral development of the premature infant and his parents in the environment of the neonatal intensive care un*it. In Sweeney JK (ed.), The High-Risk Neonate: Developmental Therapy Perspectives. Physical & Occupational Therapy in Pediatrics, 6:3-55.

Blackburn ST, VandenBerg KA (1998) *Assessment and management of neonatal neurobehavioral development*. In Kenner C, Lott JW, Fandermeyer AA (eds.), Comprehensive Neonatal Nursing, WB Saunders Company

Selected Resources

Fern, D. and Graves, C. (1996). *Developmental Care Guide for Families with Infants in the NICU* (1996), Weymouth, MA: Children's Medical Ventures.

Newborn Individualized Developmental Care and Assessment Program (NIDCAP) Program Guide (1998) National NIDCAP Training Center, Boston, MA: Children's Medical Center Corporation.

Understanding My Signals: Help for Parents of Premature Infants (1989, 1996) Hussey-Gardner, B. Palo Alto, CA: VORT Corporation

VandenBerg KA (1995) Behaviorally supportive care for the extremely premature infant. In Gunderson LP, Kenner C (eds.), *Care of the 24-25 Week Gestational Age Infant: A Small Baby Protocol*. Petaluma, CA: NICU Ink, 145-170.

VandenBerg KA (1993) Basic competencies to begin developmental care in the intensive care nursery. *Infants and Young Children* 6:52-59.

Zaickin, J. (1996). *Newborn Intensive Care: What Every Parent Needs to Know* (1996) Petaluma, CA: NICU Ink.

Contact Information

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Educational Information

SATURDAY
Breakout Session #7: 2:10 – 3:10 PM
Trillium Ballroom III, Conference Center

SUCCESFUL TEACHING STRATEGIES AND KEY COMPONENTS FOR USE IN EDUCATIONAL AND COMMUNITY SETTINGS

Martha M. Majors & Sharon Stelzer
Deafblind Program
Perkins School for the Blind
Watertown, MA

Presenter Information: *Martha Majors* is an assistant education director at the Perkins school for the Blind in the Deafblind Program. *Sharon Stelzer* is a teacher in the same program. Both presenters have been professionally involved with CHARGE syndrome for a number of years.

ADDRESS: 175 N. Beacon St Watertown MA 02472; TEL: 617-972-7509
EMAIL: Martha.Majors@Perkins.org and Sharon.Stelzer@Perkins.org

Presentation Abstract: This presentation is focused on teaching strategies and other factors that are critical in both educational and community settings for individuals with CHARGE syndrome. Some of the specific topics include sensory loss, communication, curriculum, teaching strategies, and environments. The information for this presentation was gained from the Deafblind Program at Perkins School for the Blind as well as national and international consultation with school districts and families regarding educational strategies and communication.

9th International CHARGE Syndrome Conference, Indian Lakes Resort, Bloomingdale, IL, July 24-26, 2009

CHARGE Syndrome Conference

Successful Teaching Strategies and Key Components for use in Educational and Community Settings

Presenters:
Martha M. Majors
Sharon Stelzer
Perkins School for the Blind

Overview of Presentation

- Overview of Communication
- Discussion of successful educational strategies for children with CHARGE
- Discussion of environmental influences that can create positive outcomes for children (the Checklist)
- Overview of Curriculum Components
- Share how these strategies can be applied to real life situations

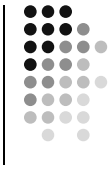
Sensory Loss and the Impact on Learning

- It is important for the team to take careful notice of the functional vision of the child as well as the hearing loss, the impact of hearing loss on communication and the total effects on day-to-day functioning.
- Several types of accommodations may be necessary in the educational setting and these should be written into the child's IEP.

COMMUNICATION

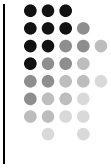
- **Many students benefit from a total communication approach**
- **This may include all or some of the following: sign language, speech, gestures, simple sign language, print/Braille, facial expression, symbols (Mayer-Johnson Symbols)**
- **Generally, the modes for each child are highly individualized and the team, including families are the decision makers.**

CURRICULUM



- Curriculum should be FUN, age and content appropriate; Children with CHARGE are very capable and must be challenged through access to a highly motivating and flexible curriculum. Children with CHARGE benefit from a curriculum that incorporates a variety of skills across settings. Once goals are achieved other skills can be added to enhance knowledge and growth.

CURRICULUM COMPONENTS



- Child Centered Curriculum
- Exciting content
- Age appropriate
- Functional Activities
- Natural learning environments
- Variety of activities
- Explore the range of curriculum options that include: Preschool, Early Academics, Academics, Functional Academics, Vocational

What Every Child with CHARGE Must Learn

- How to be an effective Communicator
- How to make choices
- How to help out
- How to be social
- How to be a part of a group
- How to Negotiate
- How to be Organized
- How to anticipate activities (calendar system)
- How to take turns
- How to cope with behaviors

Teaching Strategies

- Choice making
- Beginning-middle-end
- Partial vs. Full participation
- Motivation
- Follow student's lead
- People preferences
- Modeling
- Prompt levels (hand under hand)
- Awareness of hands/touch
- Pause time for response
- Task Analysis
- Sharing
- Clear expectations

Teaching Strategies continued

- Negotiation
- Structure and routine
- Natural routines
- Active vs. passive learning
- Organizational skills
- Boundaries

ENVIRONMENTS

- Children with CHARGE often require frequent **sensory "breaks";** there should be adequate space within classrooms and other spaces in order to ensure access for the child who needs time to relax, to do something that **calms** and allows time to be **quiet**;
- **Fatigue** can overcome a child quickly and the ability to take a quick "break" is critical the success of the child to maintain a calm demeanor

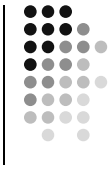
ENVIRONMENTS

- A flexible schedule that allows for “breaks” is important
- Staff should be aware of “signals of behavior” and how to respond to the child
- Prevention is the key to a successful education for a child with CHARGE!

ENVIRONMENTS

- Physical environments can be arranged to motivate and intrigue the child with CHARGE.
- A flexible environment provides opportunities for the child with CHARGE to change and grow.
- Optimal learning can take place for the child with CHARGE.

Resources



- **CHARGE Syndrome Website Professional Series (Majors, Stelzer and Ryan)**
- **Perkins School for the Blind Webcast Series on CHARGE Syndrome (Summer 2009)**

INFORMATION FOR YOUNG ADULTS

SATURDAY

Breakout Session #7: 2:10 – 3:10 PM

Trillium Ballroom IV, Conference Center

TAKING CHARGE OF YOUR LIFE, DEVELOPING A POSITIVE VISION TOWARDS EMPLOYMENT

Michael Fagbemi

Technical Assistance Specialist

National Consortium for Deaf-Blindness (NCDB)

&

Joe McNulty

Director, Helen Keller National Center

Sands Point, NY

Presenter Information: *Michael Fagbemi* has worked at the Helen Keller National Center for thirteen years in a number of different capacities in direct services as a habilitation staff, as a residence coordinator and as a senior behavioral specialist. He is currently is working on a children's education grant (NCDB) National Consortium on Deaf Blindness as a Technical Assistance specialist and he support Joe Mcnulty on special activities in Washington DC. Mr. Fagbemi also provides technical assistance to families, systems, youth and service providers in the southern region including the Virgin Islands.

ADDRESS: 141 Middleneck Road, Sands Point Long Island 11050 TEL: 516 944 8900 x 202; EMAIL: mike.fagbemi@hknc.org

Joe McNulty is the Director of the Helen Keller National Center. He participates in the National Consortium for Deaf-Blindness as a senior scientist. He has made many contributions in the area of deaf-blindness over a career that has spanned many years. On June 26 of this year, he and a contingent from the HKNC met with President Obama in the Oval Office to create an awareness of the challenges and needs associated with deaf-blindness.

Presentation Abstract: Young adults with dual sensory impairments are not often given the opportunity to live and work in their communities of choice. This lack of opportunity further limits a young adult's exposure to new experiences and as a result impacts on their ability to socialize effectively live independently and develop work skills beneficial to employers. The focus on educational mandates and the pressures that are associated with it have had a profound effect on youth who are in need of independent living skills. One important component of living independently is work. We will discuss the national picture and briefly explore an employment model based upon a negotiated employment relationship between employer and job seeker that fits the needs of both.

Taking charge of your life
"*developing positive vision towards employment*"

9TH International CHARGE Syndrome Conference

July 25th 2009

Mike Fagbemi & Joe McNulty

Objectives

- Provide an overview of employment & the national agenda.
- Provide an overview of an emerging job development model.
- Share current southeast efforts to explore this model and lessons learned.
- Offer employer perspective of hiring practices

National Transition Follow-up Study of Youth Identified as Deaf blind "Parent Perspectives" (Petroff, 1999)

- **The majority of students did not received adequate transition planning;**
- **Only 40% of the students' and/or parents' interests and preferences were identified as a component of the transition planning process;**
- **Only 12% of youth and their families were involved in person-centered planning;**
- **Of those who engaged in transition planning, the majority didn't begin until one year prior to school exit;**
- **A very few number of students received community based experiences (vocational or otherwise)**

What do we know about DB Youth

- Data suggests that there is a slight improvement in the employment outcomes for youth with deafblindness as compared to the 1999 and 2001 study
- Data suggests that more youth are volunteering than working
- Data suggests that there youth remain primarily living with their parents;
- Data suggests youth who are deafblind continue to be isolated within their communities.

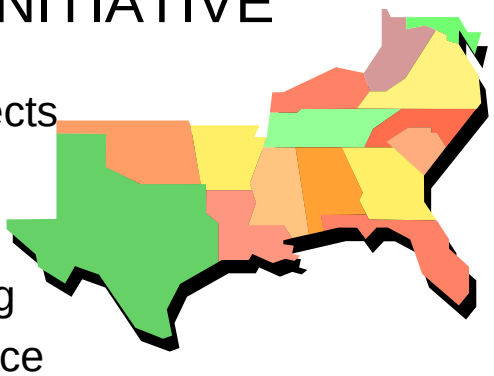
(Dr. Jerry Petroff)

Historical context…….

- Supportive Employment has stood the test of time
- Opened doors for people with complex needs that would not have historically been able to be included in typical work settings.

NCDB INITIATIVE

- Ten state DB projects
- Two young adults
- Local team
- Web based training
- Technical assistance
- On site training



Discovery

- Foundation of Customized Employment
- Exploration of a persons life
- Who is he?
- A journey to discover information about a person that can be translated to work tasks

- Discovery seeks to identify already – existing information rather than developing information solely for the purposes of evaluation or diagnosis

Scenario # 1

- A census taker makes an appointment to come to your house for purposes of gathering on family members, income, housing values. The meeting is held in your living room. The census taker asks very discrete questions from a pre – set and writes information while you respond. The offer of a beverage is declined and as soon as the interview is complete, the census taker thanks you and leaves for another appointment down the street

Scenario # 2

- Your new next door neighbor drops by to borrow the traditional cup of sugar. You welcome the neighbor in with an offer of a cup of coffee, which is accepted. You both take a seat in your living room and & a conversation begins that touches on topics as far – ranging as weather, sports, the local school system, directions to the shopping mall & your mother –in law's visit next week. After about an hour, the neighbor says its time to get back home and the conversation continues out the front walk & re –establishes itself when your spouse comes with new topics & sharing. Finally after about an hour & a half your neighbor leaves to go home with promises of getting together sometime next week.

Questions to consider :

- Which scenario was most successful in determining who we are and how we feel?
- Which scenario resulted in more discrete information that could be used for social / planning?

Discovery is most effective when we have both

- Discovery can be viewed as simplistic
- The process is often accelerated
- We think we know the person so therefore there is nothing else left to know

Types of Discovery

- Facilitated Discovery
- Group Discovery
- Self Discovery

Customized Employment

- A blend of services that combines good employment practices to assist in developing a negotiated job which is based upon the discrete needs of the employer and the interests and strengths of the job seeker (Dept. of Labor)

Talking points

- Salaries are negotiated at the prevailing wage.
- Job seeker is represented by person who can capture the best of them.
- The focus is on contribution as opposed to competition.

The Basic tenets of Customized

- Interests of person
- Conditions of employment
- Contributions

WORKERS WITH DISABILITIES:

Ready for Tomorrow's Jobs Today

ODEP

Challenges

Hiring Challenges

- Nature of the work
- Not knowing accommodation costs
- Cannot find qualified candidates
- Attitudes NOT considered a major challenge

Hiring Concerns

- Costs
- Lack of skills and experience
- Less safe and productive

WORKERS WITH DISABILITIES:
Ready for Tomorrow's Jobs Today

Persuasiveness of Information

Most Persuasive:

- Satisfactory performance, attendance, and retention
- Increases in company's productivity
- Benefits to company bottom line

Less Persuasive:

- Testimonials from line managers
- Benefited nationally recognized companies
- Addressing concerns about cost

*Only companies that do not actively recruit.

WORKFORCE OPPORTUNITY LEADERSHIP

Negotiation points

- The discrete job tasks and description
- The hours, rate,
- Natural supports , accommodations
- What can you offer my bottom line
- How do I retain you
- There are myths about disabled people

- “People seldom see the halting and painful steps by which the most insignificant success is achieved.”
  Annie Sullivan

Employment is only a piece of the puzzle

- Expectations
- Social Networks
- Independent Living
- Transportation
- Access
- Communication
- Public Perception

INFORMATION ON EARLY EDUCATION

SATURDAY
Breakout Session #8: 3:40 – 4:40 PM
Trillium Ballroom I, Conference Center

LOOK AT ME NOW: THE IMPACT OF EARLY INCLUSION ON OUTCOMES

The Costello Family
Heather, Kevin, Jessica, Melissa and Shane
Pulaski, TN
&
Jennifer Miller and Donna Consacro
Tennessee Deafblind Project (TREDS)
Monroe Carell Jr. Children's Hospital at Vanderbilt

Presenter Information: *The Costello Family* is from a small rural community near Pulaski, Tennessee. Kevin is retired from the US Navy and Heather is employed part time in her own business. Their oldest daughter Jessica is a freshman in college. Melissa, their middle child (who has CHARGE syndrome) is a senior in high school. Their son, Shane, is a sophomore in high school. ADDRESS: 221 Robinson Road, Pulaski, TN; TEL: 931-363-7257; EMAIL: kevin_costello@bellsouth.net

Jennifer Miller is the Technical Assistance and Training Coordinator for the Tennessee Deafblind Project (TREDS). She provides technical assistance, training and support for teachers, service providers, medical personnel and families. She is a Certified Child Life Specialist and Certified Educator of Infant Massage with 19 years of experience working with children who are medically fragile and those with disabilities. TEL: 615-322-7194; EMAIL: jennifer.l.miller@vanderbilt.edu

Donna Consacro has worked as the Family Information and Resource Specialist for the Tennessee Deafblind Project (TREDS) for 15 years. Donna works with the families of children on the Tennessee Deafblind Census to assist them with locating a variety of resources and information to support to their children.She is a wife and the mother of three adult daughters. Her oldest daughter was born profoundly deaf. She is grandmother to twin 4-year-old granddaughters and their 2-year-old brother, all of whom are profoundly deaf. She also has a 2-year-old grandson who is hearing. TEL: 615-322-8279; EMAIL: donna.consacro@vanderbilt.edu

ADDRESS: TREDS, 3401 West End Avenue, Suite 460 West, Nashville, TN 37203

Presentation Abstract: This panel presentation will showcase a young woman with CHARGE syndrome who has been successfully included in home, school and community throughout her life. The panel will include this young woman, her parents, siblings, and Tennessee Deafblind Project staff. As well as triumphs we will discuss barriers and strategies to overcome them. We plan to offer both practical advice as well as inspiration for families, their children and the people who work with them.

Look At Me Now: The Impact of Early Inclusion on Outcomes

Presenters:
Costello Family, Pulaski, TN
Donna Consacro and Jennifer Miller Project TREDS,
Monroe Carell Jr. Children's Hospital at Vanderbilt

9th International CHARGE Syndrome Conference
July 24-26, 2009 Chicago, Illinois

Agenda

- Introductions
- Lessons Learned Worth Sharing
  - Mom & Dad
  - Siblings
- Today & Tomorrow
  - Melissa
- Recap of Lessons Learned
- Question/Answer & Evaluation

Who Are the Presenters?

- Donna Consacro & Jennifer Miller– TN Deafblind Project TREDS
- Heather & Kevin Costello – Parents
- Jessica & Shane –Siblings
- Melissa Costello – High school student

Through the Eyes of Mom & Dad: The Tribulations and the Triumphs

- Melissa's birth and the early years
  - Medical miracles and misconceptions
- The preschool years
  - Leaning about inclusion
- The early school years
  - Family values versus professional opinions

Through the Eyes of Mom & Dad: The Tribulations and the Triumphs

- Middle school and high school
  - Out and about in the world
- What the future holds
  - Reaching for the stars

Through the Eyes of Siblings

- The early school years
  - Realizing Melissa is special
  - Affect on the family
- Middle school and high school
  - Friends and community
- What the future holds
  - Believing in Melissa

The Costello Family

Words from Melissa

- What's happening now
  - At school
  - In the community
- My hopes and dreams for the future
  - At school
  - In the community
  - After school

A Recap of Actions

- Finding emotional support
- Perseverance and persistence
- Believing in Melissa's abilities
- Dealing with naysayers
- Identifying additional resources as needed

It's a Wrap!

- Questions and Answers
- Evaluation

Resources & References

- Tennessee Deafblind Project
  - http://www.treds-deafblindproject.com
- DB-Link
  - http://nationaldb.org/ISLibrary.php
- National Consortium on Deafblindness
  - http://nationaldb.org/index.php
- Think College.Net – Post Secondary Education
  - http://www.thinkcollege.net
- Helen Keller National Center
  - http://www.hknc.org

Medical Information

SATURDAY
Breakout Session #8: 3:40 – 4:40 PM
Trillium Ballroom II, Conference Center

ANESTHESIA COMPLICATIONS IN CHARGE SYNDROME

Kim Blake, M.D.
&
Jill MacCuspie

Dalhousie University
Halifax, Nova Scotia

Presenter Biography: *Dr. Kim Blake* began her involvement with CHARGE more than 25 years ago at Great Ormand Street Hospital in the UK, where she lectured on CHARGE and published several of the earliest papers describing the syndrome. She was instrumental in organizing the UK family support group. Since moving to Canada, she has continued to be involved with the CHARGE Syndrome Foundation. She has been an invited speaker at every conference and received funding from the Foundation for several of her research projects. Kim's research career continues to focus on CHARGE, particularly the issues of the adolescent and adult population. She routinely involves students in her research, both educational and clinical. Most of her students have had abstracts and/or papers published, some even with first authorship. Kim has recruited many local medical faculty members in her research and developed a center of excellence for research and knowledge in CHARGE syndrome. She is regularly asked to present on CHARGE syndrome, both nationally and internationally. Her most recent research projects are on the effects of anesthesia in CHARGE syndrome and the feeding difficulties in children with CHARGE syndrome.

ADDRESS: Dalhousie University, 5850/5980 University Ave, Halifax, NS B3K 6R8
TEL: 902-488-0128; FAX: 902-470-6913; EMAIL: kblake@dal.ca

Jillian MacCuspie is entering her third year of medical school at Dalhousie University in Nova Scotia, Canada. She was first introduced to CHARGE Syndrome in 2006 as a student working with Dr. Kim Blake and has had a keen interest in it ever since. Her work on CHARGE Syndrome includes a paper on anesthesia management published with Dr. Blake, and a case study on the use of Botox to reduce salivary secretions in an infant with CHARGE Syndrome. After medical school Jillian is planning on pursuing a career in pediatrics.

Presentation Abstract: Why is anesthesia important? How many surgeries is my child likely to have? Why is it important to combine procedures with one anesthesia?

ANESTHESIA COMPLICATIONS IN CHARGE SYNDROME

Dr. Kim Blake, MB, MRCP, FRCPC

1st Professional CHARGE Syndrome Conference
July 24-26, 2009
Chicago, Illinois

kblake@dal.ca

There are Always Risks of Complications with Anaesthesia

- "…you sign a consent"
- Are you informed?

- Are Individuals with CHARGE Syndrome More at Risk?

Growing up With CHARGE Syndrome

Kennedy

Age 0-2 years: 7 surgeries

Age 2-4 years: 3 surgeries

Age 4-6 years: 6 surgeries

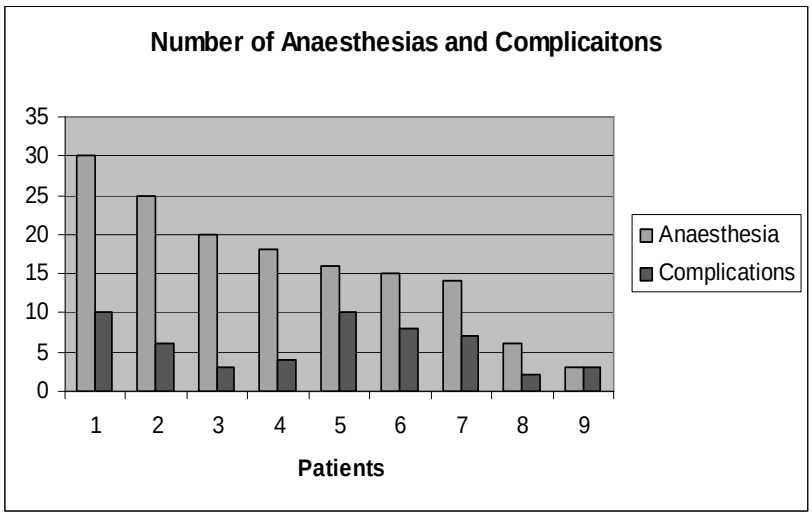
= many anesthesias

Postoperative Airway Events of Individuals with CHARGE Syndrome

- Detailed chart reviews on nine patients
  - Mean age 11.8 years (± 8.0)
  - 215 surgeries (average 22 per child)
  - 147 anesthesias (average 16 per child)
- Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Blake K, MacCuspie J, Hartshorne TS, Roy M, Davenport SLH, Corsten G.*International Journal of Pediatric Otorhinolaryngology*, *Vo. 73*, *February 2009*

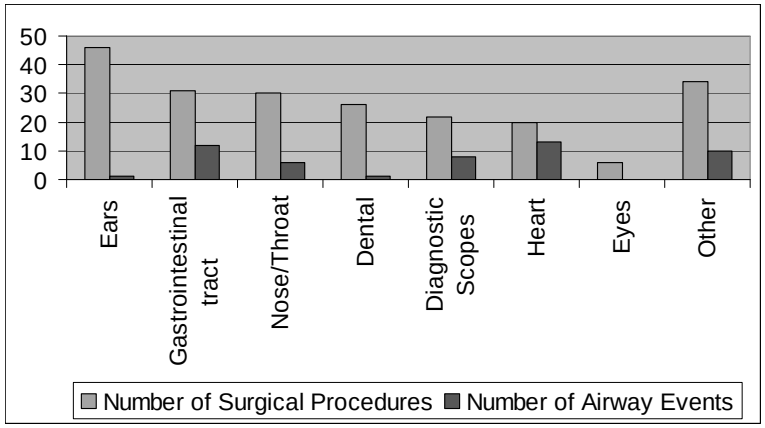
Results



35% (51/147) of anesthesias resulted in complications (>60% were major)

Results

Anesthesia related complications occurred most often with heart, diagnostic scopes (L/B/E) gastrointestinal tract procedures



Results

Number of surgical procedures per anesthetics with resulting postoperative airway events.

Number of surgical procedures	Number	Post-operative Events	Percent resulting in airway events
1	94	37	39% (n= 37/94)
2	36	8	22% (n= 8/36)
3+	15	5	33% (n= 5/15)

P=0.1 Combining multiple procedures under one anesthesia does not lead to an increase in post-operative events.

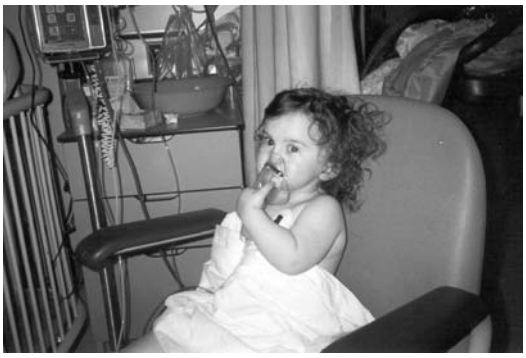
Results

Feeding procedures and rates of postoperative airway events.

	Number of Anesthesias	Airway Event	No Airway Event	Significance
G/J tube	82	36	46	Yes p=0.0092
No G/J tube	63	15	48	
Nissens fundoplication	79	33	46	Yes p=0.049
No Nissens fundoplication	66	18	48	

Having a G/J tube or Nissens fundoplication increases your child's risk of post-operative airway events

MacKenzie's Story



- 27 surgical procedures
- 18 anesthesias
- 4 complications
- Multiple ICU admissions
- Post tonsils/adenoids - improved

Discussion

- 35% of anesthesias resulted in post-operative complications
- Heart, diagnostic, and gastrointestinal tract procedures result in the most complications
- At least one complication occurred with every type of surgery except for eyes

Discussion

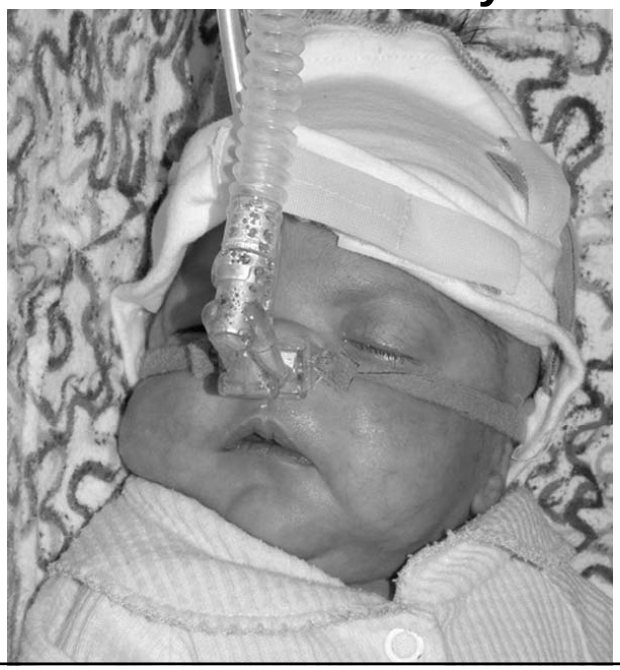
- High risk of complications in individuals with Nissen fundoplication or gastrotomy/jejunostomy tube
- Low risk with cleft palate
- What about individuals with CHD7 mutations who have mild clinical criteria?
  - Will they be at risk in the future?
  - Have they actually been challenged with surgeries?

Take Home Messages

CHARGE children are at high risk of post-operative anesthesia complications. Combining procedures during one anesthesia does not increase the risk of post-operative airway events.

The anesthesiologist needs to be aware that, even with simple procedures, the individual with CHARGE syndrome is at high risk of complications.

Frederick's Story

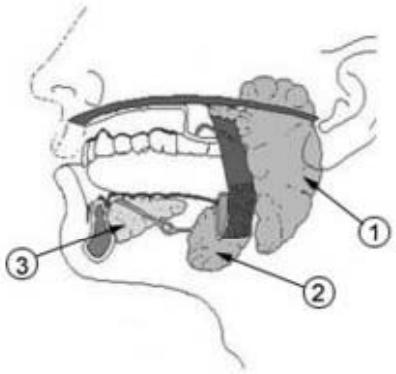


Freddy at 2 Months

- Difficulty with intubation
- ToF repair, vascular ring repair, PDA ligation
- Increased oral secretions
- Multiple attempts at extubation

Site of Botox Injections

1. Parotid glands
2. Submandibular glands
3. Sublingual glands



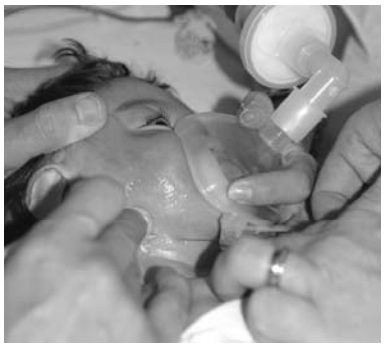
Botox 7.5 units was injected into salivary glands 1 and 2 on each side

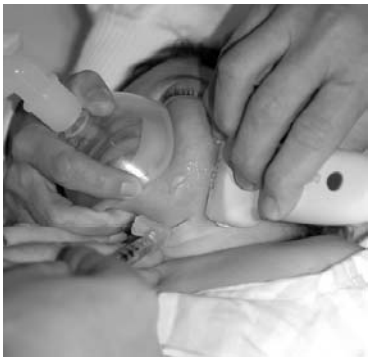
Freddy at 7 Months

- Aspiration pneumonia from oral secretions
- Gastroesophageal reflux
- Required ventilation

Botox Injection

Submandibular Gland Via Ultrasound and Parotid Gland by Palpation





10 Units/gland

Botox Injection

Prophylactic Use to Prevent Increase in Oral Secretions and Aspirations (4-5 monthly)

Waiting for picture

Thank you to all the Children and their Families

UK CHARGE Family Support Group Picnic 1991

EDUCATIONAL INFORMATION & FAMILY SUPPORT

SATURDAY
Breakout Session #8: 3:40 – 4:40 PM
Trillium Ballroom III, Conference Center

NANNY 411:

INFORMATION ON THE TOP TEN STRATEGIES FOR PROMOTING POSITIVE BEHAVIOR IN CHILDREN WITH CHARGE SYNDROME

Laurie S. Denno, MA, LMHC, BCBA
Deafblind Program, Perkins School for the Blind
&
Deanna Rothbauer
Parent of a 12-Year-Old Son Who Has CHARGE Syndrome

Presenter Information: : *Laurie Denno* is a practicing behavior analyst with over 30 years of experience working with children and adults with developmental disabilities, including over 20 years of experience working with deafblind children and young adults, many of whom have CHARGE syndrome. Laurie's main interests are in implementing proactive behavior treatment that stresses teaching socially appropriate behavior and independence to all learners and using positive behavior supports in a school-wide manner. Laurie is a Doctoral Candidate in Applied Behavior Analysis at Simmons College and will be doing her dissertation on assisting parents of children with CHARGE syndrome to speak effectively with a consulting psychiatrist about their children's behavioral difficulties.
ADDRESS: Perkins School for the Blind, 175 N. Beacon Street, Watertown, MA 02472; TEL: 1-617-972-7891;
EMAIL: Laurie.Denno@perkins.org

Deanna Rothbauer has a 12-year-old son who has CHARGE syndrome and challenging behaviors.

Presentation Abstract: Learn proactive approaches to prevent behavior difficulties. It is much easier to prevent problem behavior than to change it once the child has learned it. Behavior analysis is first and foremost a teaching strategy that stresses the interaction of the family, the school and the social environment with children and their behavior. Learn the principles from a behavior analyst and how to implement the principles from a parent who has done it.

Nanny 411:
Information on the Top Ten Strategies for Promoting Positive Behavior in Children with CHARGE Syndrome

Laurie S. Denno, MA, LMHC, BCBA
Behavior Analyst
laurie.denno@perkins.org
Deanna Rothbauer, Parent
9th Annual CHARGE Syndrome Conference
Bloomingdale, IL

Some General Information

- Most behavior is learned
- Behavior is learned by interacting with the environment
- Behavior is contextual, influenced by both what comes before it and what comes after it
- Every child has a unique context in which their behavior occurs, so every behavior treatment is unique for a specific child
- Children with CHARGE have learning challenges due to their sensory impairments. Pay attention to sensory issues

- Some behavior is not learned (tics, OCD, self stimulatory behavior) but still may be influenced by what comes before it and what comes after it

1. Be Consistent

- Have a discussion about what is important to your child and your family and make a plan. You can't work on everything at once.
- **All** family members should know and follow the plan
- Talk with school staff to increase consistency across settings
- Children with CHARGE need structure and predictability (many have anxiety disorders)
- Teaching new skills and learning new skills takes time, especially with hearing and vision impairments. Be patient!
- If you don't know what to do, walk away or give yourself some time and space to think

2. Be Positive

- Tell the child what “to do”
- Use positive feedback, praise and other reinforcement to increase behavior
- Rules for reinforcement
- 10:1 rule
- Use positive feedback to decrease undesirable behavior
- Teach alternatives
- Focus on strengths
- Tell the truth

3. Have Behavioral Expectations

- Use rules and reminders about acceptable behavior. Write the rules down. Not too many.
- Use positive feedback when rules are followed, even with reminders.
- Be respectful. Sometimes children “can” say no. Honor their request.
- Behavioral expectations should be the same for all children.
- Offer choices about what, when and how to do things.

Behavioral Expectations (cont.)

- A compromise can be a good solution to a stand off
- Not following the rules can teach a valuable lesson through natural consequences
- When you make exceptions, label the experience as “special”
- Your behavior influences the behavior of your child, so have your own “good behavior”
- Be an authoritative parent, not an authoritarian parent
- If you threaten with a negative consequence, follow through

4. Communication

- Use communication at the child’s functional level and in their primary mode
- Make sure you know the child’s vocabulary
- Give directions in short clear sentences in the child’s field of vision
- Tell the child what “to do”
- Recognize your child’s strengths and limitations
- Coordinate with the Speech and Language Pathologist

Communication (cont.)

- Anticipate the child's needs but expect her to ask for what she wants
- Teach the child to ask for a "break" or "relax time"
- Do not try to talk your child into "being good" or expect anyone else to do so
- Action speaks louder than words
- When you can't say anything nice, say nothing at all
- Use a "script" with your child when you are discussing less than desirable behavior

5. Implement a Schedule

- Follow a daily routine
- Make a schedule at the child's functional level (words, pictures, symbols, etc.): "to do" list
- Build in and teach changes, cancellations
- You can use a schedule with a behavior plan very easily
- The schedule should follow the regular flow of the day
- Save time for "down" or "alone" time
- Teach relaxation skills
- Teach play skills
- Plan for regular SI
- Have the child help around the house
- Have some fun

6. Child Should "Do" for Herself

- The child should participate in her own life
- ASAP have her help around the house, feed the dog, get the mail, clean up toys
- Start teaching ADLs at an early age
- Remember the "delay" in developmentally delayed
- Expect the child to "try", help only if necessary
- Praise "by yourself", "independent"
- Have the child spend some time alone with appropriate toys, objects or stimulation in a safe space

7. Things to Avoid

- ***Punishment*** suppresses behavior, but only when the punisher is present
- Punishment has some very bad side effects
- Punishment does not teach new behavior
- Reprimands should be used sparingly or not at all
- Giving up too soon
- Shopping for the magic solution
- "She'll grow out of it" syndrome
- She's fine at home (school)
- Making excuses
- Blaming others
- Blaming CHARGE
- Being a martyr (especially the moms)

8. Educate Yourself

- Read a book about Behavior Management, Applied Behavior Analysis or Functional Analysis of Behavior (See next page)
- Take a class at a college or through a parent group
- Network with other parents
- Talk to school staff at school about resources
- Call the CHARGE Syndrome Foundation for materials

Educate Yourself (cont.)

- The Good Kid Book, Howard Sloane
- Parents are Teachers, Wesley Becker
- Living with Children, Gerald Patterson
- Relaxation, J. Cautela and J. Groden
- Functional Behavior Assessment and Intervention in Schools, J. McDougal, S. Chafouleas and B. Waterman
- When Your Child Is Difficult, Mel Silberman

Educate Yourself (cont.)

- Solving Child Behavior Problems at Home and at School, E. Bleckman
- Applied Behavior Analysis, J.Cooper, T. Heron and W. Heward
- Freedom from Meltdowns: Dr. Thompson's Solutions for Children with Autism, Travis Thompson (good advice that also applies to CHARGE syndrome)

9. Talk To School Staff

- They may not know about CHARGE but they may know about behavior
- Don't try to solve all the problems for them, listen and brain storm with them
- Speak up when you have a problem, they can brain storm with you
- Everyone brings different skills to your child's education
- Expect high quality education
- Educate yourself about materials, homework, communication and the strategies being used

10. Get Professional Help

- Look for a Board Certified Behavior Analyst
- Use the website www.bacb.com
- **A good BCBA will not necessarily know about CHARGE**
- You can educate them
- Behavior is learned the same way by all children
- Ask at school
- Call a mental health center or a children's hospital
- Call other organizations who serve developmentally disabled children
- Check with a local university in special ed or psychology department

11. Call a Psychiatrist

- Find a child psychiatrist who specializes in developmental disabilities
- Outline the symptoms that are troublesome
- Organize your information
- Collect a little data
- Try to coordinate with the school
- Psychiatrists mostly prescribe medicine
- Medicine is used to treat symptoms
- Medicine is not to make your child lethargic, quiet or different
- Prescribing medicine is a trial and error process
- Psychiatry takes time and patience

A Note About Interfering Repetitive Behaviors

- Many children with CHARGE display interfering repetitive behaviors
- The behaviors appear to be neurological and are probably not learned, although they can be influenced by the environment
- Children are “driven” to complete the behaviors
- When you interrupt, interfere with or try to stop these behaviors, children may become aggressive or self injurious (SIB)
- It is the aggression and SIB that often cause problems at school and at home, but the repetitive interfering behaviors are the basis of the problem
- Aggression and SIB will respond much better to traditional behavior treatment strategies if the interfering behaviors also are treated directly

Thank you.

Sensory Information

SATURDAY
Breakout Session #8: 3:40 – 4:40 PM
Trillium Ballroom IV, Conference Center

VISION ISSUES FOR PEOPLE WITH CHARGE SYNDROME

David Brown

Education Specialist
California Deaf-Blind Services

Presenter Information: *David Brown* began his career in the United Kingdom but now provides educational services in California. He has written and spoken widely on CHARGE syndrome and deaf-blindness.. He is an extraordinarily perceptive observer of individuals with deaf-blindness. His descriptions of the challenges faced by specific individuals with multiple anomalies and the effects on individual behavior have provided a model for the study of CHARGE syndrome and have shaped the body of knowledge that has been amassed over the past two decades.

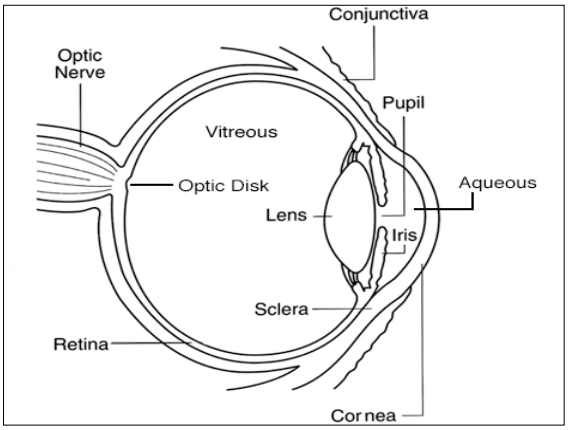
ADDRESS: 885 Corbett Avenue, San Francisco CA 94131
TEL: 415-405 7559; EMAIL: davidb@sfsu.edu

Presentation Abstract: Many of the anomalies found in CHARGE Syndrome carry significant implications for the development of functional vision skills. Some of these anomalies are specifically associated with eye defects, but many are not so are constantly overlooked or misunderstood as contributing to functional vision difficulties. This session will examine all these anomalies, their behavioral implications, and strategies for intervention.

Visual issues for people with CHARGE Syndrome

CHARGE Foundation Conference 2009
Chicago, Illinois

David Brown
Education Specialist
California Deaf-Blind Services
San Francisco State University

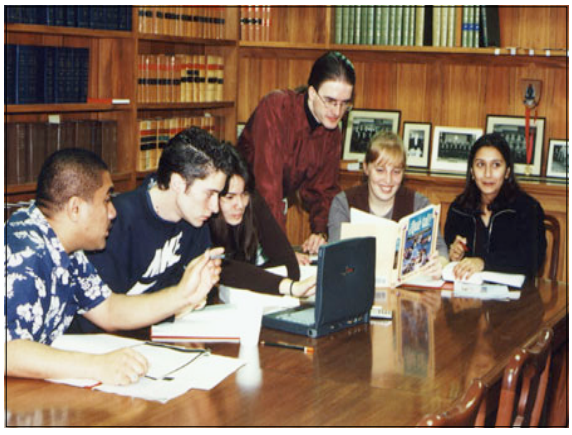


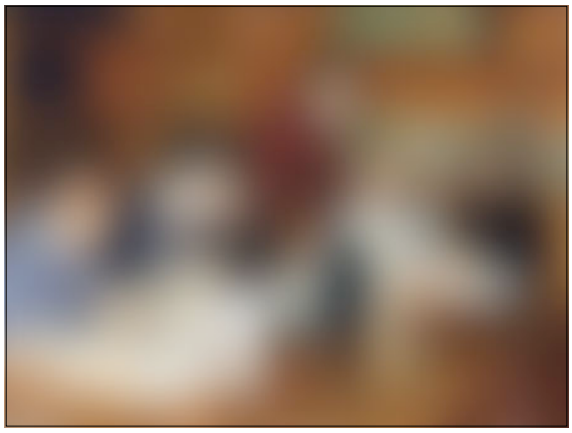
High Risk Signs of Vision Loss

- Atypical appearance of the eye
- Unusual eye movements
- Unusual gaze or head positions
- Absence of visually directed behaviors

The Five Types of Vision Loss

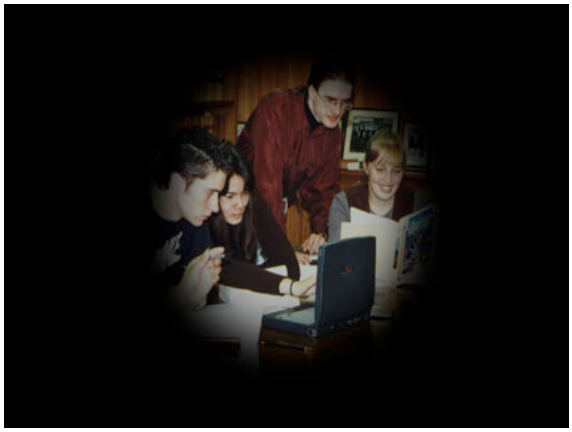
1 Loss of visual acuity: visual images appear blurred, visual detail is missing



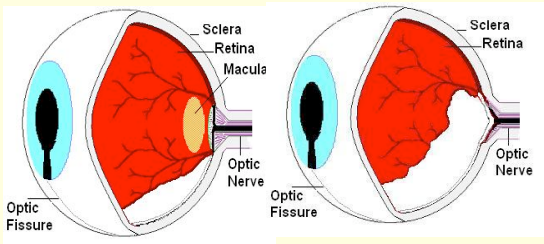


The Five Types of Vision Loss

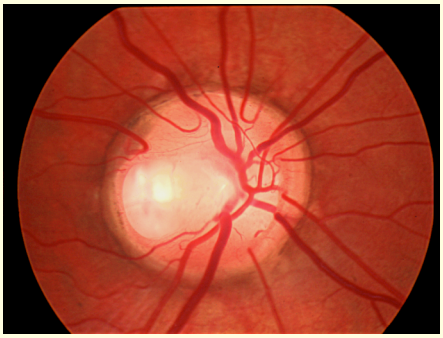
2 Loss of visual field: part (or parts) of the visual field is blurred or completely missing



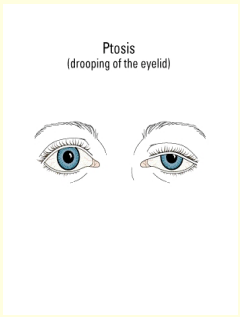
Retinal Coloboma



Optic Nerve Coloboma

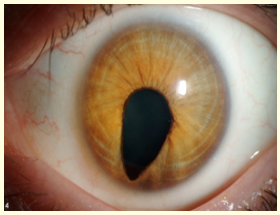


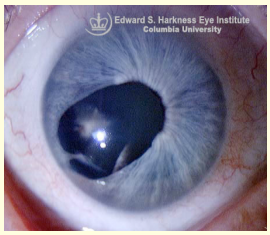
Facial palsy & ptosis



Facial palsy & ptosis

Iris coloboma



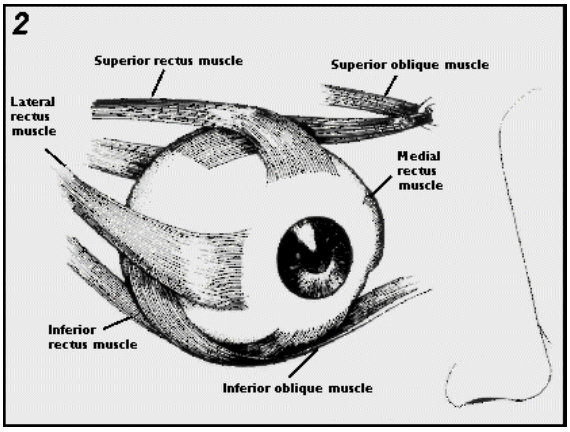


The Five Types of Vision Loss

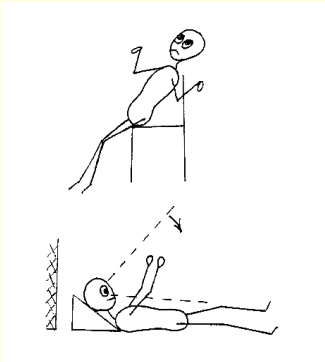
3 Loss of contrast sensitivity:the relative difference between the lightness and darkness of objects and their background is hard to perceive

The Five Types of Vision Loss

4 Loss of ocular motor control: the ability to control eye movements, particularly when performing visual tasks (eg fixating, tracking, scanning) is compromised



T Geniale (1991)



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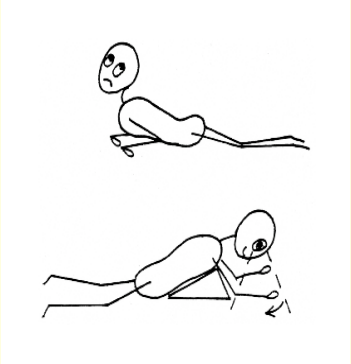
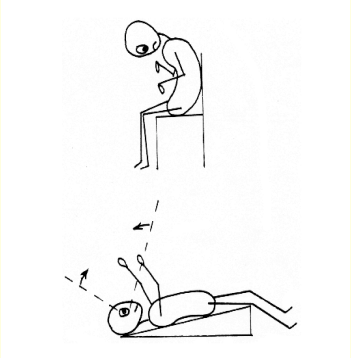
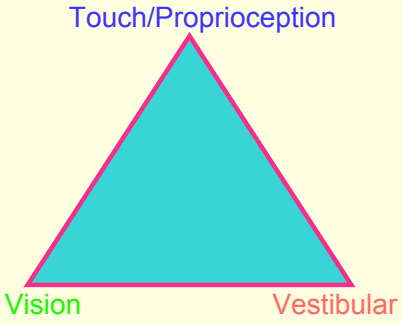


T Geniale (1991)

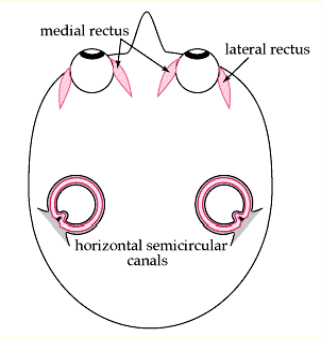


T Geniale (1991)

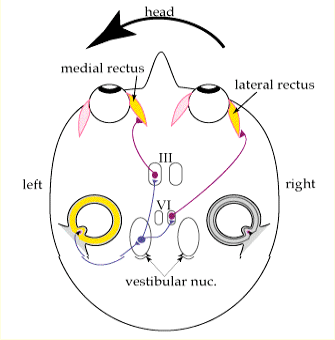
The Equilibrium Triad



The Vestibulo-ocular Reflex (1)



The Vestibulo-ocular Reflex (2)

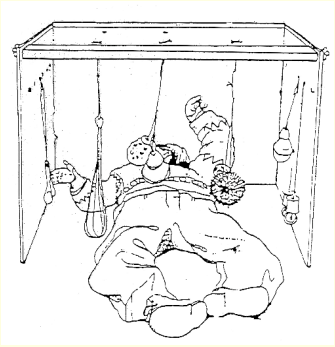


This is normal viewing posture…

…when you have no vestibular sense, upper visual field loss, poor tactile & proprioceptive perception, & low muscle tone.

The Little Room

The Be-Active Box



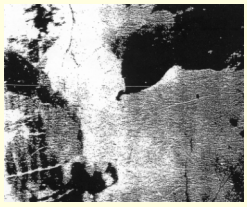
The Five Types of Vision Loss

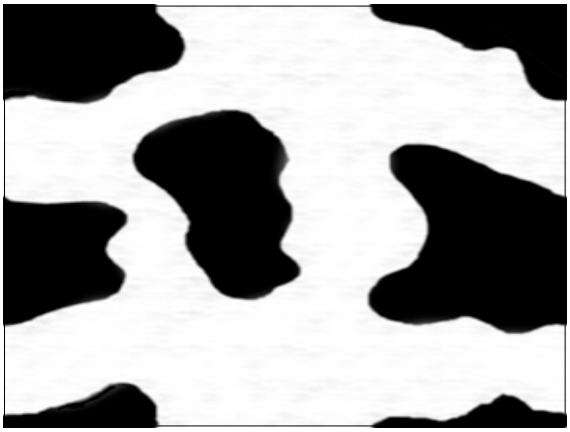
5 Loss of visual processing: the brain is unable to make correct sense of the information it is receiving through the eyes

Whose perception counts?

The brain, the organ that is responsible for your conscious experience, is an eternal prisoner in the solitary confinement of the skull…and must rely on information smuggled into it from the senses…the world is what your brain tells you it is, and the limitations of your senses set the boundaries of your conscious experience.

Coren, Porac & Ward "Sensation & Perception" (1984, p2)





We don't see with our eyes - we see with our brains

When you are assessing vision - don't think 'eyes', think 'child'

We don't hear with our ears - we hear with our brains

When you are assessing hearing - don't think 'ears', think 'child'

Natalie Barraga (1976)

Visual functioning is related in part to the condition of the eye. More explicitly, visual functioning is determined by the *experiences*, *motivations*, *needs* and *expectations* of each individual in relation to whatever visual capacity is available to *satisfy* *curiosity* and accomplish activities for *personal* *satisfaction*.

Vision Issues for People with CHARGE Syndrome

David Brown, CHARGE Foundation Conference 2009

In this presentation I plan to focus on the multi-layered complexity of the vision issues faced by many people with CHARGE Syndrome, with a special emphasis on early childhood when good visual motivation and good visual behaviors can be established.

There is a common tendency to assume that the actual condition of the eyes, particularly any eye defect that might be present, is what determines functional vision skills. While it is, of course, important to know about the condition of the eyes, and to seek appropriate help from medical specialists to evaluate this and to intervene where possible, there are also many other factors that need to be understood, evaluated, and worked on.

For individuals with CHARGE Syndrome the following would be a helpful list to remember:

- The eyes, and ocular defects
- The nerve pathways that connect the eyes to the brain
- The brain itself
- Muscle tone, and the obstacles that abnormal muscle tone present to effective use of vision
- Broader issues of postural control, and energy levels and fatigue
- Distractibility and the place of vision in the individual's hierarchy of the senses (ie. is vision an important resource for this person, or does it seem very low priority for them?)
- Expectations, previous experience, and motivation of the individual (and of the people around them)
- Environmental factors such as visual clutter, physical placement of things in relation to the individual, lighting levels, other distractions that might compete for the individual's attention

We have to remember that we don't see with our eyes, we see with our brains – all that the eyes can do is collect visual information for our brains to 'see' (ie. it is the brain, not the eyes, that has to perceive, to interpret and recognize, and to make decisions based on what the eyes are showing it). We also need to remember that in any group of people with visual impairments the individual with the best functional vision skills might not be the individual with the most vision!