



**Friday, July 31, 2015
Breakout Session #1: 10:45-11:45am
Schaumburg A-B**

Planning for the Future Part 1

Brian and Benjamin Rubin

Rubin Law

Presenter Information:

Brian Rubin has been a practicing attorney since 1976 and is the parent of three children, one of whom, Mitchell, has Autism. Brian's law practice, since 1982, when Mitchell was one year old, has been dedicated to serving the legal and future planning needs of his fellow families of children and adults with intellectual disabilities, developmental disabilities, and/or mental illness. 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.

Benjamin (Benji) Rubin, Brian's youngest son, Mitchell's "little/big" brother, graduated from the University of Illinois College of Law, Magna Cum Laude, received his undergraduate degree from Northwestern University, and currently is pursuing his Graduate Law Degree, an LLM (Tax) at Northwestern University. Having Mitchell as a brother profoundly shaped who Benji is today, and thus the type of law he chose to practice. His personal experiences as a sibling offer a unique perspective into the responsibilities that come with caring for a sibling with special needs. Now, as an adult, those sometimes present and future responsibilities he will share with his older sister regarding his brother's care are a concern that he shares with all brothers and sisters of individuals with special needs.

Presentation Abstract:

Government Benefits: The who, what, where, when, why and how of Government Benefits, including:

- SSI & SSDI
- Medicaid & Medicare

Special Needs Estate & Future Planning: The who, what, where, when, why and how of:

- Types of and appropriate uses of Special Needs Trusts;
- The ABLE Act Account;
- Gifting by grandparents & extended family members;
- The impact of divorce & child support on government benefits;
- Determining the appropriate amount to leave;
- Guardianships & Alternatives;
- Providing guidance, education and information for the "Future Team";
- Other related topics



**Friday, July 31, 2015
Breakout Session #2: 10:45-11:45am
Schaumburg C-D**

**Can't Stomach This: A Discussion on
Gut Motility in CHARGE Syndrome
Including Data from Recent Research**

**Kim Blake, MD,
Department of Pediatrics,
Dalhousie University,
& IWK Health Centre, Halifax NS
Shelby Steele
Meghan Macdonald**

Presenter Information:

Dr. Kim Blake's research in CHARGE Syndrome has been ongoing for 35 years. She has completed research in many clinical aspects of CHARGE Syndrome including; post-operative airway events, sleep apnea, bone health, cranial nerve anomalies and feeding disorders. Her clinical understanding of CHARGE Syndrome has given her the ability to ask research questions that can be investigated with the Zebra Fish model. Her goal is to understand more about the cranial nerves associated with CHARGE Syndrome with respect to the cardiovascular and gastrointestinal systems.

Presentation Abstract:

A common and long term issue for CHARGE syndrome individuals is eating and gastrointestinal motility problems. This presentation will outline the common gastrointestinal (GI) problems; identify professionals and treatments that are available; and present the clinical and basic science research that is hot off the press.



**Friday, July 31, 2015
Breakout Session #3: 10:45-11:45am
Schaumburg West**

Father's Forum

**Timothy S. Hartshorne, Ph.D.
Central Michigan University, Department of
Psychology**

Presenter Information:

Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project, which provides support to children who are deafblind in Michigan. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. He has been awarded the Star in CHARGE by the CHARGE Syndrome Foundation. He is first editor of the book *CHARGE Syndrome*.

Presentation Abstract:

CHARGE affects every member of the family. This meeting is only for fathers who have a child with CHARGE syndrome. It is their opportunity to have an open discussion and share experiences with other fathers.



**Friday, July 31, 2015
Breakout Session #4: 10:45-11:45am
Schaumburg East**

***Looking at the world through rose-colored glasses:
Approaches to positive assessment***

**David Brown & Julie Maier
California Deaf-Blind Services**

Presenter Information:

David entered the specialist area of deaf-blindness in 1983 and has become a leading and well-published practitioner in the field particularly with reference to CHARGE syndrome. In his roles as Head of the Family Education and Advisory Service of SENSE (the National Deafblind Association) in the UK (1983-2000), and as Educational Specialist for California Deaf-Blind Services (since August 2000), he has worked with over 150 children and young people with CHARGE. For 25 years David has been writing training manuals for universities and other training organizations, and publishing articles on a variety of topics in journals and magazines such as "Talking Sense", "Deafblind Education", "Eye Contact", "Special Children", "Deaf-Blind Perspectives", "reSources", "Deafblind International Review", "The American Journal of Medical Genetics", and "Child: Care, Health and Development". His writings have been translated into French, Italian, Spanish, Portuguese, Danish, Norwegian, Swedish, Finnish, German, Greek, Japanese, and Russian. David has contributed to staff training courses in 14 countries, and he has presented at CHARGE Syndrome Conferences in England, the USA, Australia, New Zealand, France, Norway, Sweden, Denmark, Switzerland, and Germany. David has also worked as a consultant with the state deaf-blind projects in 18 states of the USA. David has a credential in the Education of Students with Multi Sensory Impairments from the University of Birmingham, an Honorary Doctorate of Science from Central Michigan University for his research work with children with deaf-blindness, and in 2013 was awarded the Deafblind International 'Lifetime Achievement Award' for "innovative and visionary work with people with deafblindness".

Presentation Abstract:

This presentation is based upon two articles that Julie and David wrote for the CDBS newsletter in 2014. We will examine and discuss the perspectives and assumptions educators, medical professionals, and other related service providers often employ when viewing students with CHARGE Syndrome. These perspectives and assumptions then influence the direction taken during assessment and planning development of educational goals and programs for individuals with CHARGE Syndrome. We will share a more effective, child-centered approach.

California Deaf-Blind Services



The contents of this PowerPoint presentation were developed under a grant from the US Department of Education, #H326T130031. However, those contents do not necessarily represent the policy of the US Department of Education, and you should not assume endorsement by the Federal Government.
Project Officer, Jo Ann McCann.



“Looking at the world through rose-colored glasses” Approaches to positive assessment

David Brown & Julie Maier
California Deaf-blind Services
CHARGE Syndrome Foundation Conference
Chicago July 2015



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The uniqueness (and complexity) of CHARGE syndrome

The changing nature of the population of children with CHARGE syndrome

David Brown - Am.J.Med.Gen. 2005

“Children with CHARGE syndrome are truly “multi-sensory impaired”, having difficulties not only with vision and hearing but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell, as well as problems with breathing and swallowing, eating and drinking, digestion, and temperature control.”

CHARGE - the most ‘multi sensory impaired’ of all syndromes

Problems with the perception of:

- Vision
- Hearing
- Touch
- Proprioception
- Temperature
- Pain
- Vestibular
- Smell
- Taste

Why do people assess children with CHARGE syndrome?

Challenges to the Assessment Process

- CHARGE presents a very diverse and complex population of learners
- CHARGE presents a wide variety of idiosyncratic behaviors
- People doing assessments usually only know one type of assessment process
- There are limited resources and assessment tools available
- People doing the assessment often forget “The reason why” of assessment

A CAPACITY BUILDING FRAMEWORK

- Using a CAPACITY model allows you to look for and recognize the unique characteristics, skills, talents, and interests of a person.
- Is a way of understanding an individual that assumes their competence and values their contribution and participation
- Allows you to recognize POTENTIAL and POSSIBILITIES.
- It is INDIVIDUALIZED.

A DEFICIT-FINDING FRAMEWORK

- In contrast, a DEFICIT model focuses on labels, limitations, barriers, and remediation.
- It does NOT recognize POTENTIAL and instead focuses on “fixing”, or “getting ready”, or “meeting specific criteria”.
- Views support needs as evidence of child’s problems.
- It is LIMITING.

Why use a capacity-building framework?

- Actively re-positions students
- Removes “problems” from students and shifts them to educational professionals
- Leads to more meaningful and student-focused educational decision-making and learning opportunities

Presuming competence

“Presuming competence is the underlying assumption that even those individuals who behave, move, communicate, learn, and interact with others in different ways share the same human desires we all have for personal growth, community membership, and fulfilling relationships and work. Often these differences disguise the competencies of these individuals due to our society’s narrow view of competency which does not allow us to recognize the atypical or unusual skills, abilities, behaviors, and contributions demonstrated by individuals with disabilities as competent or valuable.”

~J. Maier (2014)



Presume Competence

"Let's consider the first few weeks Annie Sullivan spent with the young Helen Keller. What if she had only recognized a "wild child" who held no desire to communicate with others or potential to learn? What if she believed Helen was only a spoiled child with a bad temperament that would be permanently locked inside herself because she had lost her vision and hearing as a baby? Fortunately for Helen and her family and society as a whole, Ms. Sullivan took the approach of "presuming competence" and understood that Helen's different behaviors and actions were certainly a form of communication and an indication of a deep desire know, learn, and grow."

~J. Maier (2014)



Two perspectives

Deficit model description	Capacity-building model
<i>Assessment of environmental signs from the PCI Environmental Print series:</i> "While Juan was able to expressively identify many of the environmental signs by category, it is the opinion of the examiner that Juan's responses were often not specific to the function of the sign in a true context. Examples include Juan stating, "What Mr. P does." for recycling, as well as "You might fall" for slippery when wet sign. This indicates that Juan is able to rely on visual cues yet he may have difficulty when found within the natural environment, such as buildings and outdoor settings."	<ul style="list-style-type: none">•Juan is a visual learner with a good memory.•Juan can make inferences and associations when presented with information. (e.g. Recycling symbol associated with an activity his teacher performs)•Juan can predict events or problems and assess risk and act accordingly (e.g. be careful when you see "slippery when wet" sign and walk around it or walk carefully).•Juan would benefit opportunities in natural settings to learn to identify, read, and use environmental print.

The criterion of the least dangerous assumption...

"The criterion of the least dangerous assumption holds that in the absence of conclusive data, educational decisions ought to be based on assumptions, which, if incorrect, will have the least dangerous effect on the likelihood that students will be able to functional independently as adults."

~Anne Donnelan (1984)

Two different assumptions

Case #1	Case #2
A child with CHARGE Syndrome who significant vision loss and profound hearing loss is provided with an object calendar and taught signed communication, basic literacy, functional math skills, and the use of a cane from early school years on. She is provided with daily integration opportunities to with same age peers who are taught how to communicate and engage with her. She receives support from an intervener and her team regularly meets to discuss her progress and support needs.	Now consider that same child assigned all day in special education class and receiving primarily custodial care and no academic instruction or communication system because it was assumed that she is “not really aware of anything or anyone around her” and an academic curriculum will be too difficult for her due to her global delays. She must be led by hand to different activities in class and school by whichever staff is working with her at that time because she cannot see and hear and it isn’t safe for her to use a cane yet. She plays alone by choice and because she is just not interested in her peers.

Which assumption is more dangerous to follow?

Same view, different perspective

“This lens is not limited to identifying strengths, but rather **shifts the focus**. The view is of the same student, yet the perspective is very different. Educators who use a capacity lens **look beyond what the student with disabilities is “doing”** by describing the different or missing skills or behaviors, and instead **consider why the student is or isn’t doing something**, and will then identify and **consider systems of support** that could enhance and develop the student’s capabilities. “These educators also understand that the “problems” that challenge them the most are actually evidence of student capability.”

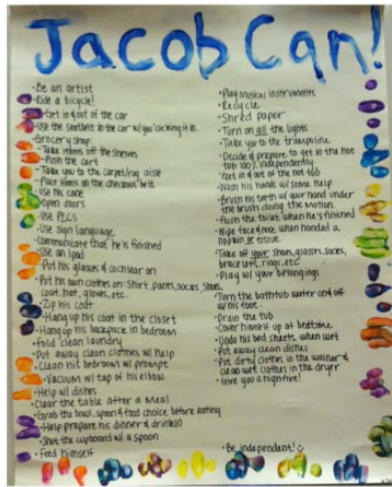
~J. Maier (2014)

Support Needs

- A deficit-framing view judges the type and intensity of supports as another way to acknowledge the limitations and deficits of the individuals.
- What if supports were viewed as a means to expand the individual’s participation and contribution and self-determination?
- Why is a visual or tactile checklist used to complete a job or task often viewed as, “This person cannot perform this job without help,” instead of “Wow, with this checklist this person can independently complete this job.”
- We all rely on supports in our lives, but when the need for supports is viewed as a deficit or limitation, then that colors a person’s view and perspective of the capacity and contributions of another.

~J. Maier (2014)

From 2013
International
CHARGE
SYNDROME
Conference
Poster Session



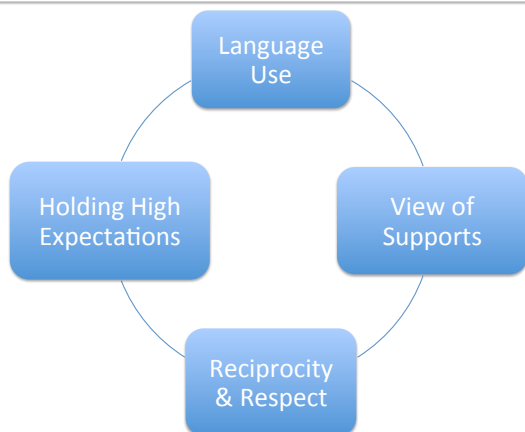
Two views of Jacob

What Jacob CAN do...	What Jacob CANNOT do...
<ul style="list-style-type: none"> • Ride a bike and can buckle seatbelt • Use sign language and picture symbols to communicate • Put his glasses and cochlear on • With some help c wash hands and brush teeth • Push a grocery cart & put items on the checkout conveyer belt • Shred paper • Put away clean dishes • Put dirty clothes in the washer and wet clothes in the dryer • Choose to use hot tub & get in and out independently 	<ul style="list-style-type: none"> • Cannot drive a car • Cannot speak • Is deaf-blind • Cannot wash his hands or brush his teeth independently • Cannot read a recipe • Cannot cook a meal • Cannot grocery shop, wash dishes, or do laundry independently • Rips paper, so he he should not have paper • Cannot live alone

The IMPORTANT question here is...

Which list would be more helpful in helping Jacob live a life that will be fulfilling for him and bring him happiness?

Capacity-building Assessment Practices



Our view of assessment.....

- Is unusual!
- Is positive
- Looks at positive skills & achievements
- Looks at learning styles
- Looks at preferences & interests
- Looks at the whole child
- Credits the child with intelligence

Our view of assessment (2).....

- Seeks to improve our understanding of the child
- Seeks to help us to build a positive relationship with the child
- Seeks to help us to know what to teach and how best to teach it
- Seeks to give us a clear focus for measuring progress

Jan van Dijk (1966)

“In the educational atmosphere I describe, the child holds the central position, the teacher ‘follows’ the child and, when the child responds, the teacher is present to answer the child’s request”

Van Dijk & Nelson
“Principles of Assessment” (2001)

- Make the child at ease
- Determine the child’s bio-behavioral state
- Determine the child’s interest
- Follow the child’s interest

The Van Dijk Approach to Assessment

- Child-guided
- Fluid
- Looks at the processes children with multiple disabilities, including sensory impairments, use to learn & to develop
- Assessment is summarized in terms of strengths and next steps for intervention

Areas of the Van Dijk Assessment Framework

- Ability to maintain & modulate state
- Preferred learning channels
- Ability to learn, remember & anticipate routines
- Accommodation of new experiences with existing schemes
- Problem solving approaches
- Ability to form social attachments and interact
- Communication modes

Assessment Questions

D Brown, "Follow the Child" (2001)

- How do you feel?
- What do you like?
- What do you want?
- What do you do?

D Brown 'Follow the Child' (2001)

- Consult those who know the child better than you do
- Identify the child's motivators
- It's okay to match different sensory inputs
- Relax/arouse the child
- Position the child to facilitate functional skills
- Allow the TIME necessary for the loop of sensory perception, interpretation, and response

CHARGE Communication

*communication with one's own body

*communication with one's immediate environment

*communication with the wider world

Assessment should be regarded as
a Process not a Product

◆ Trial and Error (child-guided, fluid)

◆ Always a work in progress

◆ Questions not answers

◆ Why?

References for “Looking at the World Through Rose Colored Glasses” presentation
12th International CHARGE Syndrome Foundation Conference,
July 31st 2015

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Friday, July 31, 2015
Breakout Session #5: 10:45-11:45am
Schaumburg E-F

***Breathing and Posture: More Linked
than you Think!***

Mary Massery, PT, DPT, DSc

Presenter Information:

Dr. Massery received her BS in Physical Therapy from Northwestern University in 1977, her DPT from the University of the Pacific in 2004 and her DSc from Rocky Mountain University in 2011. Her publications and interests focus on linking motor behaviors to breathing and/or postural mechanics in both pediatric and adult patient populations. Dr. Massery has been invited to give over 800 professional presentations in 49 US states, 9 Canadian provinces, and 14 countries worldwide. Mary received the **American Physical Therapy Association's** highest clinical award, *The Florence Kendall Practice Award*. She continues to maintain a private practice in Chicago, specializing in ventilation and postural dysfunction for children with complex medical conditions like CHARGE.

Presentation Abstract:

Each child with CHARGE presents with unique physical deficits that can progress to significant postural deficits by adulthood. This session is designed to educate families about the link between breathing mechanics, postural alignment, movement strategies, and balance deficits, and to demonstrate how these interactions may adversely affect the child's postural maturation. The focus is on educating the families about the big picture, the long haul, to empower them about the value of periodic physical therapy and other supportive services throughout childhood and into adulthood, rather than just during Early Intervention.

12th International CHARGE syndrome Conference
Chicago Friday, July 31st, 2015
10:45 – 11:45 am

BREATHING & POSTURE: MORE LINKED THAN YOU THINK!

Mary Massery, PT, DPT, DSc
Massery Physical Therapy, Glenview, IL
www.MasseryPT.com

CHARGE

- Rare syndrome 1/10,000 births
- Complex medical conditions, many which can be life threatening
- However, each child is unique in his/her presentation making a one-size fits all approach to PT impossible



Cole and his family

CHARGE

- My goals today as a PT...
- Educate families about short term & long term postural issues that can adversely affect the child's physical development, health and participation.
- Suggest how PT may help minimize or prevent postural and breathing problems throughout the lifespan.



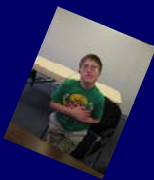
Cole and his family

Research on Postural Interventions Secondary to CHARGE

■ **NONE!** (PubMed)

My direct experience with CHARGE

- 6 patients: 1 girl, 5 boys
- Young: 15 months – 13 y/o
- Amazing families! I love working with you!



Major Postural Trends observed with this small population

- Shoulders: Protracted (sloped) & elevated (short neck) 4/6
- Airway: Tracheostomies 4/6
- Spine: Thoracic kyphosis 4/6, lumbar kyphosis 1/6. No scoliosis (yet).
- Scars: 6/6. Severe restriction 1/6.



George

Major Postural Trends observed with this small population

- Why?
 - Genetics?
 - Compensations?
 - Vestibular deficits (balance)?
 - Visual deficits (protection)?
 - Increased work of breathing (survival, increased accessory recruitment)?
 - Trunk muscles low tone (postural instability)?
 - Scar restrictions (limiting ROM)?
 - Or all of the above?!!!!



Balance

- 3 systems of feedback to the brain keep us upright and stable:
 - Visual
 - Vestibular
 - Proprioception
- Children with CHARGE often only have proprioception to give them postural feedback.
 - "Heavy Feet" – children may stomp and clomp to give themselves more feedback.

Major Postural Trends observed with this small population

- All these impairments restrict mobility and balance, hence participation and potentially health.





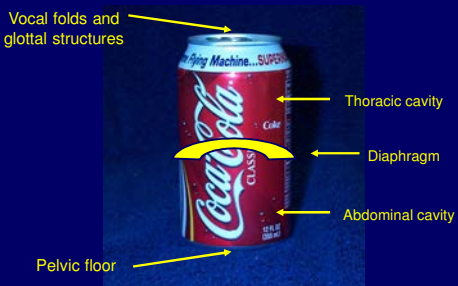
Photo by Lois Greenfield

A Postural Control Model Using a Soda-Pop Can

- Closed System
- Positive pressure from internal can is > atmospheric pressure
- Pressure creates functional strength to an otherwise weak external structure



A Postural Control Model Using a Soda-Pop Can



Vocal folds and glottal structures

Thoracic cavity

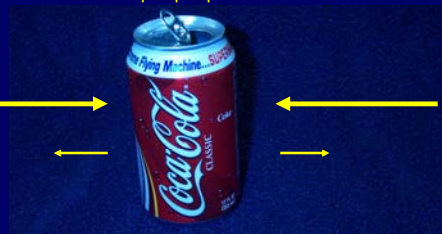
Diaphragm

Abdominal cavity

Pelvic floor

Positive Pressure Compromised

Pop-top Opened



External Pressure > Internal Pressure

Potential Results: Collapse of the Skeleton and Internal Organs



PT's Role

- Identify postural deficits and potential limitations to movement and health (especially breathing mechanics)
- Educate families so they can become the primary member of the PT team in the future!
- Home programs are critical to success!



Postural Changes Over Time



Cole: Trach Scar Revision




Cole: Trach Scar Revision



What can PTs and Families do?

- Assume there are motor consequences for all persistent physiologic impairments! ☺
- Teamwork!
 - Recognize the conflict.
 - Treat the breathing / postural control impairment.
 - Refer to the medical team for further testing and medical management when appropriate.
 - Together, we can help our children meet their physical and physiologic maturation needs, achieving greater adult health and participation goals.

It's all about the can!



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Idea from Kathy Martin



**Friday, July 31, 2015
Breakout Session #6: 10:45-11:45am
Schaumburg G**

Grandparents Forum

**Pamela Ryan
Perkins**

Presenter Information:

Pam has been at Perkins School for the Blind for more than three decades. As the School Psychologist in the Deafblind Program at Perkins as well as her other positions within the school, she has worked with and learned from countless students with CHARGE and their families. Pam is also on the board of directors of the CHARGE Syndrome Foundation.

Presentation Abstract:

CHARGE syndrome affects every member of the family. This meeting is only for grandparents who have a grandchild with CHARGE. It is their opportunity to have an open discussion and share experiences with other grandparents who have had to deal with the issues associated with CHARGE syndrome.



**Friday, July 31, 2015
Breakout Session #7: 10:45-11:45am
Schaumburg H**

**Young Adults with CHARGE Only:
Share experiences, concerns, and ask
questions**

**Kasee Stratton, Ph.D., NCSP & Dan Gadke,
Ph.D., NCSP, BCBA
Mississippi State University, Assistant
Professor/Licensed Psychologist**

Presenter Information:

Dr. Kasee Stratton and Dr. Daniel Gadke are assistant professors of school psychology at Mississippi State University and licensed psychologists who specialize in CHARGE and other developmental disabilities. Dr. Stratton currently runs the Bulldog CHARGE Syndrome Research Lab at MSU and Dr. Gadke is the director of the Autism and Developmental Disabilities Clinic at MSU. Dr. Stratton and Dr. Gadke enjoy meeting with young adults and recently held a young adult group at the CHARGE Syndrome Australasian Conference in 2014 and received positive feedback from participants. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome*, the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Gadke is published and well versed in the area of developmental disabilities. Dr. Gadke and Stratton completed their post-doctoral training at Johns Hopkins School of Medicine and the Kennedy Krieger Institute.

Presentation Abstract:

This time is devoted to young adults only--ages 16 and up. We will provide a safe and confidential space for young adults with CHARGE to ask questions, discuss concerns, and share with each other. Previous groups have discussed sexuality, getting and keeping jobs, having relationships and friendships, planning for independent living, and so much more. No question is off limits.



**Friday, July 31, 2015
Breakout Session #8: 1:00-2:00pm
Schaumburg A-B**

Planning for the Future Part 2

Brian and Benjamin Rubin

Rubin Law

Presenter Information:

Brian Rubin has been a practicing attorney since 1976 and is the parent of three children, one of whom, Mitchell, has Autism. Brian's law practice, since 1982, when Mitchell was one year old, has been dedicated to serving the legal and future planning needs of his fellow families of children and adults with intellectual disabilities, developmental disabilities, and/or mental illness. 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.

Benjamin (Benji) Rubin, Brian's youngest son, Mitchell's "little/big" brother, graduated from the University of Illinois College of Law, Magna Cum Laude, received his undergraduate degree from Northwestern University, and currently is pursuing his Graduate Law Degree, an LLM (Tax) at Northwestern University. Having Mitchell as a brother profoundly shaped who Benji is today, and thus the type of law he chose to practice. His personal experiences as a sibling offer a unique perspective into the responsibilities that come with caring for a sibling with special needs. Now, as an adult, those sometimes present and future responsibilities he will share with his older sister regarding his brother's care are a concern that he shares with all brothers and sisters of individuals with special needs.

Presentation Abstract:

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- Medicaid & Medicare

Special Needs Estate & Future Planning: The who, what, where, when, why and how of:

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- Determining the appropriate amount to leave;
- Guardianships & Alternatives;
- Providing guidance, education and information for the "Future Team";
- Other related topics



**Friday, July 31, 2015
Breakout Session #9: 1:00- 2:00pm
Schaumburg C-D**

Mothers and Fathers and Siblings, Oh My!

**Timothy S. Hartshorne, Ph.D.
Central Michigan University, Department of
Psychology**

Presenter Information:

Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project, which provides support to children who are deafblind in Michigan. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. He has been awarded the Star in CHARGE by the CHARGE Syndrome Foundation. He is first editor of the book *CHARGE Syndrome*.

Presentation Abstract:

A child with CHARGE syndrome clearly impacts all the family members, but little is known about exactly how. This presentation reports on research conducted by the CHARGE lab on the experience of mothers, fathers, and siblings of children with CHARGE, and then discusses the findings in the context of family systems. It concludes with some thoughts on family wellbeing.



Mothers, and Fathers, and Siblings, Oh My!

The Family in CHARGE

Tim Hartshorne

Mothers



Mothers

- Most research is on mothers
- Burden of care giving may fall on mothers most of all
- Parental roles may be more well-defined
- Mothers may experience the most guilt

- Direct care
- Medical issues
- Education issues
- Transition issues
- Family management
- Need to protect
- May feel at fault

“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 View in 1973

The impact of a handicapped child on a family is never negligible, usually damaging, and sometimes catastrophic. A few families with great spiritual strength may be bound more firmly together by the experience, but in most, the stresses imposed far outweigh any benefit. Mitchell, 1973.

Negatives?

- I don't know enough to do this
- There is no support out there
- Someone will need to stay home
- Getting the right medical help
- Fear for my child
- Guilt that I am not doing enough
- I may spend the rest of my life doing this
- There is not enough time in the day
- There is not enough time for spouse and other children

I can't cope!!!

The View Today

Families of children with disabilities report positive perceptions in addition to negative perceptions and stress, and there is some data to suggest that positive perceptions are common, and that they assist the family in adapting to having a child with disabilities.

Hastings & Taunt, 2002

Positives?

- Pleasure/Satisfaction in providing care
- Child is a joy
- Sense of accomplishment
- Strengthened family
- New sense of purpose
- New skills, abilities, even career
- Have become a better person
- Increased personal strength/confidence
- Social networks
- Increased spirituality
- Changed perspective on life
- Making the most of every day

Fathers



Fathers

- Often the forgotten parent
- Researchers have generally ignored
- Participation in child care related to higher marital satisfaction – also a way to cope
- May have an intense reaction to the diagnosis
- Child raises question of life's meaning
- May feel cut off from social support
- Negative view of professionals
- Acknowledge personal growth

Why do we know so little

- Fathers have traditionally delivered discipline and life lessons, not care
- Assumed to have the same views, thoughts, concerns and stress as mothers



Shantell Johnson study

- 93 fathers

United States	69
Australia	6
United Kingdom	5
Canada	4
New Zealand	4
Germany	4
Ukraine	1
Costa Rica	1

Single	5
Married	82
Divorced	6

21-31	5
32-41	31
42-51	42
52-61	11
62-71	6

Some high school	1
High School	13
Some college	20
College graduate	34
Graduate course work	2
Graduate degree	23

Fathers' Reactions

- Fathers' reactions to birth and diagnosis

Scared, concerned, anxious, fearful	66
Sad, heartbroken, lost, pessimistic	49
Shocked, stunned, offended, confused	41
Overwhelmed, challenged, disappointed, helpless	29
Love, pride, appreciation, dedication	23
Anger, resentful, bitter, rage	17

- 68.8% did not know mutation is in sperm

Impact on career

- No change in leisure time
- Half took only some of allotted vacation
- Career significantly less central to identity since birth of child with CHARGE
- Birth led to career or job change for 29%
- Most fathers felt job was “father friendly”
- Importance of career has not change

Impact on Friendships

- Quality of friendship is lower since birth
- They have fewer friends since the birth
- Were less satisfied with the number of friendships since the birth
- Did not rate “time with the guys” very highly
- In general did not want more friendships nor did they miss those they had before

Dads' Active Disease Support Scale

- Top rated tasks performed
 - Recognize and respond to child's symptoms
 - Give up sleep if child's condition needs it
 - Take care of child so spouse can go out
 - Come to child's medical appointments
 - Reward/praise child for cooperating with treatment
- Overall score very similar to other conditions

Dad's Active Disease Support Scale

- Top five tasks perceived to help family
 - Take care of child so spouse can go out
 - Take over household tasks giving spouse more time to attend to medical issues
 - Engage in leisure activities with child(ren)
 - Come to child's medical appointments
 - Recognize and respond to child's symptoms
- Overall scores much lower than other conditions

Meaning in Life

- How happy are you taking all things together?
 - Significantly less happy
- All things considered how satisfied are you with your life as a whole these days?
 - Significantly less satisfied
- How often, if at all, do you think about the meaning and purpose of life?
 - Significantly less often

Feel a bit helpless?

- Fathers are fixers
 - CHARGE is hard to fix (may not know best)
- CHARGE is so complicated and involved, fathers may not feel that they are able to contribute enough that is useful
- Family may need more attention
 - Career still important, but less central
 - Friends still nice, but less central
- Happiness and life satisfaction impacted, but no time to worry about meaning in life

Siblings



Siblings: What we knew

- May be more empathic
- May be more likely to go into social services occupations
- May be sometimes embarrassed
- Sisters give more caretaking
- Brothers give more caretaking than those who do not have a sibling with disabilities
- Brothers more likely to be proud of and take care of a brother with disabilities
- Siblings may be reluctant to add to parent burdens

Sibling Study

- Tracy Olson

29 participants

14 males, 15
females

Ages 13 to 42,
with a mean of
20.8

- Rachel Vert

Average age of
sibling with
CHARGE 17.4,
range 3 to 29

Sibling Evaluation Scale

- Largest impact
 - Felt isolated
 - Became more tolerant
 - Became more responsible
 - Obtained accurate information
 - Concerned about sibling's future

Social Isolation

- Loneliness
 - Significantly less lonely than college student sample
- Utilization of social support
 - Similar to college student sample in openness to social support
- 12 had attended a CHARGE conference
- 7 had attended a support group
- 10 had friends with a sibling who has disabilities

Perceptions of Family Stress

- All but two believed their parents experienced at least a moderate amount of stress, and a majority attributed at least part of that to the sibling with CHARGE
- 21 tried to avoid adding to their parents' stress levels
- On a measure of family hardiness, mean was significantly less hardy than norms

Sense of Well-Being

- Scored high on a measure of personal well-being
- Those with a higher sense of personal well-being were significantly less lonely and more likely to utilize social support.

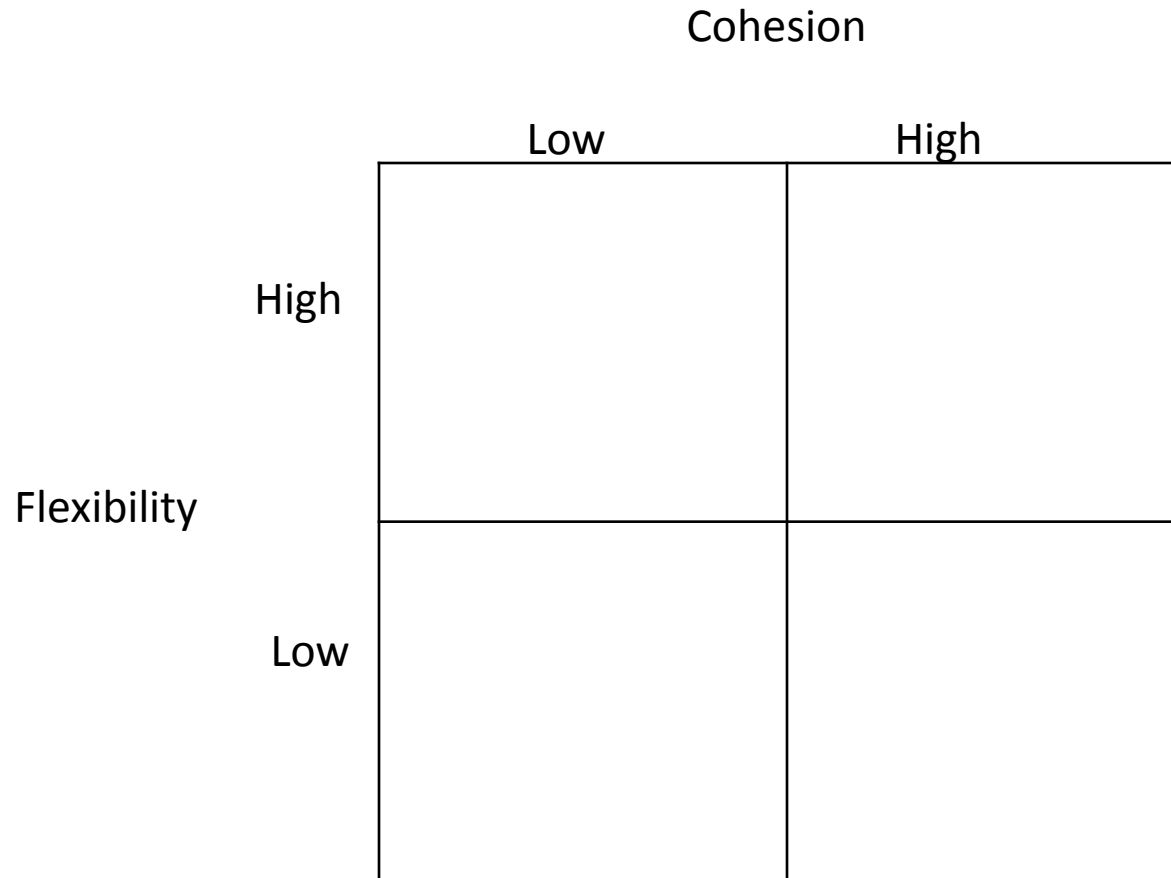
Other variables

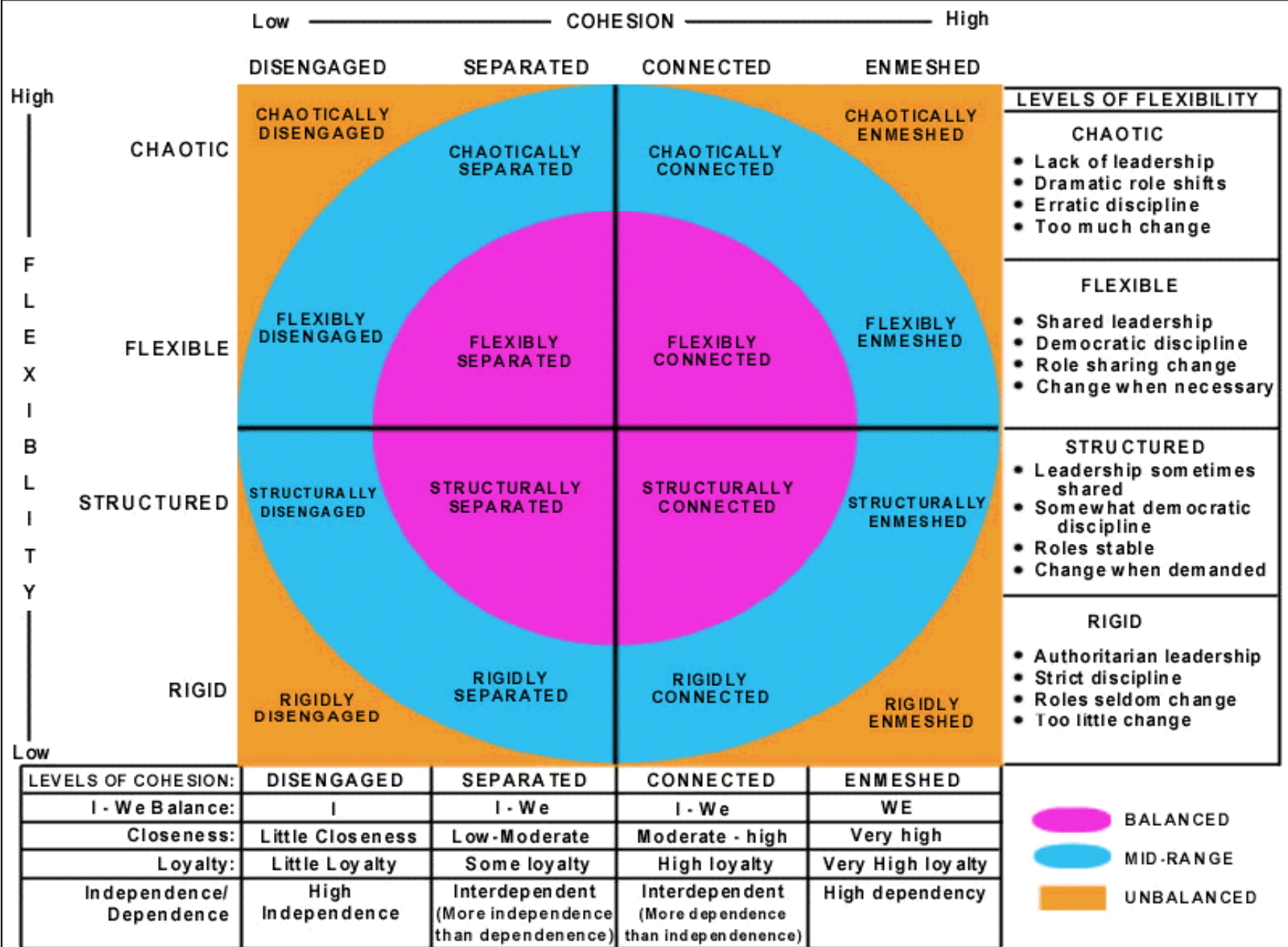
- No differences by
 - Sex
 - Birth order
 - Age of participant
 - Age of sibling
- Choice of career
 - No differences between those going into helping vs. non-helping careers
 - Except those going into helping careers believed their sibling influenced the choice

Not so different?

- Generally highly value their sibling with CHARGE
- Recognize parent and family stress, and try not to add to it
- Do not feel particularly lonely or isolated
- Generally perceive themselves as doing well

Family system





System Flexibility/Adaptability

- Leadership – who is in charge?
- Rules/roles – who can do what?
- Negotiation – how are decisions made?
- Organization – how orderly are things?
- Values – what is important?

Chaotic

Flexible

Structured

Rigid

System Cohesion

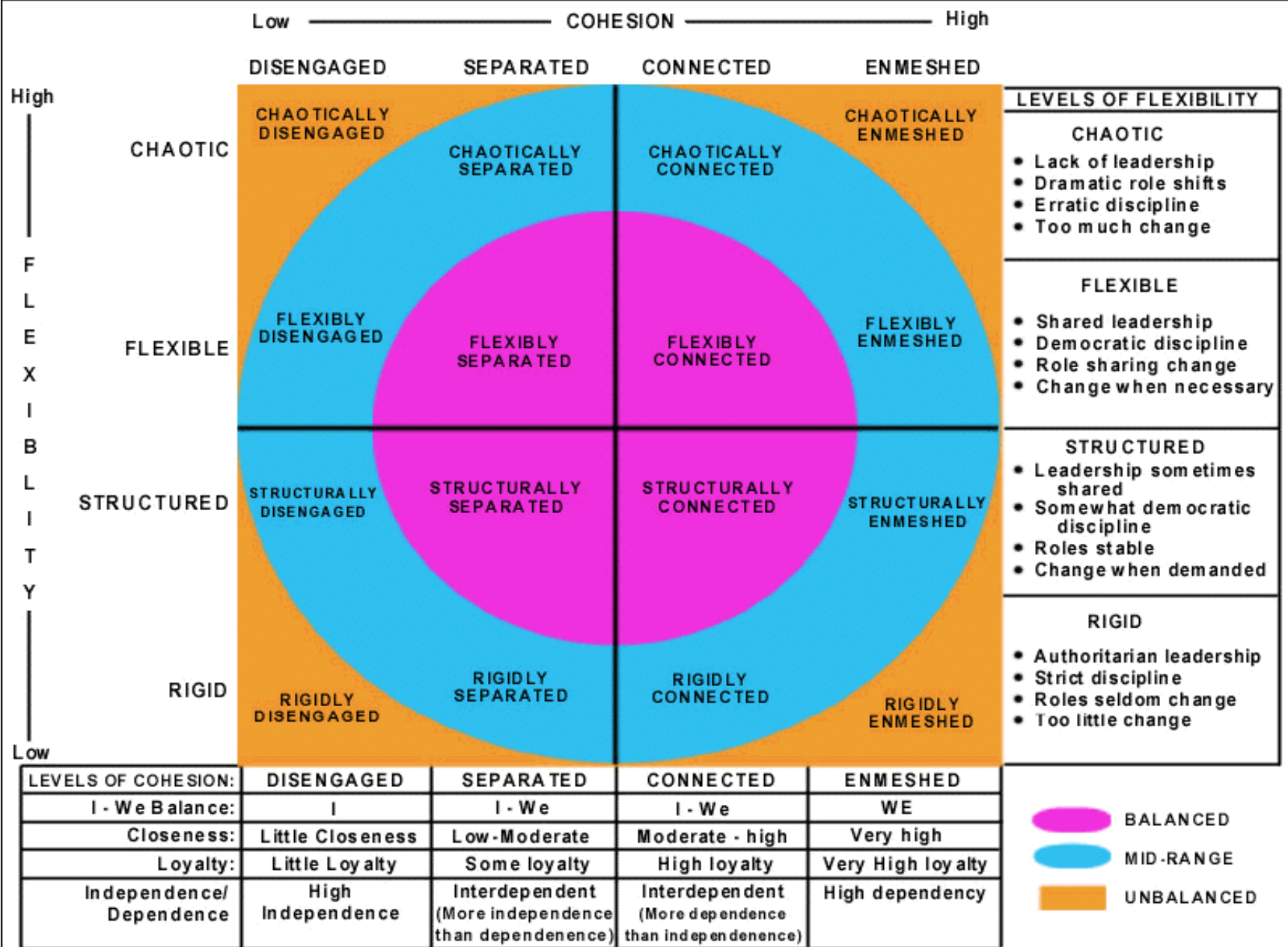
- Closeness – involvement
- Support – how much backup
- Decision-making – who benefits
- Commonality – what is shared
- Unity – how is morale

Disengaged

Separated

Connected

Enmeshed



In the end, it is about family

- Sources of meaning
- Sense of belonging
- Family motto
- Family ritual
- The courage to be imperfect
- Getting along and contributing
- How we define ourselves



MARATHON SKILLS

Ann P. Turnbull

- Meet basic needs
- Know your self and your family
- Love unconditionally
- Establish relationships
- Experience and benefit from emotions
- Take charge
- Anticipate the future
- Establish balance



**Friday, July 31, 2015
Breakout Session #10: 1:00- 2:00pm
Schaumburg West**

**How to talk to your child about having
CHARGE**

**Kasee Stratton, Ph.D., NCSP
Mississippi State University, Assistant
Professor/Licensed Psychologist**

Presenter Information:

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She currently runs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome*, the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Stratton specializes in challenging behavior and improving adaptive skills. Prior to joining faculty at Mississippi State University, Dr. Stratton completed her pre-doctoral and post-doctoral fellowship at Johns Hopkins School of Medicine and the Kennedy Krieger Institute. Dr. Stratton carried a heavy caseload of individuals with a variety of developmental concerns and helped many families start the discussion with their child about their syndrome. This work continued in 2013-2014 as Dr. Stratton was employed as a licensed psychologist at a center for children with disabilities.

Presentation Abstract:

When and how do I tell my child they have CHARGE syndrome? While each family will decide when the time is right for them, it is never too early or too late to learn how to begin this discussion. This presentation will cover the benefits of letting your child know about CHARGE and will address specific talking points. Child- and family- friendly strategies for introducing the diagnosis to your child will be presented.



**Friday, July 31, 2015
Breakout Session #11: 1:00- 2:00pm
Schaumburg E-F**

CHARGE 101

**Meg Hefner, MS, Genetic Counselor,
Saint Louis University
Nancy Salem-Hartshorne, Ph.D.,
Delta College**

Presenter Information:

Meg is a genetic counselor with more than 30 years' experience with CHARGE syndrome. She was a founding Board member of the CHARGE Syndrome Foundation, Inc and has written extensively on CHARGE for families (editor of the Management Manual for Parents), geneticists (American Journal of Medical Genetics special issue on CHARGE syndrome), and professionals working with children with CHARGE (CHARGE Syndrome, Plural Publishing). Awards for CHARGE syndrome work include the first Stars in CHARGE award and the Diane Baker Alumni Award from the University of Michigan. She has presented at every CHARGE Syndrome Conference and actively answers genetic and medical questions on several CHARGE Facebook pages. Meg's most recent endeavor is the CHARGE syndrome database, which will help provide baseline information to CHARGE researchers around the world.

Presentation Abstract:

The "CHARGE 101 series" (presentations #11, 17 and 23) is intended for parents of babies and young children with CHARGE and first-time conference attendees. We will present an overview of what is most important about your child with CHARGE: diagnosis and features of CHARGE, behavior, communication and outcomes. We encourage you to attend all three presentations.

In the first hour, Meg will begin with a review of the features used in making a clinical diagnosis (how do we know it is CHARGE and not something else) and then cover many of the other medical features seen in babies and children with CHARGE. Then she will talk about how the sensory deficits in CHARGE (e.g. hearing, vision and balance issues) affect early development, with emphasis on the communication bubble. In the second hour, Nancy will present an overview of what we know about behaviors and developmental outcomes in CHARGE and introduce communication options. Finally, Rob will continue with the importance of communication, what constitutes communication and the importance of play and other interactions in learning.

CHARGE Syndrome 101 Series for New Families

Meg Hefner, M.S.

Genetic Counselor

Adjunct Associate Professor of Pediatrics
Saint Louis University School of Medicine

meg@chargesyndrome.org

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

Name features should NOT be used for diagnosis

Epidemiology of CHARGE Syndrome

- ~1 in 10,000 births worldwide
- Most often a new dominant mutation
 - Copy error in sperm production
 - Increased but low (1-2%) recurrence risk
 - Major gene identified in 2004 (CHD7)
- Mild end of spectrum is still being investigated

Major CHARGE gene: CHD7 on chromosome 8q12

- Very large gene with many unique mutations
 - Gene sequencing required, expensive
 - What the gene does is being worked out
- If CHD7 mutation is identified
 - It confirms diagnosis in questionable cases
 - Help define the mild end of the spectrum
 - *Not finding a mutation does NOT rule out CHARGE*
 - Possible to test other people in the family
- Prenatal diagnosis /Pre-implantation genetic diagnosis
 - First identify a CHD7 mutation in the person with CHARGE
 - Presence or absence of a mutation but *not the severity of the features*
- *See presentations on Genetics of CHARGE*

Chance of Recurrence

- Parents who have one child with CHARGE:
1-2%
- Children of individuals with CHARGE:
50%
- Extended family members:
- $<<1\%$



Two children who have CHARGE, three (two pictured) without

Three generations of CHARGE syndrome



Diagnostic Criteria and Medical Features

What features help us know it is
CHARGE

What are some of the other features

Evolution of the Diagnostic Criteria

- 1981 – Acronym (CHARGE) features
- 1998 – Blake criteria (Major and Minor features)
- 2003 – Verloes criteria (added inner ear features)
- 2004 – CHD7 mutation
- 2015 – refined again

2015 Criteria

- Positive gene test (CHD7+) with some features
 - Is a CHD7 mutation enough for a diagnosis?
- ***Clinical Diagnosis*** based on a combination of Major and Minor clinical features

Major diagnostic features

- Common in CHARGE but rare in other syndromes
 - Coloboma of the eye
 - Cranial nerve anomalies
 - Choanal atresia/stenosis
 - Characteristic CHARGE ears
 - Outer
 - Middle
 - Inner

Coloboma – cleft of the eye



Iris Coloboma

Increased sensitivity to light



Retinal Coloboma

Increased risk of retinal detachment with retinal coloboma



Retinal coloboma causes visual field defects

Retinal coloboma: upper visual field defects



Children will position themselves to be able to see

Macular coloboma: central vision loss

- The child will look *above* the point of interest and *not appear to be making eye contact*
- Note also facial palsy in this child
- This combination can make child appear inattentive



Cranial nerves 9 & 10: **Swallowing problems**

- Many require G-tube feeding
- May last for years
- Some adults avoid certain textures and continue to have choking episodes

Swallowing complication: Aspiration

- Aspiration is the most common reason for hospitalization in first 2 yr. of life
- GE reflux is common
 - Surgery (Nissen fundoplication) 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

Most have 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

Choanal atresia/stenosis

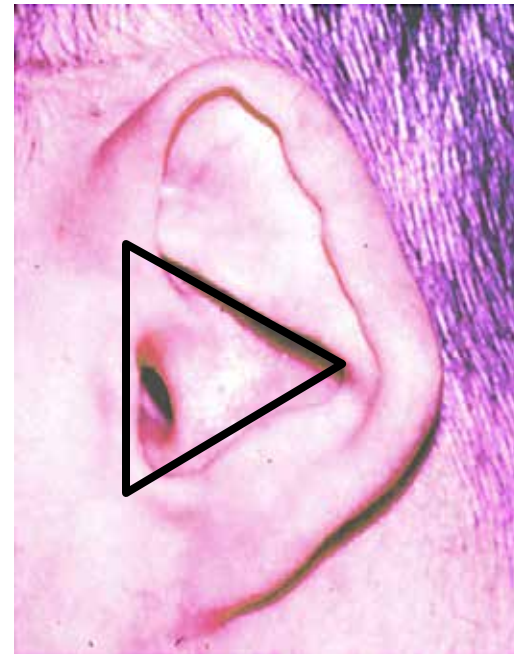
- Two sides (BL) or one side (UL)
- Bony or membranous
- Blockage (atresia) or narrowing (stenosis)
- UL stenosis may be difficult to diagnose:
 - goopy nose all the time
 - lots of infections, fluctuating hearing loss
- Often requires several surgeries to remain open



Baby with stents

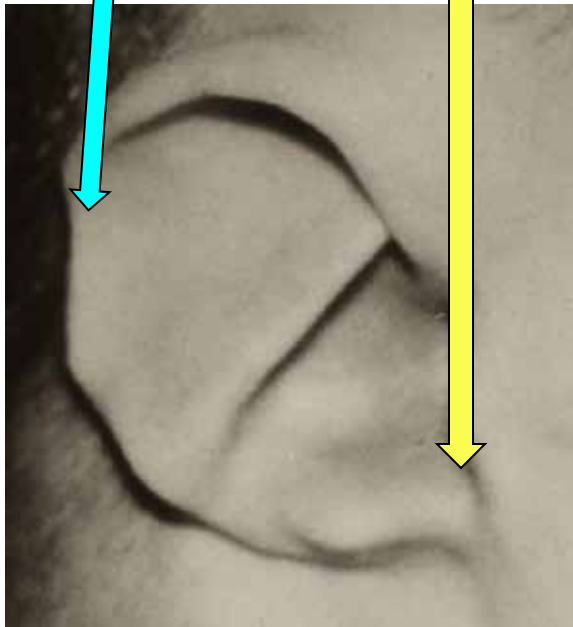
Characteristic CHARGE Ear: **OUTER EAR**

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



Characteristic CHARGE Ear

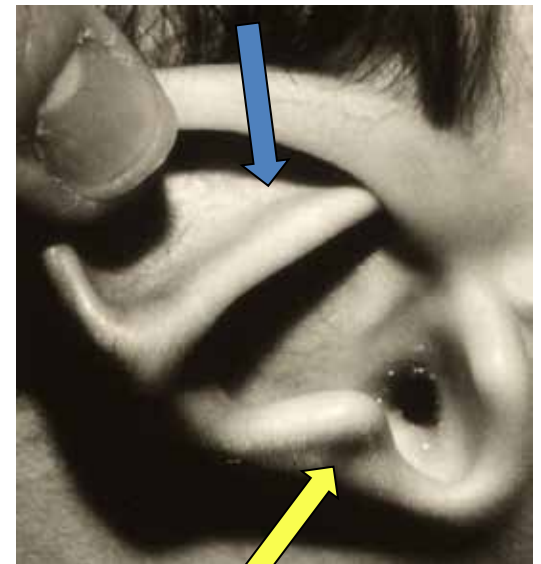
Snipped off helix with
small or absent
earlobe



Discontinuity between the
antihelix and antitragus



antihelix



antitragus

Characteristic CHARGE Ear:

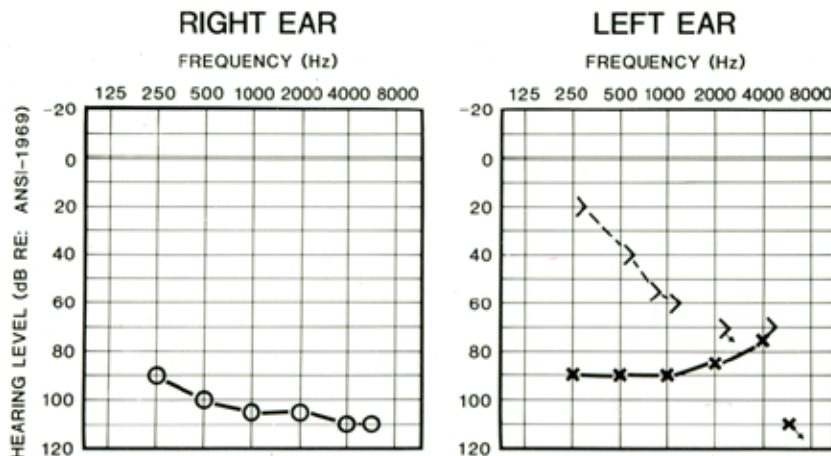
MIDDLE AND INNER EAR

- Middle ear bones – ossicular malformations (stapes, incus)
 - Conductive hearing loss – not helped by tubes
- Inner ear (*CT scan*)
 - Mondini defect: 1-1/2 turns to the cochlea
 - Small or absent semicircular canals
- Hearing Loss
 - Test early, test often
 - Know your child's hearing status

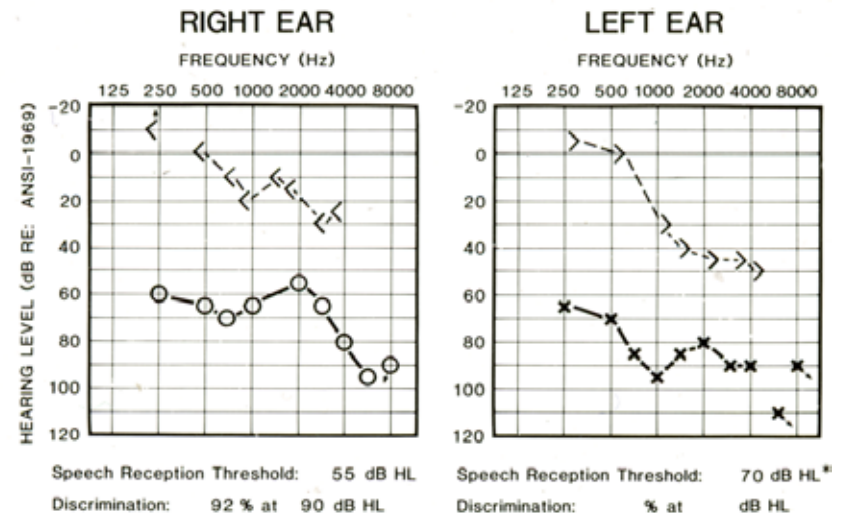
Hearing in CHARGE

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

27 MONTH OLD MALE (C.K.)



25 YEAR OLD FEMALE (C.R.)



*60 dB HL with earphone on side of head directly above ear

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”
 - Cruise for very long time
- Age of independent walking
 - Walk at 24-32 months if no other problems
 - Walk at 3-4 years if hypotonia and visual impairment

CHARGE Syndrome

Minor Diagnostic Features

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

CHARGE Syndrome Minor Features: Uncommon in other syndromes

- Upper body (?truncal) hypotonia
 - Slumping posture
 - Crawl on back
 - Bottom shuffle
 - Delayed walking
 - Complicated by poor balance
- Hockey stick palmar crease
- Characteristic CHARGE face



Characteristic CHARGE Face

- Square face
- Broad forehead
- Round eyes
- Nose

Prominent bridge

Square root

Prominent columella

- Small chin, gets bigger with age



CHARGE Syndrome Diagnosis

Minor Features common in many syndromes: **Heart defects**

- Tetralogy of Fallot
- Aortic arch anomalies
- Complex heart defects
- Can be any heart defect
- Same spectrum of heart defects as in VCFS (aka 22q deletion syndrome, DiGeorge)



Heart surgery at four years old

CHARGE Syndrome Minor Diagnostic Feature: **Facial 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
- Tracheomalacia
 - weak, collapsing trachea

CHARGE syndrome minor feature: Renal (**kidney**) anomalies

- Hydronephrosis (retained fluid)
- Reflux
- Horseshoe kidney
- Small or absent kidney
- Cystic kidneys

40% have renal anomalies

CHARGE Syndrome Minor Features

Short Stature

Due to:

- Medical problems
 - Heart
 - Feeding
- Growth hormone deficiency
- Short stature with unknown cause

Genital hypoplasia

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

Variability in CHARGE

- 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
- CHD7 testing is especially helpful in less clear cases, but should always be done

Other features of CHARGE

- Not necessarily helpful with diagnosis, but could be important medically or developmentally
- Differences have been noted in pretty much every organ system

Other features of CHARGE

- Brain anomalies seen on MRI or CT
- Seizures – can develop at any age
- Apnea
- Laryngomalacia
- Floppy cartilage
- Nipple anomalies
- Thymic or parathyroid abnormalities
- Immune system abnormalities

Other features - continued

- Webbed neck
 - Sloping shoulders
 - Absent trapezius muscle
- Abdominal wall defects
 - Umbilical hernia
 - Omphalocele
- Limb/skeletal anomalies
 - Limbs: missing or extra fingers or bones
 - Vertebral anomalies
 - Scoliosis
- Others (any organ system)

Additional CHARGE *management* issues

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

Intelligence in CHARGE

- Assume normal intelligence until proven otherwise - long-term prognosis may be very good
- “Input impairment” due to combined vision & hearing loss – see CHARGE 102
- 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

Delayed? Yes. But may have a lot of potential to catch up – you can't know yet.

CHARGE 101 – part 2

Sensory deficits and early
development in babies and
children with CHARGE syndrome

Senses in CHARGE

- *Vision loss – field and acuity
 - *Hearing loss – mixed, often severe
 - Balance abnormalities
 - Touch
 - Sensitive to certain textures
 - Light touch??
 - Altered pain sensation
 - *Smell - absent or decreased
 - Taste - possibly normal
- * distance senses

***What Percent of All Available
Information Do We Access
With Each of Our Senses?***

**Estimates when all senses are
working at full capacity**

Percent

100

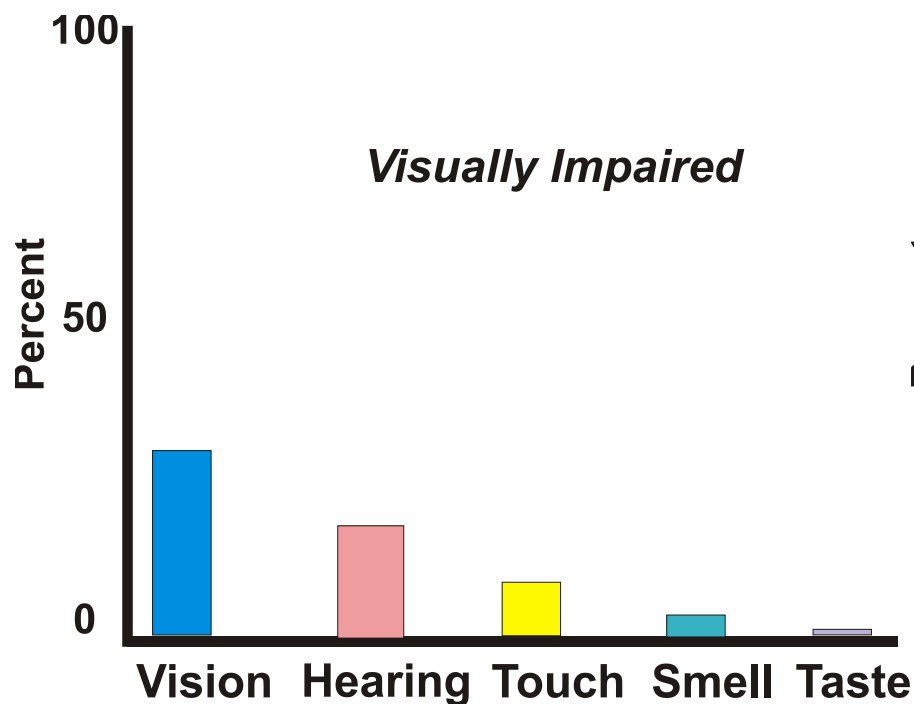
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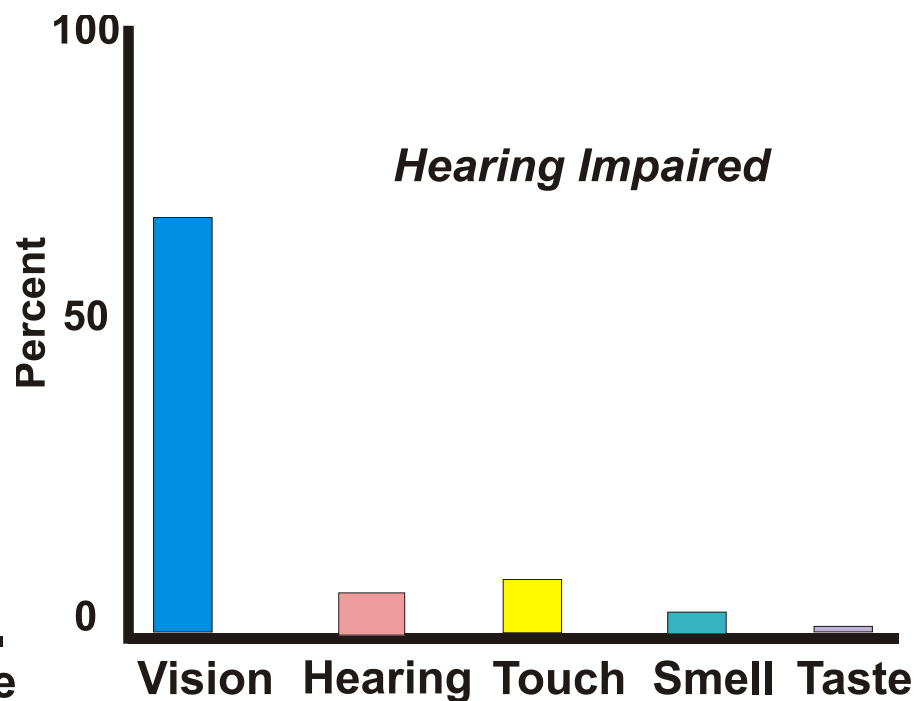
Vision Hearing Touch Smell Taste



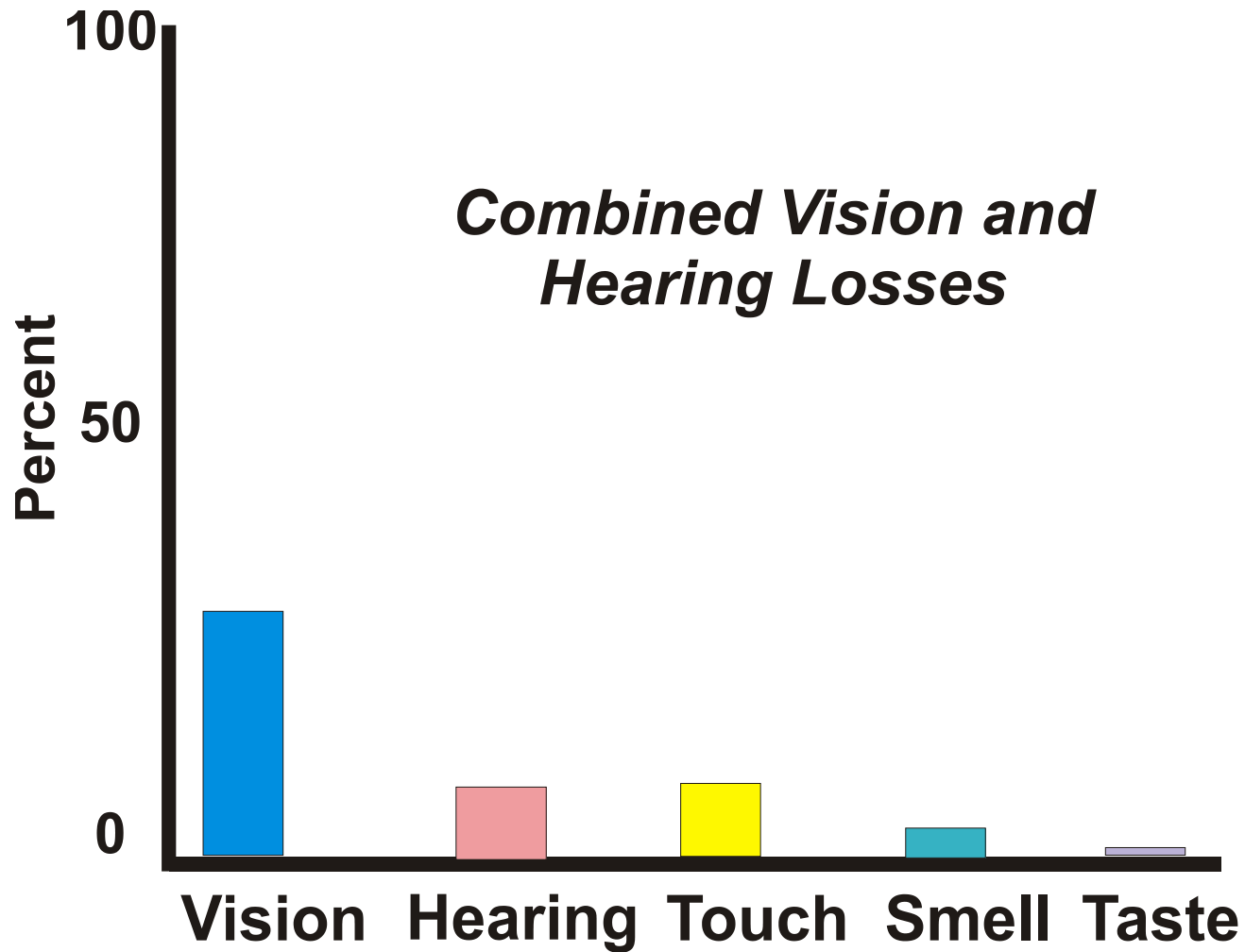
Missing half of vision or hearing



Less information from vision, full information from other senses



Less information from hearing, full information from other senses



Less information from vision, less from hearing:
Far less input overall – less information coming in

Distance Senses and the Communication Bubble

- How far out and how well can the child *see*?
 - Coloboma (visual field)
 - Acuity
 - Other
- How far out and how well can the child *hear*?
 - Sensorineural loss
 - Conductive loss
 - Fluid, infections
 - Amplification
- Can the child *smell*?

Communication Bubble

- The area *within which* the individual can
 - See
 - Hear
 - Smell
- Touch and taste do not give information at a distance
- Anything outside the bubble is “off the radar”

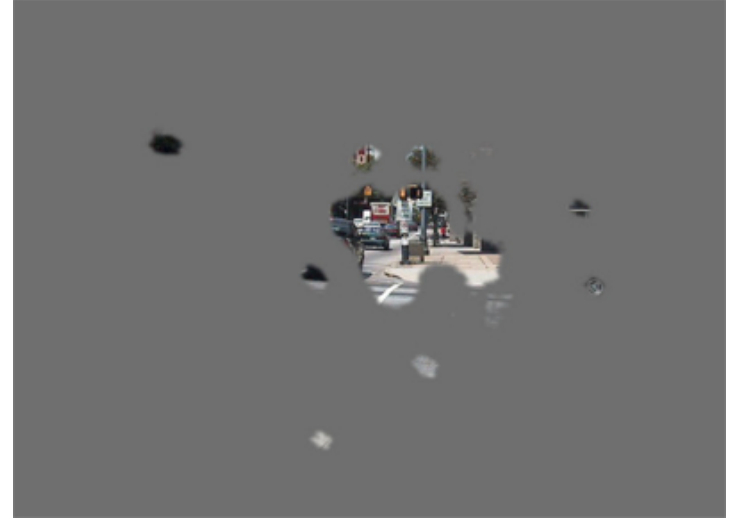
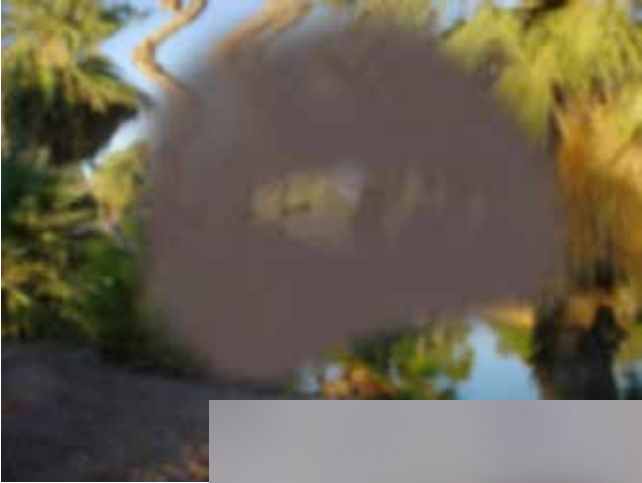
Living in a bubble - think of the bubble as opaque...



Anything outside the bubble is “off the radar”



Vision loss



If you are not in the child's visual bubble, she is not seeing you, not seeing you sign to her

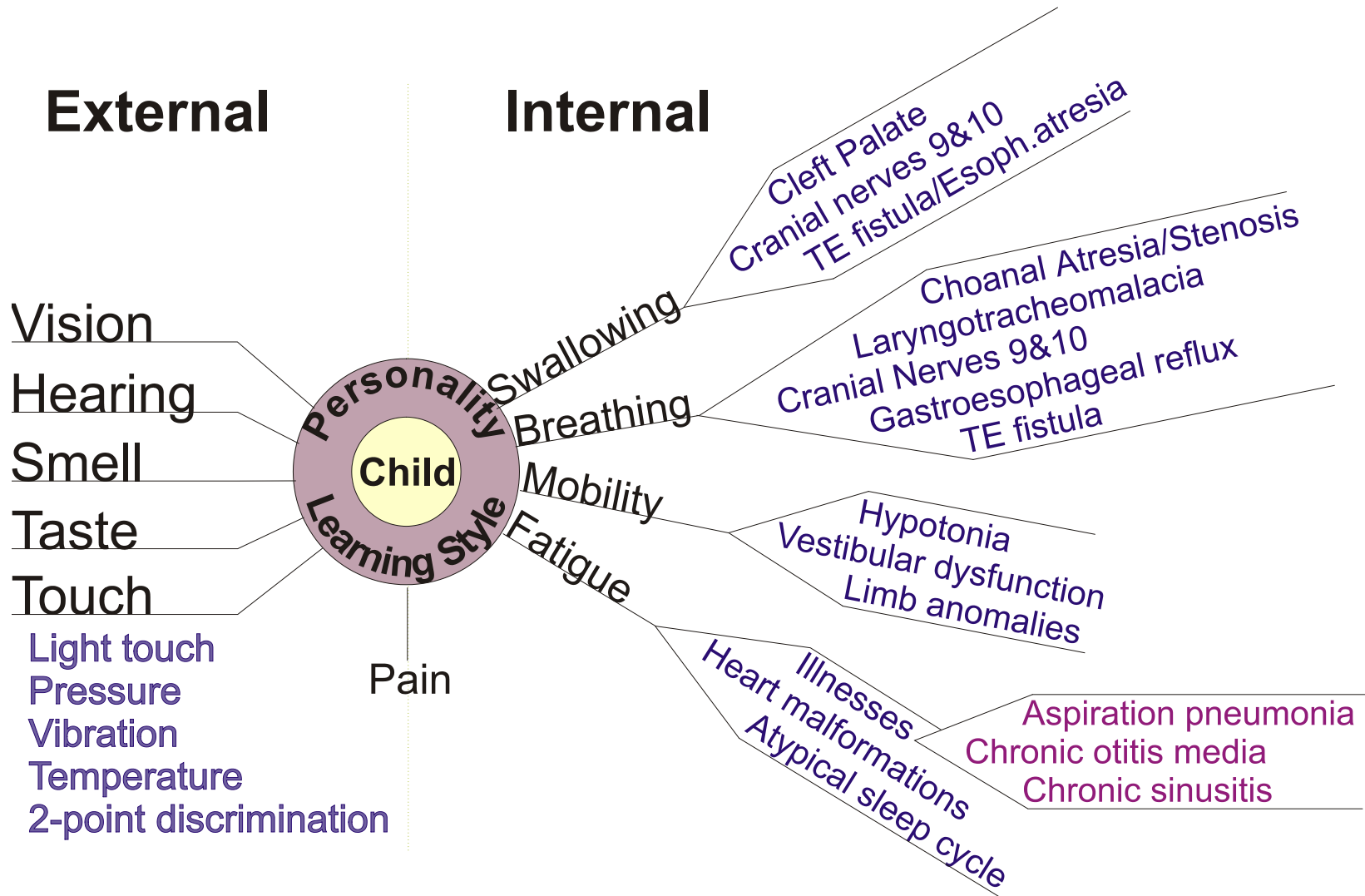
Sensory loss and development

- Early motor development – vision is important:
 - Head control
 - Sitting
 - Cruising, walking
- Communication: vision and hearing
 - Communication is learned by seeing and hearing – what if you don't see and hear?

Kids with CHARGE may be “Hard of” many things

- seeing
- hearing
- smelling
- feeling
- empathizing
- breathing
- balancing
- multitasking
- spacial awareness
- swallowing

Physical Influences on Development in CHARGE



Know your child

- Vision status (bubble)
- Hearing status (bubble)
- Balance
- Overall health, moods
- Observe carefully (learn from David Brown), communicate with caregivers
- Work on communication, communication, communication

Walk a mile in her shoes

- It is impossible to understand hearing loss and vision loss at a purely intellectual level
- Do a simulation yourself – try out your child's world
- Get the therapists and IEP team to participate in a simulation
- Find and use (read) the resources on the CHARGE Syndrome Foundation website

What is it like to have combined vision and hearing loss?

A simple (classroom) simulation

- Vision loss: plastic sandwich bags
 - Add tape for blind spots
- Hearing loss: foam ear plugs
 - 30 dB loss
 - “mild” hearing loss

How hard is it to

- See the teacher
- Hear the teacher
- See your work in front of you
- Hear others in the class
- Move about the room
- Stay focused
- Keep alert and awake all day



**Friday, July 31, 2015
Breakout Session #12: 1:00- 2:00pm
Schaumburg G**

**“What about me?” Guiding your
standard-developing child with their
experience of being a sibling to a child
with CHARGE**

**Michael Braga BS
Ruth Braga BA, MSN, RN
Founders of Super Incredible Brothers &
Sisters (S.I.B.S.)**

Presenter Information:

Michael & Ruth Braga's lives were changed forever in 1998 when they adopted their oldest son Brandon at one week old. Aside from knowing he had CHARGE Syndrome, they were clueless as to what that meant. Through their 21 years of marriage, they have experienced infertility, adoption, IVF, illness, loss, and a transcontinental move for Brandon's treatments/education-twice. Parents to three children now, they share their story and sense of humor with professionals and parents.

Despite being a high school dropout, Mike received his GED and ended up at the University of Utah, earning a BS, a spot on the Deans' list, and was named Inspirational Student of the Year in 2002. He ran the torch for the 2002 Paralympic Winter Games in SLC, pushing Brandon in his wheelchair. He will complete his MEd in Deaf/Blind Education in Spring 2015.

Ruth received a BA, but later returned for a nursing degree. She then received her MSN from Drexel and now teaches medical students, surgery residents and nursing school. Mom, wife, writer, instructor and home organizer with a sense of humor!

Presentation Abstract:

As parents, our homes are often consumed with the care of our sons and daughters with CHARGE. Between doctor visits, therapy, education and other issues, there comes a point when our typically developing children are left asking, "What about me?" With help from our 14-year-old daughter, this course will address the challenges that siblings face and how to implement survival strategies. Learn, share and hear from others and return home with new ideas to help each of your children flourish!

“What About Me?”
Guiding Your Standard Developing Child Through Their Experience of Being A Sibling
To An Individual With CHARGE Syndrome

Michael Braga BS, M.Ed. mbraga79@gmail.com
Ruth Braga MSN, RN ruthbraga1@gmail.com
With Mindy Braga-Age 14

We are parents to Brandon (CHARGE)-17, Mindy-14, and Nicholas-7. Brandon has led us through multiple adventures, but Mindy inspired us to create a sibling support group for kids in our area who have a sibling with disabilities.

Mindy will join us and talk about some of the challenges that come with being a sibling to a brother or sister with CHARGE from her perspective. As parents, we will discuss some of the ideas for meeting the emotional & developmental needs of your children without disabilities, how to find ways to improve their relationships with each other, and hopefully how to avoid going crazy in the process. We want to share what has worked, what flopped, and help you to find ideas that will work in your family unit.

Ultimately, we hope to help everyone understand that they are not alone. We conducted a survey about your children with CHARGE and their siblings and we will be sharing the results. We are from many places and backgrounds, but we are all here because of an individual with CHARGE! We look forward to learning from everyone!



Our presentation is sponsored and made possible by the Utah Developmental Disabilities Council



**Friday, July 31, 2015
Breakout Session #13: 1:00 - 2:00pm
Schaumburg H**

**Supporting Healthy Sexuality for
People with Disabilities
(Individuals with CHARGE 16+ Only)**

**Krescene H. Beck, QIDP, LNHA
Penina Simon**

Presenter Information:

Krescene Beck has a lifetime of experience supporting people with intellectual and developmental disabilities. Her enthusiasm and zest for life provide inspiration to everyone she meets. As a licensed nursing home administrator and former executive vice president of a large social service agency, Krescene has spent the past several years working on statewide grants focused on self-advocacy and on building collaborations to better respond to people with disabilities who experience sexual violence. Krescene's vision is simply put: Find your passion. Live your purpose. Celebrate your journey.

Penina Simon is from Montreal, Quebec, Canada. She will be turning nineteen in august and is studying creative arts at cegep (the pre-university program unique to the province of Quebec). She has grown up under the CHARGE diagnosis and is hard of hearing and has low vision. Penina enjoys art and education and has an interest in humanities.

Presentation Abstract:

Self-advocates age 16 and over only are invited to come together for a time of active learning and teaching about healthy relationships; healthy sexuality; rights, boundaries, and consent; and safety and support. People with disabilities experience sexual violence at a disproportionate rate. They are also less likely than others to identify abuse and seek services when violence does occur. One of the factors that contribute to the increased risk is the lack of information and education people with disabilities receive on issues related to healthy relationships, sexuality, sexual violence prevention and body safety. This type of education, long denied or overlooked, is central to empowering people with knowledge of healthy relationships as well as information on how to report sexual violence when it occurs.

Prevention Education Program Guide

Disability service agencies and rape crisis centers can help prevent sexual violence against people with disabilities. They can also respond effectively to women who experience sexual violence. Achieving these goals requires collaborative efforts to educate women with disabilities about healthy sexuality, sexual rights and sexual violence, and safety planning. This guide will help disability service agencies and rape crisis centers:

- Be aware of basic information about people with disabilities and healthy sexuality;
- Understand the sexual rights of people with disabilities; and
- Prepare and deliver education sessions to people with disabilities regarding sexual violence, healthy relationships, healthy sexuality, and sexual violence.

www.icasa.org/docs/illinois%20imagines/mini%20module%204%20education%20guide.pdf

Prevention Education Program Picture Supplement Guide

This module can be used as a supplement with the Prevention Education Program Guide. It includes concrete examples and activities based on the preferred learning styles of some people with intellectual and developmental disabilities. The supplement includes strategies, activities, handouts, and information for teaching sexual violence education for people with disabilities to enhance the process for both participants and teachers.

www.icasa.org/docs/illinois%20imagines/module%204b%20picture%20guide.pdf

A Guide for Parents and Guardians

Parents and guardians are key to the work of preventing sexual violence against people with disabilities. As a parent or guardian, you are in an ideal position to educate and support the education of loved ones in relation to healthy sexuality, sexual rights and sexual violence, and safety planning. You are also likely to be the first responder to people with disabilities who experience sexual violence. This guide will help you, as a parent or guardian, to

- Be aware of basic information about people with disabilities and healthy sexuality
- Understand the sexual rights of people with disabilities
- Be aware of risks and needs of people with disabilities in relation to sexual violence
- Support and supplement education for people with disabilities regarding sexual violence, healthy relationships, healthy sexuality and sexual violence risk reduction

www.icasa.org/docs/illinois%20imagines/soft%20cover%20guide%20for%20parents%20and%20guardians%20module%204.2.pdf

Additional Materials

Illinois Imagines has produced a variety of education and other material for use by self-advocates, family members and guardians, rape crisis centers, and disability service agencies.

www.icasa.org/index.aspx?PageID=1045



**Friday, July 31, 2015
Breakout Session #14: 2:15 - 3:15pm
Schaumburg A-B**

**Identifying Effective Positive
Behavioral Supports for Young Adults
with CHARGE Syndrome**

**Susan M. Bashinski, Ed.D.
Associate Professor, Special Education
Missouri Western State University
Susan M. Bruce, Ph.D.
Professor, Special Education
Boston College**

Presenter Information:

Susan M. Bashinski has 38 years' experience with learners who experience multiple disabilities. She has directed numerous federal and state grants in low-incidence disabilities and deaf-blindness, including: personnel preparation, research, model in-service training, and assistive technology. Dr. Bashinski has extensive experience in providing professional development and technical assistance nationally and internationally, particularly in the areas of augmentative and nonsymbolic communication for learners who have low-incidence disabilities, including deaf-blindness and CHARGE syndrome. Her research interests and areas of expertise include early communication and language development, augmentative communication, and cochlear implants, with numerous publications and presentations related to these topics. Dr. Bashinski authored the chapter on assessment of prelinguistic communication for the Hartshorne, Hefner, Davenport, and Thelin 2011 book, *CHARGE*. She has given both paper and platform presentations at the 2009 and 2011 CHARGE Conferences.

Presentation Abstract:

This presentation will share findings from an action research study that examined three elements of positive behavioral interventions & supports (PBIS): adult use of language, environmental arrangement, and sensory system sensitivities and needs. The presenters will utilize videos to demonstrate the application of proactive and reactive behavioral intervention strategies with five young adults who have CHARGE syndrome. Excerpts from profiles of these young adults will also be shared.



Strategies to Support Positive Behaviors in Students with CHARGE Syndrome

Susan M. Bashinski, Ed.D., Missouri Western State University
Susan M. Bruce, Ph.D., Boston College



Review of Literature: PBIS



Positive Behavior Intervention Supports apply behavioral principles to support emotional regulation

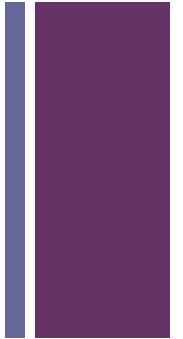
Learners with CHARGE syndrome:

- n often have problems with emotional regulation
(controlling one's emotions)
- n usually have high anxiety levels

(Davenport & Heffner, 2010; Hartshorne & Salem-Hartshorne, 2010)



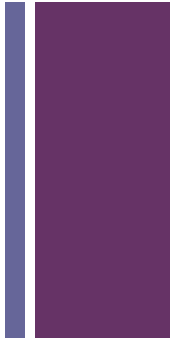
Review of Literature (con't.)



- n PBIS is proactive rather than just reactive
- n PBIS looks beyond the learner
 - n Considers sensory integration needs
 - n Considers environment
 - n Uses information from Functional Behavior Assessment (FBA)-on purposes of behavior to build the Positive Behavior Intervention Support Plan
 - n Cites alternative/replacement behaviors that must be taught (such as communication) (Horner, 2000)



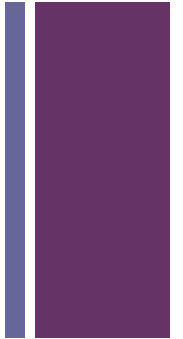
Cognitive Behavior Therapy (CBT) Elements



- n Cognitive Behavior Therapy: Action oriented form of psychosocial therapy-replacing maladaptive thinking and behavior with constructive thinking and behavior
- n We applied 10 elements of CBT: Individual is active participant, implementation in familiar settings and activities, *cognitive rehearsal* through social stories, *role play*, reinforcement to condition positive responses, *systematic desensitization* through carefully controlled exposures, *cognitive restructuring*-choice making, labeling, redirection, & relaxation techniques



Action Research



- n Recursive, problem solving form of research
- n Cycles of action-reflection-action-reflection
- n Four types: classroom, collaborative, critical, and participatory
- n **OUR STUDY**: Collaborative action research with collective case study design

(Bruce & Pine, 2010; Hendricks, 2009)



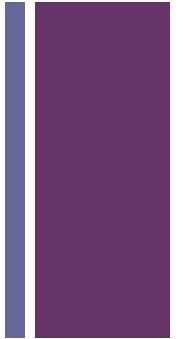
Research Questions



- n What positive behavior intervention (PBIS) supports are most effective with each of the young adults who are deafblind?
- n What language supports, especially adult use of sign language, will support positive behaviors in students?
- n How does environmental engineering support positive behavior in each student?
- n What sensory-motor integration strategies are helpful in promoting behavior in each student?
- n Which strategies are important to preventing a negative behavior and which are important to addressing a negative behavior?



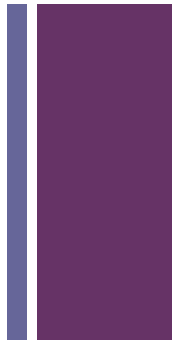
Young Adult Student Participants



- n 7 young adult students*
- n 5 with CHARGE syndrome (4 who are deafblind, one who is deaf with additional disabilities)
- n Ages: 18-22 years
- n 3 males, 2 females
- n *This presentation will focus on the 5 students have CHARGE syndrome: Joe, Jon, Gail, and Nathan, and Renee



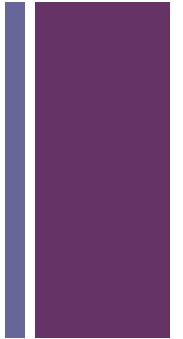
Five Students with CHARGE Syndrome



- n **Joe:** Profound bilateral hearing loss, 20/900, right field restriction, severe delays, expresses in signs and sign combinations
- n **Jon:** moderate bilateral hearing loss, left eye-20/300, right eye-20/30, superior field restriction, moderate delays, expresses in sign language
- n **Gail:** Severe bilateral hearing loss, 20/360, superior & inferior left and lateral left field losses, moderate delays, expresses in speech, sign language as back-up (especially receptive)



Five Students with CHARGE Syndrome (con't.)



- n **Nathan:** Moderate-severe hearing loss, left eye-20/70, right eye-20/800, moderate delays, expresses in sign language
- n **Renee:** Profound hearing loss, normal vision, severe delays, expresses in pictures/line drawings, body language, some signs/gestures



Adult Participant/Co-Researchers



- n 3 classroom teachers with expertise in deafblindness
- n 1 teacher liaison
- n 1 psychologist with expertise in Cognitive Behavior Therapy (including adaptations for those with developmental delays)
- n 6 paraprofessionals
- n 1 university faculty member



The Intervention

Our study focused on three areas of PBIS:

- n Adult use of language
- n Sensory needs of the learners
- n Environmental engineering/arrangement



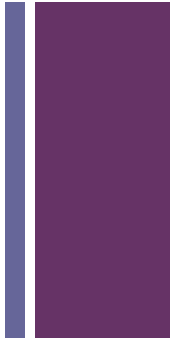
The Intervention (con't.)



- n Teachers recorded data on behavior
- n Teachers videotaped at times that were often problematic for each student (specific transitions and activities)
- n Teacher and psychologists discussed data
- n All research team members discussed a focus student each month and generated a PBIS Profile
- n Students participated for 6-9 months, depending on their graduation date



Data Sources



- n Behavior chart addressing our 3 areas of focus
- n Some students had additional behavior sheets provided by the psychologist
- n Classroom journals (for teachers and paraprofessionals to record ideas about each student's behavior)
- n Videotaped observations that captured proactive and reactive strategies (transcribed)
- n Ongoing development of Positive Behavior Intervention Support Profile for each learner. Completed profiles were member checked by teachers.



Data Analysis



- n Constant comparative, elements of grounded theory
- n During data collection-observing for themes for each student/case with influence on teaching (action cycles)
- n After data collection-identification of themes that cut across cases (collective case analysis)
- n Open coding (capturing the specific) and axial coding (identifying families or categories of data)

(McHatton, 2009)

+ Environmental Engineering & Sensory Sensitivity: Sample Flip Chart Data from Meeting re: **Joe**

Asking him: "it's going to be really loud - do you want to leave?"

Designated area that is his area - showing him how much space he has - what space is "his"

last year - sitting @ desk and/or doing his schedule were triggers

being "jostled" by others

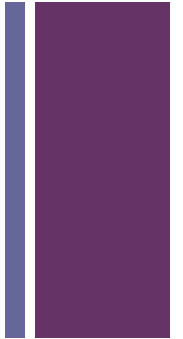
being active - likes

Sensory making backpack heavier (sensory)

discontinued "signing in" for workroom - trigger - begins crying

+ Findings

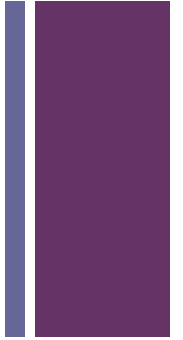
8 Major Themes/Super-Categories



- n Provide structure
- n Support students to cope with anxiety
- n Address students' sensory needs
- n Support on task behavior
- n Support transitions between activities & environments
- n Support mature behavior (cooperating & adult-like)
- n Establish & maintain positive climate
- n Adult language supports positive behavior



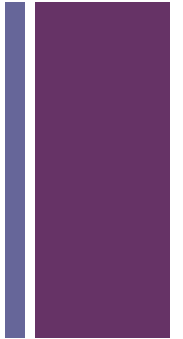
Theme #1: Provide structure



- n Directly teach rules, practice, remind, have learner restate the rule
 - n Individual nuance: Give **Nathan** the direction, step back to allow him time to process, don't repeat the direction. At times, remind him that everyone has rules to follow.
- n Use schedules & calendars(related to learning routines)
 - n Individual nuances: **Joe** needs novelty built into schedule; **Jon** requires that the entire schedule is reviewed whenever an activity is canceled; **Renee** needs first/then language (work first, then computer)



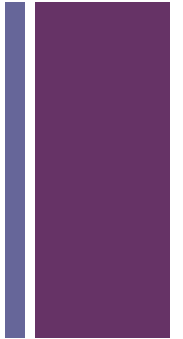
Provide structure (con't.)



- n Establish & teach routines (within & between activities)
- n Structured physical spaces
 - n Well defined workspace essential for staying calm (**all students**)
 - n Secure place to leave unfinished work (**Jon**)
 - n Chill out space (when overwhelmed/stressed) (**Joe**)



Theme #2: Support students to cope with anxiety



- n Structure (Theme #1) helps
- n Keep students informed
- n Gross motor exercise, rough & tumble play (3 students)
- n Calm spaces
- n Consider influence of sensory sensitivities (Theme #3)
- n Support students to learn & identify their individual strategies:
 - n **Gail**: Use of mature behavior sheet, turning down hearing aids when overwhelmed, establishing physical distance from irritations

+ Support students - cope with anxiety

(con't.)

- n Redirect to new activity if obsessional compulsive disorder (OCD) behavior is difficult for student to control
- n Switch communication forms: Use sign instead of speech with **Gail** and fingerspelling or tactile sign instead of visual sign with **Joe**
- n Relaxation techniques: For example, deep breathing, sighted guide and deep pressure for **Gail**
- n Use counting-1st, 2nd, 3rd for what will happen (**Joe & John**) and count to 5 to relax (**Gail**)
- n Suggest he not worry about that now (**Nathan**)

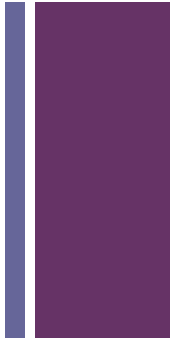
+ Support students - cope with anxiety

(con't.)

- n Unexpected touch as source of anxiety for most students (deafblindness impact)-avoid situations where unexpected touch is likely to occur-or restructure
- n Introduce something new to break up OCD (**Renee**)
- n For repetitive talk: Know their topics; acknowledge communication and redirect topic to appropriate time and place, may need to reassure
 - n Example: **Gail's** talk about Wizard of Oz
 - n Example: Repetitive talk about future event-such as holiday (3 students)



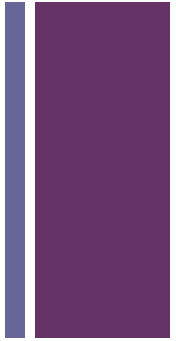
Example of Repetitive Talk: Excerpt from PBIS Profile Draft re: **Gail**



- n Behavior = Repetitive talk about Wizard of Oz
- n Staff must be aware of manipulations to get into such conversations
 - n She might say “Call me Dorothy”
 - n She might say “Do you like shoes?” Who wears red shoes?” This leads to Dorothy conversation.
 - n She might say, “I like yellow.” This leads to talking about the yellow brick road.
- n Adult use of language: Staff talk about real/not real



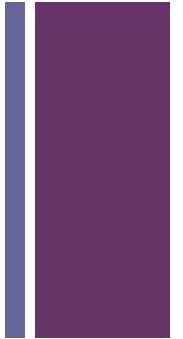
Theme #3: Address Students' Sensory Needs



- n Examples of sensory needs/sensitivities:
 - n **Joe:** Comfy chair with body parts touching specific parts of chair, deep pressure, weighted backpack
 - n **Jon:** Prefers chairs without arms, weighted beanbag & cart, sensory diet, spinning
 - n **Gail:** Sensitive to sounds and accidental touching-avoid crowded elevators. She knows her strategies for sound.

+ Address Students' Sensory Needs

(con't.)



- n **Nathan:** Hot and cold temperatures, must have goggles for swimming, likes rough and tumble play, needs exercise and stretching to stay positive (treadmill), beanbag and computer access for calming after meltdown
- n **Renee:** Needs lots of sensory input-body sock, weighted blanket, beanbag, clapping, squeezes, deep pressure, she often leans on surfaces to satisfy a sensory need

+ Theme #4: Support On Task Behavior

- n Provide sufficient wait time for response
- n Reinforcement-higher for non-preferred tasks
 - n Remind of upcoming reinforcer
- n Counting down to give foreseeable end (how many more)
- n First/then language
- n Reduce distractions (including watching what you do)
- n Redirection
- n Consider preferred activity-end each session w/ preference(**Joe**)
- n Token economy (**Nathan**)

+ Theme #5: Support Transitions Between Activities & Environments



- n Consistent use of finished sign
- n Consistent use of daily schedule
- n Preview what will happen in next environment
- n Sufficient wait time
- n First/then language (**Renee**)
- n Simple language (**Renee**)
- n Adult use of language (Theme #8 applies here)

+ Theme #6: Support Mature Behavior (cooperation & “adult-like ‘ behavior)

- n Wait time to process (**Joe, Jon**)
- n Verbal compromise (**Gail**)
- n Use of mature/adult behavior sheet
- n Talk about “mature” and “not mature (**Nathan, Gail**)
- n Use of self-evaluation sheet (**Gail**)
- n Know student’s types of refusals & be prepared
- n Set limits
- n Give physical space as needed



Journal Excerpt re: **Joe's** Behavior



“When he starts having negative behavior (pushing things, ignoring staff, not aggressive-but maybe on edge), give him a reminder—If you want to _____(name preferred activity), you need good behavior”....Seems to be more effective since it gives him an incentive instead of just reminding him about the rules.”



Theme #7: Establish & Maintain Positive Climate



- n Keep staff attitude/behavior positive-wording, signs, voice, facial expression
- n Use reassuring language and tone
- n Physical distance from upsetting events-some students can initiate this, others need adult support
- n Adjust expectations on difficult days
- n Appropriate types/levels of reinforcers
- n Preferences: Favorite topics (**Gail**), computer (**Renee**)
- n Consistency in environments

+ Establish & Maintain Positive Climate

(con't.)

- n Provide choices, but consider each student's requirements (open ended or choice of two)
 - n **Nathan** likes choices of reinforcers
- n Watch your position-stay in close proximity-but where depends on student
- n Restructure environment to keep it positive (sensory inputs)
- n Provide socialization opportunities
 - n **Joe** needs silly time with others
 - n **Gail** needs contexts for relationship building

+ Theme #8: Adult Language Supports Positive Behavior

- n Use very few directives (decide what you will save this for)
 - n Example-Use of “no” for touching others
- n Keep your language positive
- n Be aware of trigger topics
 - n Topics you don’t have info about/vague topics (**Joe**); home, people who have disappeared from his life, deaf, death, skinny vs. fat (**Nathan**)
- n Be aware of students’ trigger words and possibly substitute other words
 - n **Joe**: Use “celebration” not “party”
 - n **Nathan**: “death” and “deaf”
 - n **Renee**: “no” and “you need to ...”



Adult Language Supports

Positive Behavior (con't.)

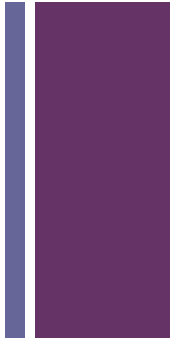


- n Provide specific information
 - n **Joe, Jon, & Gail** need info about environments—who is there, what they will do, noise levels...**Nathan** seeks much information, but be careful it doesn't become task avoidance
- n Refer to past events that are similar—point out student success in past event
- n Talk about mature/not mature behavior (**Gail, Nathan**)
- n Provide language for their concerns—words/signs
 - n **Nathan**: Model correct American Sign Language structure
Encourage students to use their strategies



Adult Language Supports

Positive Behavior (con't.)



- n Modify forms (from visual to tactile sign..)
- n Suggest thinking before action (**Nathan** only)
 - n Adult Messages:
 - n “I want you to think”
 - n “Think before, what’s next?” (for schedule reinforcement)
 - n “Try and think before doing”
 - n “Stop and wait. I want you to think. Slow. Patient. One minute.”



Journal Excerpts on Adult Use of Language with **Gail**



- n For disorganized thinking, thinking in the past or too far in the future-help to refocus **Gail** on the NOW. “Let’s save that until June. You can ask me in June. When can we talk about that? Why don’t we save that for _____(name month).”
- n In response to her talking in detail about past events- because she wants things to happen exactly the same way----”Use language that points this out to her- acknowledge that via language. Are you asking if same people are going/we’re doing same thing because you like things to be the same?”

+ Video: **Gail** reviewing her self-evaluation sheet & strategies



+Video: Identifying Positive Behavioral Interventions & Supports fro **Joe**





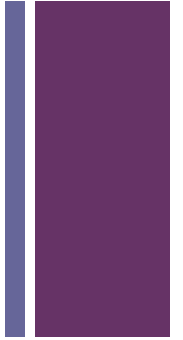
Discussion



- n Common needs (such as defined and organized spaces, sensory sensitivities)
- n Unique needs-to each student
- n Must know each student well-their likes/dislikes-level of detailed knowledge held by teachers-amazing
- n Adults could act in specific ways to prevent negative behaviors



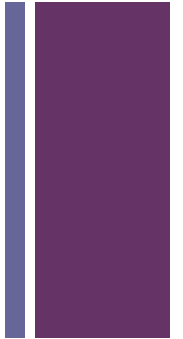
Discussion



- n Collaboration important-in our study students were known by all three teachers
- n Consistency important-yet must be willing to adjust in the moment
- n Profiles can be used to shape PBIS plans
- n Profiles are helpful to share across environments and for transitions to adult environments



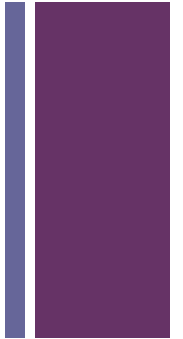
Study Limitations



- n Difficult to identify clear action research cycles (and connections between specific strategies: behaviors as instructional adjustments occurred frequently)
- n Study of just 7 students, 5 with CHARGE syndrome (including one who was not deafblind)



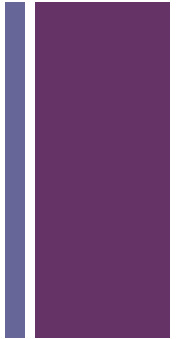
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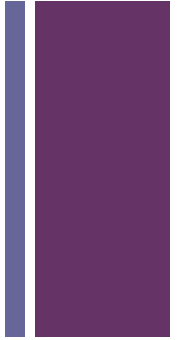
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Acknowledgements



Thank you to the following supporters of this research:

- n Michael & Susan Argyelan Education Research Fund
- n Perkins School for the Blind Research Fund



**Friday, July 31, 2015
Breakout Session #15: 2:15 - 3:15pm
Schaumburg C-D**

**Addressing Feeding Issues in CS:
Implications for Future Oral Motor and
Speech Development**

**Whitney Pimentel, MA, CCC-SLP
TalkTools**

Presenter Information:

Whitney B. Pimentel, MA, CCC-SLP is a Speech Pathologist that specializes in oral placement, feeding, and speech therapy for clients of all ages and ability levels. She has a private practice in Park City, UT and provides individualized evaluations for children with Oral Placement, Sensory, and Feeding Disorders. Trained under Sara Rosenfeld-Johnson, MS, CCC-SLP and member of the TalkTools speakers bureau.

Presentation Abstract:

Early feeding skills are critical in development of later oral motor skills, mature feeding skills, and speech clarity. Participants will learn what to look for in “good” feeding skills rather than simply “safe and adequate” feeding. Addressing these skills early is critical and will reduce later “bad habits” that evolve around spoon feeding, cup drinking, straw drinking, chewing and swallowing. However, these skills can also be taught in older children who have not had feeding and/or Oral Placement Therapy. Participants will learn techniques to facilitate lip closure, tongue retraction, tongue lateralization, and jaw grading for chewing. Families will also become aware of medical, environmental and sensory experiences and how they can alter feeding outcomes. Videos of both positive and negative experiences will be shown as part of the learning process.

Addressing Feeding Issues in CS: Implications of future Oral Motor and Speech Development

TALKTOOLS®
DISCOVER THE FEEL OF FEEDING AND SPEECH

Presented by Whitney B. Pimentel
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TalkTools Therapist
Whitisp@gmail.com

Disclosures

Content Disclosure: This presentation will focus on assessment and treatment of children with feeding, oral placement, and speech difficulties. Other treatment approaches will receive little to no coverage

Speaker Financial Disclosure-Whitney Pimentel is the owner of Discovery Therapy and is a member of the Talk Tools speaker bureau and receives a speaker honorarium.

Non Financial Disclosure-Whitney Pimentel has no relevant non financial relationships to disclose.

Common Characteristics in CHARGE Syndrome

1. Oral Hypo/Hyper/Mixed Sensitivity: feel/taste
2. Low muscle tone-oral hypotonicity: weakness in muscles of abdomen, velum, jaw, lips, tongue.
3. Significant gap between receptive and expressive language skills
4. Hearing loss
5. Vision impairment
6. Other Medical complications

Where do we start?

What is Sensory?



The Sensory System

1. **Tactile Hyposensitivity:** An under-reaction to tactile input.
 2. **Tactile Hypersensitivity:** An over-reaction to tactile input.
 3. **Mixed Sensitivity:** Any combination of hyper, hypo or normal sensitivity.
 4. **Fluctuating Tactile Sensitivity:** Responses that change over time.
-
- Tactile Defensiveness:** A learned tendency to respond negatively or emotionally to tactile input.

- Why is feeding so important to an oral-motor (oral placement) therapy program?

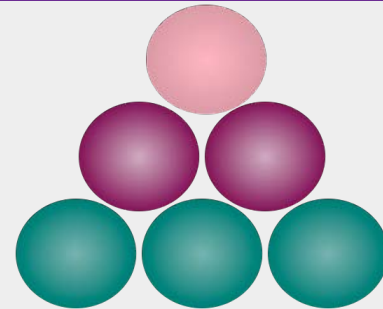
**Nutritional Concerns

**The muscles that are used in feeding are the same muscles that are used in speech.

- Why does my child bite and put everything in their mouth and avoid certain foods?
- Why does my child suck their thumb, grind their teeth etc?

Addressing Feeding Issues in CS

Before and After



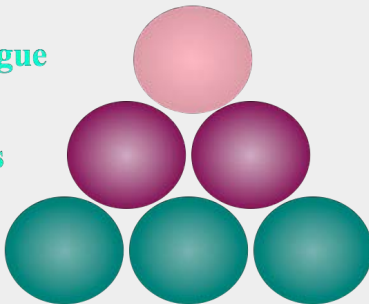
Addressing Feeding Issues in CS

Before and After

↑Tongue

↑Lips

↑Jaw



Addressing Feeding Issues in CS

Lip from Jaw Dissociation

- 1. Lip closure skills:** spoon feeding, cup drinking
/m, p, b/
- 2. Lip protrusion/retraction skills:** straw drinking, /oo,
w, ee, ih, eh, sh, ch, j
- 3. Lower lip retraction/tension**
Lower lip protrusion/tension
skills: remove food from upper/lower lip
/f, v, er/

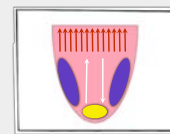
Addressing Feeding Issues in CS

Before and After

- 1. Suckle- Protrusion/Retraction**
skills: bottle feeding, breast feeding
no speech relationship
- 2. Tongue Retraction**
skills: swallowing, tongue at rest in mouth, all sounds except /th/, co-articulation, vocalic /er/
- 3. Tongue Tip Lateralization**
skills: moving foods side to side, keep a bolus on the molars during chewing no speech relationship
- 4. Tongue Tip Elevation/Depression**
skills: mature swallow, t, d, n, l, s, z, sh, ch, j, k, g

Addressing Feeding Issues in CS

Before and After



gue does not move as one unit. Each part independently mov

Addressing Feeding Issues in CS

Before you begin....

1. Establish a supported feeding posture: Stability in the body will allow for maximum mobility in the mouth.
2. Evaluate the sensory system: Motor skills are developed when a muscle receives sensory input telling it to move.

Sensory activities are recommended to facilitate activation of a specific muscle prior to using that **muscle**

Addressing Feeding Issues in CS

Tools used when Assessing the Oral Sensory System



Addressing Feeding Issues in CS

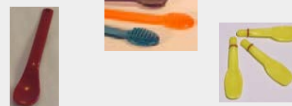
Typical Feeding Schedule

- **Spoon Feeding:** 4-6 months or as directed by your pediatrician
- **Cup Drinking:** 6-8 months or when spoon feeding is fully established
- **Straw Drinking:** 8-10 months or when spoon and cup drinking are fully established or earlier if therapeutically more appropriate for oral motor development.
- **Soft Solids:** 8-12 months or when the motor plan for chewing has been established.

Addressing Feeding Issues in CS

Therapeutic Feeding Techniques

- Spoon Feeding
 1. Lateral Placement
 2. Front Placement
 3. Pointed Spoon Slurp



Addressing Feeding Issues in CS

Therapeutic Feeding Techniques

- Goals: Lip closure (m, b, p) and Tongue Retraction for all sounds except /th/



Addressing Feeding Issues in CS

Liquids Optimal Feeding Tools and Techniques

- a. Cup Drinking
- b. Straw Drinking

Goals: Lip closure (m,b,p)
Tongue retraction for all sounds except /th/



Addressing Feeding Issues in CS

Why is Straw Drinking So important

- Straw Drinking

1. Honey Bear
2. TalkTools(R) Straw Hierarchy



Addressing Feeding Issues in CS

TalkTools(R) Straw Hierarchy #1-#4



#1



#2



#3



#4

Goals: Lip Protrusion, Tongue Retraction, Jaw Stability. Repetitive suck.
Last sip looks same as first. Able to drink 4 oz in less than 2 minutes.
Use only thin liquids.

Addressing Feeding Issues in CS

TalkTools(R) Straw Hierarchy #5-#8



#5



#6



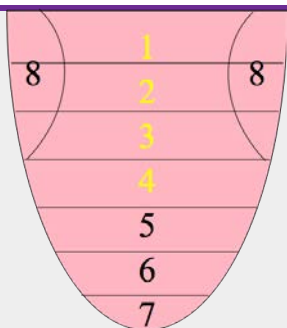
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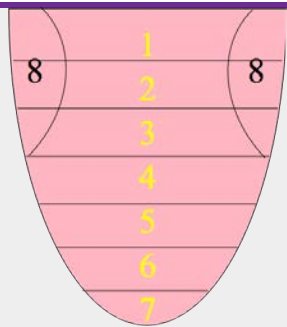
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Goals: Lip Protrusion, Tongue Retraction, Jaw Stability. Repetitive suck.
Last sip looks same as first. Able to drink 4 oz in less than 2 minutes.
Use only thin liquids.

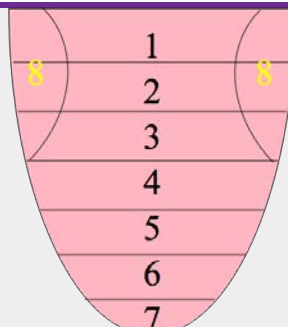
Addressing Feeding Issues in CS



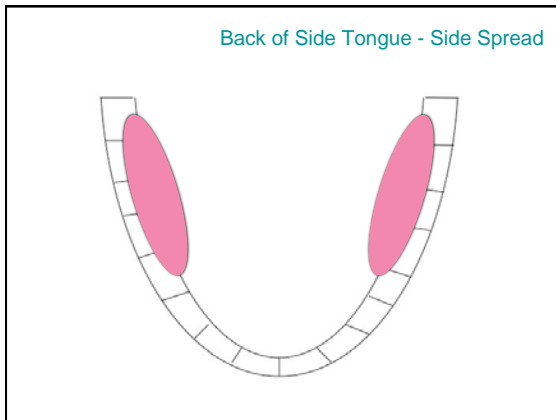
Addressing Feeding Issues in CS



Addressing Feeding Issues in CS



Addressing Feeding Issues in CS



Overview of Optimal Feeding Positions and Techniques

Solids:

1. Teach the motor plan for safe feeding of cubed solids, using non-food items.
 - a. Gloved Finger
 - b. Infadent
 - c. Ark Probe

Addressing Feeding Issues in CS

Overview of Optimal Feeding Positions and Techniques

- d. Ark's Z-vibe
- e. Bite Tubes (Red & Yellow)
- f. Ark's Grabbers (Purple & Green)
- g. Jaw Grading Bite Blocks

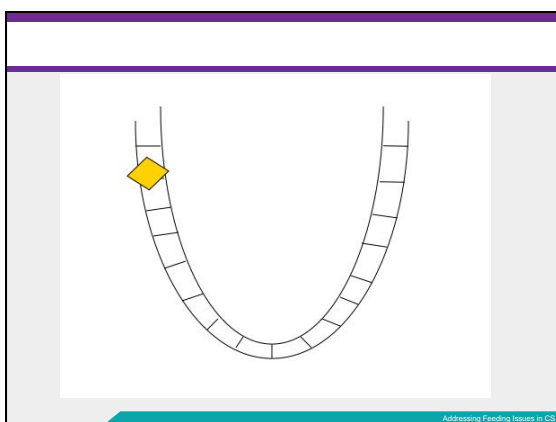
**Hierarchy as taken from *Assessment and Treatment of the Jaw*

Addressing Feeding Issues in CS

Solids

2. Chewing on Back Molars
 - a. Small Plate-Big Plate: Cocktail Fork Upside Down Position
 - b. Self-Feeding

Addressing Feeding Issues in CS



A Three Part Treatment Approach?

Addressing Feeding Issues in CS



What is a Pre-Feeding Program

GOAL: to develop the motor skills to support safe, effective, nutritive feeding.

- Clients that do not have adequate nutrition
- Clients that have poor speech clarity
- Program should start with a motor goal

A Sensory Motor Approach to Feeding: Lori L. Overland MS, CCC-SLP, C/NDT; Robyn Merkel-Walsh, MA, CCC-SLP

What is a Therapeutic Feeding Program

GOAL: to develop a set of techniques that support safe feeding and maximize a client's postural stability, muscle tone, sensory processing, and oral sensory-motor skills.

A Sensory Motor Approach to Feeding: Lori L. Overland MS, CCC-SLP, C/NDT; Robyn Merkel-Walsh, MA, CCC-SLP

Techniques for Pre-Feeding

<u>Motor Goal</u>	<u>Pre-Feeding Technique</u>
Bottle Feed	Tongue Bowling
Spoon Feed	Mickey Mouse Ear
Mastication of Solids	Gloved Finger

Why is this Effective?

- Consider the Sensory System
- Use of TACTILE Feeding/Oral Placement Method
- Feeding/Oral Placement Method
- Traditional Methods: Eclectic



**Friday, July 31, 2015
Breakout Session #16: 2:15 - 3:15pm
Schaumburg West**

ENT Issues in CHARGE Syndrome

**Daniel I. Choo, MD
Susan E. Wiley, MD
Cincinnati Children's Hospital Medical Center**

Presenter Information:

Dr. Choo currently serves as the Director for the Division of Otolaryngology Head and Neck Surgery at Cincinnati Children's Hospital Medical Center (CCHMC) and is also a Professor of Otolaryngology at the University of Cincinnati College of Medicine. He joined CCHMC in 1999 after completing his clinical Otolaryngology/Neurotology fellowship in Sarasota, FL and a research fellowship at the National Institutes of Health in Bethesda, MD. His work centers on ear and hearing disorders in children and is heavily focused on early communication skill development; including children requiring cochlear implants as well as children with congenital ear anomalies. Dr. Choo has become deeply engaged in the care of children with Charge syndrome (CS) and is co-directing (with Dr. Wiley) the development of an interdisciplinary CS Center in Cincinnati. Their efforts are directed at optimizing the coordinated care for children with CS and to develop the best practices for effectively navigating these children and their families through an increasingly complex medical course.

Susan Wiley is a developmental pediatrician in the Division of Developmental and Behavioral Pediatrics at Cincinnati Children's Hospital Medical Center. She has a clinical and research interest in children who are Deaf/Hard of Hearing Plus. She has worked in collaboration with the Ear and Hearing Center and Cochlear implant team with clinical evaluations of children who are Deaf/HH. She oversaw CCHMC's involvement in a national study on children who are deaf/blind with implants and works closely with the Ohio Center for Deaf-Blind Education.

Presentation Abstract:

The proper evaluation and management of Otolaryngologic (ENT) issues in the setting of children with CHARGE syndrome (CS) can pose a significant challenge. A multitude of clinical issues can range from choanal atresia to cleft lip and palate, to ear malformations and hearing problems, to subglottic stenosis. This discussion will focus on 3 of the main challenges (in the ENT domain) among children with CS.



**Friday, July 31, 2015
Breakout Session #17: 2:15 - 3:15pm
Schaumburg E-F**

CHARGE 102

**Nancy Salem-Hartshorne, Ph.D.,
Delta College**

Presenter Information:

Nancy is an Assistant Professor of Psychology at Delta College in Bay City Michigan and a School Psychologist. She has authored articles and book chapters about developmental outcomes for individuals with CHARGE syndrome. Her young adult son Jacob has CHARGE syndrome and lives in his own home. Nancy has been involved with language and education of children with CHARGE for 20 years. She advocates for individuals with disabilities, teamwork, thorough planning, and forward thinking for quality life outcomes for all individuals.

Presentation Abstract:

Nancy will present an overview of what we know about behaviors and developmental outcomes in CHARGE and introduce communication options

COMMUNICATION

Communication, communication, communication!!!!

After Medical/Health Issues...

- Enabling expressive and receptive communication for the individual with CHARGE syndrome is the most crucial goal. It underlies all other learning.

Language: What's so hard? Isn't it a Natural Process?

**Sure it was, for you and most of the people you know. But
Imagine...**

- you were just born into the world.
- you are hooked up to tubes, monitors, and pumps, some of which are painful.
- you have just been given a tracheostomy or gastrostomy.
- you have been in the hospital for weeks on end.
- you are recovering from surgeries, tests, and procedures, with pain and fatigue.
- your body isn't working right and you just plain don't feel good.

Now: How “ready” are you to learn?

Let's add a few things: Imagine...

- your vision is limited: You can only see through one eye, or part of one.
- your hearing is limited: You can't hear much from one ear, and nothing from the other.
- you cannot move to what you want to explore: Your muscles are weak; you can't tolerate the feel of the carpet/floor on your hands or feet.
- your sense of balance is limited or nonexistent. What you see moves around and you can't get oriented. Being held and carried may even cause you distress.
- all of this doesn't allow for growthful parent/baby interaction.

Now: How “available” are you to learn?

Factors affecting language development in CHARGE syndrome

(Adapted from Swanson, L., in CHARGE Syndrome, 2011)

- Vision and mobility/balance
- Hearing loss
- Facial palsy may cause difficulty with speech
- Tracheostomy: problems vocalizing
- Difficult to stimulate language early because of medical issues
- Language delays may affect cognitive skills
- Parent-child interactions: difficult when the child is less available because of sensory/medical issues.
- Serious illness and hospitalizations

Distance Senses

- Smell
 - Can identify a person
 - Can evoke strong memories
 - Is smell available? Not always, in CHARGE syndrome



Distance Senses

- Hearing
 - Important for understanding oral language
 - Also gives environmental cues
 - How much hearing is available? From one ear or both? How much does an ear infection affect this? Do they wear hearing aids or cochlear implants?

Distance Senses

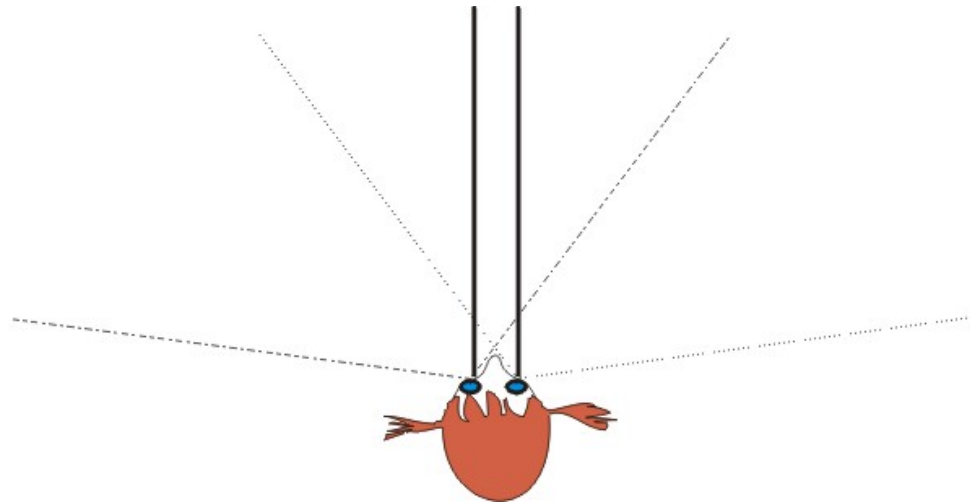
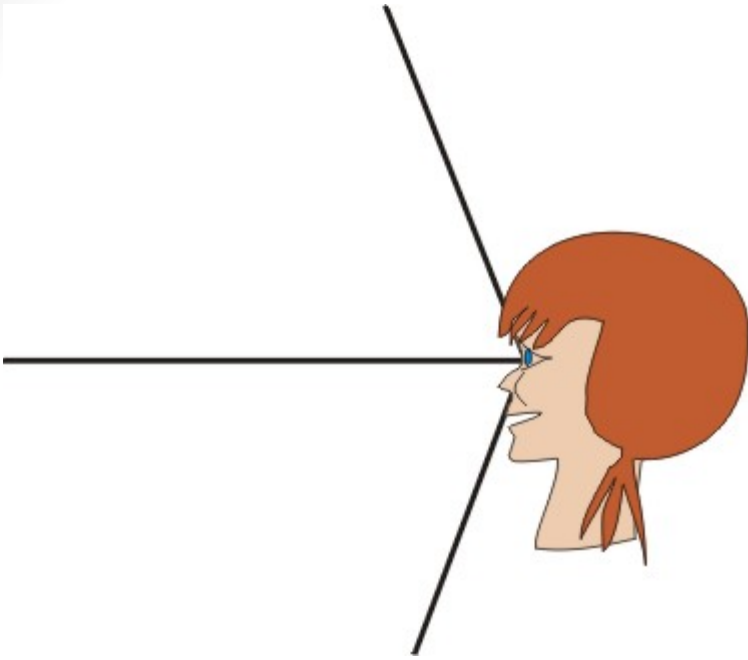
- Vision
 - Tells you what is in your environment
 - Tells you if things or people are moving toward you or away from you, or standing still
 - Tells you if people are trying to communicate with you through sign or gestures
 - How far up, down, and side to side can they see? Do they have blind spots? How far away can they see clearly? How does lighting affect their vision? What about contrast? These things may be able to be determined through a Functional Vision Assessment.

The Communication “Bubble”

“A child with CHARGE may not see you unless you are at a specific distance and in the individual child’s visual field, or the child may see only parts of you and not as a person. These children also may not hear your natural voice, or they may not hear you at all. These same children may not smell properly in order to identify food, perfumes, or other common odors and may not have enough balance to attain normal motor milestones. These children are *input impaired*.”

<http://www.dbproject.mn.org/commbubble.html>

Does your child use
postures that explain this?





What do you need to do to accommodate the individual person's communication bubble?

Communication/Language in Children with CHARGE Syndrome: Some data

Makes reactions or noises or behaviors which can be difficult to interpret	n =20	16.1%
Uses behaviors such as gestures, sounds, body movements	n = 12	9.7%
Uses single words, signs, picture symbols, or object symbols to represent basic needs	n =15	12.1%
Uses some 2- to 5-word phrases and sentences using speech, signs, picture symbols, etc.	n = 17	13.7%
Uses verbal or sign language in complete sentences	n = 59	47.6%

All children were 4 or older. Hartshorne, T.S., Unpublished Data

Communication Modes Used by Individuals with CHARGE Syndrome (Adapted from Swanson, L., CHARGE Syndrome, 2011.)

- Manual Sign

- *Sign Language*: The “official” language used by Deaf culture in your country
- *Signing Exact English*: Using English word order, instead of the “language” of sign
- *Cued Speech*: Using gestures while speaking to enhance understanding of lip-reading
- *Fingerspelling*: Spelling out words letter by letter using a signing alphabet
- *Tadoma*: a method of touching a person’s face and throat to feel what they are saying
- *Home sign*: Signs used/invented by an individual that are specific to them

- Speech
 - Using verbal forms of language, both formal and informal
- Visual Symbols
 - Using objects, pictures, or textures to communicate
- Voice Output Communication Aids (VOCA)
 - Using electronic equipment to communicate a message
- Gestures and Vocalizations
 - Pointing, showing, push-pull, vocal noises to communicate, without the use of signs
- Idiosyncratic Behaviors
 - The individual's own form of communication when other forms are not adequate, available, or accessible. Examples: crying, smiling, pain behavior, or any other way of trying to get a message across.

- Total Communication: recommended for ALL
 - The use of any form of communication available to the individual. Often, individuals choose to use more than one form.
 - For example, a person may use some sign language to answer questions, some picture symbols to ask for food, some push-pull to show you what he/she wants, and some idiosyncratic behaviors when in pain, because that is what works for them.
 - There is a growing consensus to allow for this to happen, as any communication helps the individual to be heard, no matter how it's done.
 - Limiting the individual to only one mode of communication may cause frustration as it blocks their ability to make themselves heard.

Some General Guidelines

- Let the child take the lead in communication: follow them. Get into their world; don't force them into yours.
- Turn-taking is an important first skill in communication, even if there are no words involved. How? Respond to signals to get a conversation started, just like with a developmentally typical baby; mimic sounds and gestures, extend them, expand them. Don't ignore them. All behavior is communication!

- Validate the individual's attempts at communication. Don't assume you know what they are going to say. Let them say it before you respond.
- Make sure the individual has access to all of their communication tools at all times, (all pictures, all devices) even if the answer to their request is "no." Never take away their ability to use their words, in whatever way they use them.

- Use total communication: Allow/encourage the individual to tell you what they need to tell you in any way they can.
- Ask for clarification if you are not sure what the individual is saying. Use a consistent signal/sign/word for “What?”
- Use calendar systems: help the individual to anticipate what will be happening during the time period. This helps to alleviate anxiety, to give them choices about their activities, and to understand time: before/during/after.

Behavior in CHARGE Syndrome

Common Deafblind Behavior

- **Eye pressing**
- **Finger flicking**
- **Rocking**
- **Tapping body/objects**
- **Self-injurious behavior**
- **Mouthing objects**
- **Tactile defensiveness**
- **Clinging**
- **Spinning**
- **Vocal tics**
- **Feces smearing**
- **Lining things up**
- **Extreme preferences**
- **Darting/running off**
- **Learned helplessness**
- **Submissive**
- **Stare at lights**
- **Inappropriate vocalize**

We see all of these in children with
CHARGE

-Beth Kennedy, DeafBlind Central

Tim Hartshorne (CHARGE Syndrome, 2011) has studied behavior and found that “certain similarities and patterns are emerging.” These could possibly related to the genetic differences in CHARGE syndrome. Not all individuals with CHARGE will have all features. However, these seem to come up often.

1. Low-normal cognitive functioning: There is a wide range of abilities, but Salem-Hartshorne and Jacob (2003) found that about half have low-average to average abilities, which was surprising, given that medical reports had often said all had mental retardation.

2. Very goal-directed and persistent with a sense of humor:

“Children with CHARGE syndrome seem to know what they want and persist in their intentions. This stubborn persistence helps them to learn to walk, to eat, and to achieve beyond all expectations.”

However, sometimes they have trouble letting go of the idea of what they want, and this can be a problem. At other times, they may be unable to communicate what it is they want, causing frustration and escalation of behavior. Happily, they often have a great sense of humor as well.

3. Socially Interested but Immature: Children with CHARGE syndrome often have trouble making and keeping friendships. At the same time, they seem to be more interested in relating to others than others who are deafblind or have autism. However, they often “have difficulty understanding other children and often miss or misunderstand social cues.” Part of it may be because of their problems with vision and hearing, but they seem to have difficulty with taking turns, backing off when necessary, and playing nicely with others.

4. Repetitive Behaviors that Increase under Stress: Many children with CHARGE are diagnosed with Obsessive Compulsive Disorder because of these behaviors. OCD is an anxiety disorder, and behaviors will increase with stress. The behaviors tend to soothe the anxiety. Whether or not individuals with CHARGE syndrome have OCD is still in question. Still, they often engage in repetitive behaviors, lining things up, and keeping strict routines. It seems to make sense that with limited sensory input, the world can be a bit chaotic. Using these behaviors may simply help to keep anxiety levels down.

5. High Degree of Sensation Seeking: “Light flicking, hand flapping, body shaking, and rocking are among the self-stimulatory behaviors that are common in children with CHARGE.” Although these behaviors can get in the way of learning at times, they seem to serve a function for individuals with CHARGE syndrome. Because they often lack sensory stimulation from vision and hearing, these behaviors may serve to make up for that lack, to keep the brain alert (just as you or I might tap a pencil, chew gum, or sway our calf back and forth to keep alert.) Some individuals with CHARGE may simply need a sensory break to pull themselves back together before working again. “A hard-working student might suddenly drop everything and shake his or her whole body for a moment, and then be able to go back to work.”

6. Under Conditions of Stress and Sensory Overload, Find it Difficult to Self-Regulate and Easily Lose Behavioral Control:

Most of us are able to self-regulate well. We can concentrate even though there's a little bit of noise in the background, or we know that when we are overwhelmed in a noisy environment we need to get someplace quiet. Everyone is different in what they can tolerate to their senses, and in how they deal with it. Individuals with CHARGE have difficulty regulating this.

Sometimes they may want extra stimulation, such as rubbing their hands on a textured surface repeatedly. Then, suddenly, it becomes too much. The sensation "might send them over the sensory edge and cause loss of behavioral control." They may crave the sensation of staring at a colorful disco light toy, but then knock the light away when it becomes too much.

7. Difficulty with Shifting Attention and Transitioning to New Activities; Easily Lost in Own Thoughts.

Many individuals with CHARGE syndrome become very focused internally. This may in part be because of sensory impairments, but there seems to be more. They seem to have difficulty shifting their attention, and “moving freely from activity to activity.”

Additionally some may have difficulty initiating new activity. They may want to, and may be ready, but might have difficulty getting started.

They may also have trouble with monitoring their own behavior and how it affects others.

These individuals will need assistance to learn to do these things.

Behavioral Threshold:

This is the point at which someone loses control. Consider anger in you or me. Most people may be able to control their behavior when they are angry, but then someone does or says something, and it's the "last straw," and they may lose control.

Likewise, an individual with CHARGE syndrome may hold it together as long as they are under the behavioral or sensory threshold. Once things become too much, they can have an abrupt change in behavior, seemingly coming out of the blue.

Three things can put an individual with CHARGE syndrome over the threshold for behavior:

1. Sensory stimulation: As stated before, what begins as a welcome need for stimulation can often escalate to be too much. It can change from “pleasant or bearable to painful and upsetting very quickly.”
2. Buildup of stress in the environment to the point that they can no longer cope. Consider all of the medical interventions they must go through, for example. In addition, consider the mere exhaustion factor when trying to see, hear, stand, walk, and all of the other things these individuals must work hard at, all day.
3. Pain: This may be the most important thing to look at. Often, pain is subjective. We cannot see it. A child with CHARGE may not be able to tell us that their throat or ear hurts, or that they feel achy. There are many other types of pain that are specific to CHARGE syndrome (see next slide). A sudden change in behavior without explanation should always lead to an immediate investigation of what may be causing the person pain.

Frequent Sources of Pain in CHARGE

- Otitis Media (middle ear infections)
- Sinus infections
- Migraines
- Abdominal Migraines (migraines felt in the belly)
- Gastroesophageal reflux disease (GERD)
- Gas
- Constipation
- Muscle pain
- Tactile defensiveness (too much sensation at the skin level)
- Stoma pain (inflammation around the skin at the entrance of the feeding tube in the stomach.)

Why pain is so important

- **Can affect normal brain and nerve development**
- **Can affect sleep**
- **Can interfere with exploration of the environment and learning**
- **Can interfere with the development of attachment and trust**
- **If they begin to get used to it, it may affect their health if not taken care of.**

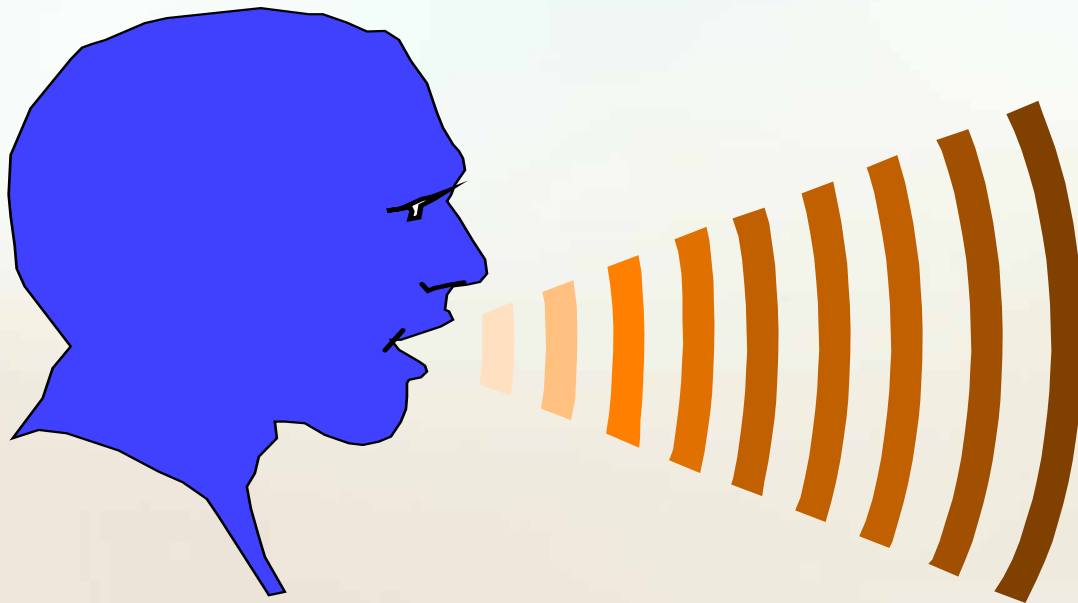
(Hartshorne, T.S. 2012)

When you are in pain, do you ...

- **Whine?**
- **Complain?**
- **Seek attention?**
- **Make demands?**
- **Act out?**



ALL BEHAVIOR IS COMMUNICATION



Sometimes behaviors make perfect sense!

This is normal viewing posture...

...when you have
no vestibular
sense, upper
visual field loss,
poor tactile &
proprioceptive
perception, & low
muscle tone.

(-David Brown)



How can you help?

- **Communication, communication, communication**
- **Always interpret behavior as communication.**
- **Develop a pain management plan.**
- **Figure out if the behavior serves a purpose for the individual. It doesn't always have to be changed or eliminated!**
- **If it is determined that the behavior should be changed or eliminated (maybe they are injuring themselves or others), we must teach them alternative ways to communicate their needs.**
- **Sensory diet: deep pressure/squeeze hugs, brushing and joint compression (ask your occupational therapist for help). These things can help regulate the sensory and behavioral thresholds by getting the nervous system reorganized.**
- **Break down larger tasks into smaller, more manageable chunks.**
- **Help the individual learn ways to self-soothe.**
- **Build sense of safety and trust**
 - **Consistency**
 - **Routine**
 - **Calendar System**

EXAMPLES

David Brown (2005).
CHARGE Syndrome
“Behaviors”: Challenges or
Adaptations? *American
Journal of Medical
Genetics, 133A*, 268-272.



. A young child who was said to be self stimulating "all the time" was actually practicing and developing his mobility and orientation skills, and using vision and touch to explore objects, very creatively. While doing this he needed to get onto his back on the floor to reorganize his sensory system with brief episodes of limb shaking and hyperventilating every 10 to 20 minutes.

(sensory break)

- A kindergartener was often self-abusive when he got distracted and over-aroused by incidental touch and air movement caused by people repeatedly walking behind his chair. Once his chair was placed with its back securely against a wall he was less self-abusive and more amenable to social interaction. (Too much stimulation)

. People were concerned when a young boy began to insist on the unusual idea of wearing band-aids wound tightly around the tips of all his fingers and thumbs every day. He was expressing his need for more and stronger pressure and touch inputs as a part of his sensory diet. (Needed more pressure/stimulation)

. Every morning in a pre-school program a student refused to sit on the floor with her class to watch the teacher sign a story. When an appropriate chair was provided the student sat and attended with great interest and a growing level of participation. (balance problems)

. A girl was described as very disruptive during sessions that required the class to sit still and participate in a signed conversation with the teacher for up to 30 minutes. When the teacher used a strategy of asking the student to move periodically to carry out small chores during these sessions (to fetch a pen, open a door, bring a book, take a paper to the school office) the disruptive behaviors largely ceased. (visual fatigue)

. A teenager enjoyed, and was quite good at, soccer in the school gym, but was unwilling or unable to play it outdoors due to the absence of strong vertical visual markers to aid equilibrium. The ability to participate in such complex physical activities outdoors did not develop until significant adaptations were introduced. (need for visual “handles” in the environment to know where his body is in space)

. During Orientation and Mobility sessions a teenager was refusing to stand still to receive spoken/signed instructions, but the problem was solved when the student was allowed to stabilize himself by leaning against a pole or a tree or a wall, or by placing one hand on the instructor's shoulder during these conversations.

(balance problem/stabilizing body/head)



Developmental Considerations

Three Developmental Domains

- › In studying human development, we look at three domains:
- › Physical Development
- › Cognitive Development (Intelligence)
- › Social/Emotional Development

Physical Development

- › We've already discussed the physical differences in individuals with CHARGE syndrome. We have discussed senses, sensory systems, surgeries, medical problems, and many other aspects. What's left?
 - › **Motor Milestones**

Normal Gross Motor (Large Muscles) Milestones

- › What's typical for a non-CHARGE baby?
 - › Hold head up
 - › Roll over
 - › Sit up
 - › Crawl on belly, then on all fours
 - › Pull to stand
 - › Cruise holding onto furniture
 - › Walk independently (Average Age=12 months)
 - › Stairs, hopping, running, skipping, etc.

CHARGE Gross Motor Milestones

- › Arching back
- › Back scooting (rub hair off back of head)
- › 5-point crawl (four limbs plus head)
- › Sitting
- › Bottom Shuffling
- › Crawl
- › Pull to Stand
- › Cruise around furniture
- › Walk independently (average age = 42 months)
 - › (Age ranges from 30 to 84 months)
- › May continue to have unsteady gait

With what you know...

- › About sensory, vestibular, and other CHARGE systems, can you hypothesize why these milestones might occur differently?
 - › Visual field reasons?
 - › Vestibular reasons?
 - › Keeping head stable reasons?
 - › Tactile defensiveness reasons?

Vestibular

- › We use eyes, joint pressure, and semi-circular canals to balance. Many children with CHARGE have little vision, and malformed or missing semi-circular canals. In order to walk, they must rely on joint pressure, primarily. Does this explain the wide, slow, toddling gait many have?

Normal Fine Motor (small muscles) milestones

- › Suck, swallow
- › Eat puree
- › Chew, bite
- › Grasp with whole fist
- › Grasp with finger and thumb
- › Manipulate/trade objects between hands
- › Hold/draw with crayon, etc.

With what you know....

- › About eating, tactile defensiveness, and low muscle tone, what might this mean about fine motor milestone achievement for children with CHARGE syndrome?

- › Early tactile defensiveness (inability to tolerate some sensory input to skin) limits how much touch and object manipulation the child can handle without going over the “sensory threshold.” This may delay fine motor milestones.
- › Of course, eating/swallowing/breathing issues will delay eating milestones.

Cognitive Ability

- *We gave the Adaptive Behavior Evaluation Scale to 100 Children with CHARGE*
- They showed a higher range of ability than once thought: 54 had scores higher than 70 (Average score is 100. Below 70 is typically considered mental retardation)
- Those who walked earlier, had fewer medical problems, and had better hearing and vision scored higher. Age at walking had the strongest result.

Salem-Hartshorne N, Jacob S. 2004. Characteristics and development of children with CHARGE association/syndrome. *Journal of Early Intervention Vol 26: 292-301.*

What does this mean?

- › It means that **we can/should have high expectations** of these children from the beginning, because we absolutely cannot tell what the outcome might be, based alone on how sick they are as babies.
- › It MAY mean that we can possibly predict how well a child will do based on when they start walking. (This is definitely not set in stone, but has been found in other studies as well.)

Age at Walking? Why?

- › The ability to walk depends a great deal on the vestibular system. Some children may have more severe problems there, and therefore walk later.
- › It's not walking age that causes problems, though. The vestibular system can affect a LOT of things that have to do with learning. Walking is just one thing it delays.
- › So, does having more problems with the vestibular system mean the child may have more developmental problems, overall?
- › It's what we are beginning to think, and it makes sense, but there is no clear proof of this.

Social Skills Deficits

- › Acquisition deficit - never learned the skills
- › Performance deficit - prefers not to use the skills
- › Fluency deficit – knows the skills but not very good at applying them

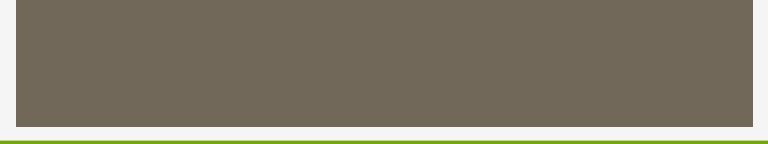
Most children with CHARGE appear to have a fluency deficit rather than the other two.

This means:

- › They know what to do.
- › They want to do well.
- › They have trouble doing it well.

Social Skills they Need

- › Getting along with peers
 - › Managing their behavior
 - › Classwork skills
 - › Cooperation
 - › Being assertive
-
- › They will need practice, guidance, and support to do well at these things.

- 
- › Management of emotions can be difficult, because of that emotional threshold that is difficult to come back from. Melt-downs may be common.
 - › Need to teach them strategies for self-soothing, talking to themselves about what's going on, and recognizing when they are approaching the threshold.
 - › How much is emotional management associated with sensory overload? We do not know. But sensory breaks seem to help.

ATTACHMENT/BONDING



Infant Attachment Problems

- › It's difficult to attach when there is little to no eye contact, and no hearing of one another's voices, between parent and baby. Add in tactile defensiveness (not wanting to be touched), and it can become very hard.
- › Medical procedures, and just trying to stay alive can have a huge effect as well.
- › The shock of having a baby with a disability can also play a part.
- › Many parents report delayed bonding with their child with CHARGE syndrome.

Reda, N. M. & Hartshorne, T. S. (2008). Attachment, bonding, and parental stress in CHARGE syndrome. *Mental Health Aspects of Developmental Disabilities*, 11, 10-21.

Toileting

Salem-Hartshorne, Blake, and Hartshorne (CHARGE Syndrome, 2011) surveyed information for 44 children with CHARGE, ages 9-21.

Independence with urine 73%
The rest needed assistance, reminders, or did not use the toilet

Independence with bowel 65%
The rest needed assistance, reminders, or did not use the toilet

Independence with overnight 54%
The rest needed protective undergarments, reminders, or physical help.

WHY?

Prerequisites for Toilet Training

Denno, L. CHARGE Syndrome, 2011

- Feel the need to eliminate
- Walk to bathroom
- Pull down pants and underpants
- Sit on toilet
- Eliminate in toilet
- Use toilet paper
- Put toilet paper in toilet
- Stand up
- Pull up underpants and pants
- Flush toilet
- Wash hands
- Dry hands
- Exit bathroom

For what reasons might these skills be difficult for an individual with CHARGE syndrome?

Balance/mobility (if you can't balance, it's difficult to undress standing up.

Can the individual button/snap/zip?

Inability to hear urine stream

Inability to smell feces

May not be socially motivated to try

There is some discussion of the possibility that they may have limited sensation because of problems with the nerves or muscles

Finally, they will likely get a late start because of all of the medical/physical problems (walking late, for example)



So....

Toilet training is likely to happen later with individuals with CHARGE.

Some may never be totally independent.

It is still a really important teaching opportunity, in terms of communication, turn-taking, and development of independence.

Why?

- Deaf-blindness: It is well-known that darkness cues the brain to release melatonin, a hormone that causes sleepiness. If you are blind or visually impaired, the darkness may not work as a cue. Therefore, is the melatonin released? Some parents have found over-the-counter melatonin to be helpful in inducing sleep, pointing to a possible deficit.
- Pain from ear infections/other problems keep child awake
- Behavioral difficulties and anxiety (think of that threshold!) leading to troubles settling at night
- Obstructive sleep apnea: Large tonsils/adenoids, choanal atresia, clefts, and other physical anomalies may get in the way of breathing while asleep. This can “fragment” sleep, so that good quality sleep is not obtained, night after night.

Effects of too-little sleep

Think about how you feel when you haven't had sleep.

Some of these children may be having that effect compounded by having fragmented sleep night after night.

Fragmented sleep means they may never enter the Rapid Eye Movement stage....this is when protein is synthesized, dreaming takes place, and the brain regenerates.

Do you think there are behavioral effects of exhaustion?

What to do?

(Heussler, H. S., CHARGE Syndrome, 2011)

- First, take care of obstructions. Surgery to remove tonsils and adenoids, or other medical management may help.
- Teach good sleep hygiene. This includes using a regular bedtime and routines (bath, read a book, teddy). The child will begin to associate these things with getting sleepy.
- Pay attention to sensory preferences. Sometimes a heavy blanket, temperature, or other things will help a child with sensory integration difficulties settle in.
- Try to make dark/light, day/night contrast as clear as possible.

ADOLESCENT DEVELOPMENT

The CHARGE Experience



Kim Blake, M.D.



Nancy Hartshorne, Ph.D.

Adolescent and Adult Issues


Blake KD, Salem-Hartshorne N, Abi Daoud M, Grandstein J. 2005. Adult and adolescent issues in CHARGE syndrome. Clinical Pediatrics. 44:151-159.

q 30 participants

q 16 female; 14 male

q Age range 13 to 30 years, mean 17.6





Medical Issues Found	Number	%
Scoliosis	19	63
Sleep Apnea	13	43
Abdominal Colic (Gas Pain)	12	40
Retinal Detachment/Cataract	10	33
Migraines	8	27
Seizures/Epilepsy	5	17
Urinary Tract Infections	5	17
Hypoglycemia	1	3

Osteoporosis also common

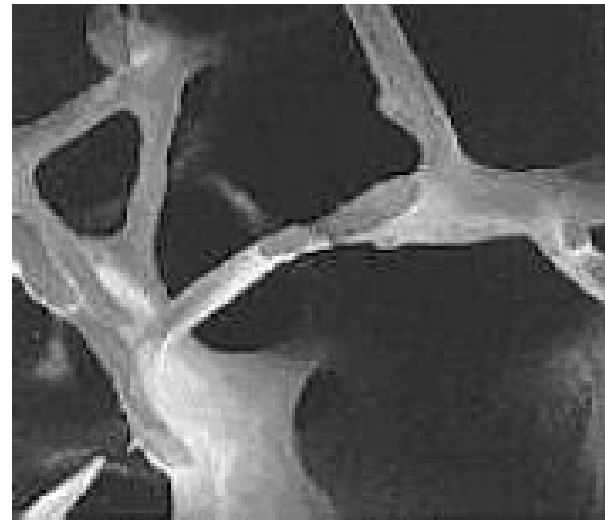
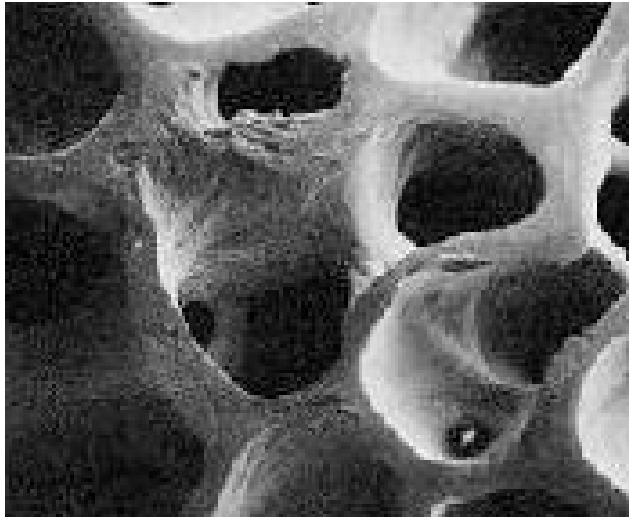
- › 15 males and 15 females, ages 13 to 34 (average 19.6)
- › Traumatic bony fractures were identified in 30% of the sample
- › Delayed puberty was experienced by 87% with only 4 individuals (2 female, 2 males) having experienced normal puberty
- › Feeding difficulties, inactivity and delayed puberty are all problems that lead to the development of poor bone health in CHARGE syndrome.

Forward KE, Cummings EA, & Blake KD. (2007). Risk factors for poor bone health in adolescents and adults with CHARGE syndrome. *American Journal of Medical Genetics. Part A.* 143A(8), 839-45.

Osteoporosis

Osteoporosis, or porous bone, is a disease characterized by low bone mass and structural deterioration of bone tissue, leading to bone fragility and an increased susceptibility to fractures, especially of the hip, spine and wrist, although any bone can be affected.

Healthy and Unhealthy Bones



Puberty Development

- Often delayed
- Girls tend to go into spontaneous puberty more often than boys, although it is usually delayed.
- Boys will often need hormone treatments to go through puberty.
- It is very important, medically, to go through puberty, to avoid osteoporosis. During puberty, we lay down bone mass.

Behavior can still be a difficult area in Adolescence

Behavioral Concern	Number	%
Aggressiveness/outbursts	16	53
Self-abuse	15	50
Sleep problems	15	50
Tactile Defensiveness	12	40

How independent were the kids in the study?

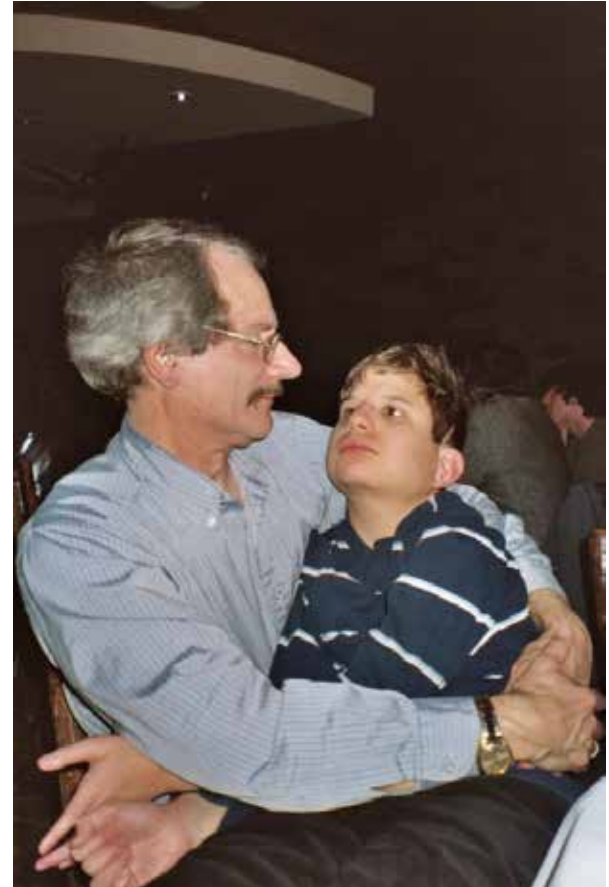
Degree of Independence (N=30)

Activity	Degree of Independence					
	None	Little	Some	Most	All	N/A
Dressing self	10		2	5	13	
Toileting self	10		4	2	14	
Washing self	10	4	3	5	8	
Getting to/from work/school	15	1	2	3	6	3
Cleaning	12	4	7	3	3	1
Cooking	16	5	7			2
Shopping	19	3	3	2	2	1
Finances	25	2	1		1	1

Can you see that there is a bit of a split? About a third have lots of trouble. The rest are learning or are somewhat independent. This gets again at the 50% who have low normal to normal intelligence.

Final Words from David Brown

"There is no other identified sub-group within the population of people with multi-sensory impairment who have so many medical problems, of such complexity and severity, and with so many hidden or delayed difficulties, and yet no sub-group has shown such a consistent ability to rise triumphantly above these problems."





**Friday, July 31, 2015
Breakout Session #18: 2:15 - 3:15pm
Schaumburg G**

The Importance of Movement in the Life of an Individual with CHARGE Syndrome

**Elizabeth (Beth) Foster, ABD, CAPE
Texas Woman's University**

Presenter Information:

Beth Foster is a Certified Adapted Physical Educator. She graduated from West Chester University in Pennsylvania with a bachelors in Kinesiology and a minor in adapted physical education, coaching, and health education in 2006. She began working as an Elementary Adapted Physical Educator at the School District of Lancaster that same year. During her time working in Pennsylvania Beth was named the 2012 Pennsylvania State Association for Health, Physical Education, Recreation and Dance adapted physical education teacher of the year. Beth has been involved in Camp Abilities which is a developmental sports camp for individuals who are blind, visually impaired or deafblind for the past 11 years. She is currently the assistant director for Camp Abilities in Pennsylvania and Texas and a dual sports coach at Camp Abilities in NY. From her experience at Camp Abilities with individuals who are deafblind, she developed a passion and determination to be knowledgeable within the field of deafblindness. Beth has completed the Minnesota Low Incidence/Deafblind Project Intervener training sessions. She also volunteered at the previous CHARGE International Conference by assisting to run the Camp Abilities. This year, she is currently in charge of providing Camp Abilities to the individuals throughout the CHARGE International Conference. Currently, she is working on her PhD in Adapted Physical Education at Texas Woman's University focusing on students who are deafblind.

Presentation Abstract:

The purpose of this session is to provide information about the importance and benefits of movement throughout the life of an individual with CHARGE Syndrome. The session will provide strategies to develop meaningful and accessible movement activities. A systematic approach based on best practices will be used to develop appropriate adaptations and explore with movement. Participants will understand the definition of physical education, student rights, and options for instruction to develop a positive, meaningful experience.

The Importance of Movement in the Lives of Individuals with CHARGE Syndrome

12th International CHARGE Syndrome Conference

Elizabeth “Beth” Foster, MS, ABD, CAPE

Session Purpose:

- Provide information about the importance and benefits of movement
- Provide strategies to develop meaningful and accessible movement activities
- Develop appropriate adaptations and explore with movement
- Understand motor development and learning and the impact movement has on individuals with CHARGE Syndrome.
- Understand the definition of physical education, student rights, and the difference between services and placements

Why is physical activity and movement important to individuals with CHARGE syndrome?

Individuals with CHARGE Syndrome may have:

- Delay in developmental milestones
- Display delayed motor skills
- Lower levels of fitness
- Balance and stability problems
- Lower levels of stamina/endurance
- Less opportunities to participate in movement and physical activities

There are a number of barriers to overcome for participation and opportunities in physical activity:

- Attitudes: Parents, Instructors (community), and students
- Professional Preparation
- Transportation
- Time
- Funding
- Knowledge of individual's needs
- Program availability
- Equipment availability

Sources: (Conroy, 2011; Lieberman & Houston-Wilson, 1999; Lieberman, Houston-Wilson, & Kozub, 2002)

With opportunities and participation in physical activities and movement, one can increase the following:

- Social Skills, Self-esteem, and Self-determination
- Common bond, Sense of belonging, and a feeling of Ownership
- Perceptions
- Independence
- Orientation and Mobility Skills
- Physical endurance and fitness
- Spatial awareness
- Gross motor skills

Individuals with CHARGE syndrome may have a delay in motor development due to:

- Vision and Hearing loss
- Vestibular Dysfunction
- Muscular weakness
- Orthopedic disorders
- Hospitalization and Surgeries
- Balance and Posture

Sources: (Dammeyer, 2012; Hartsthorne, Hefner, Davenport, & Thelin, 2011; Hartsthorne, Nichols, Grialou, & Russ, 2007; Pogrund & Fazzi, 2002; Salem-Hartshorne & Jacob, 2004; Smith, Smith, & Blake, 2010; Thelin, Curtis, Maddox, & Travis, 2007; Travis & Thelin, 2007; Williams & Hartshorne, 2005)

In research age of walking in individuals with CHARGE syndrome has been

- LINKED to
 - Communication
 - Language
 - Adaptive Behavior Scores (Intelligence)

Sources: (Dunlap, 1985; Hartshorne & Cypher, 2004; Petroff, 2001; Salem-Hartshorne, 2003; Salem-Hartshorne & Jacob, 2004; Thelin and Fussner, 2005)

Physical Activities and Movement that need to be provided:

- Various positions and postures
- Reach, grab, and release
- Push and pull- Proprioceptive input
- Kick and strike
- Cross midline and body awareness
- Use of auditory and vision
- Tactile surfaces

4 Key Areas to Movement & Learning to keep in mind:

- Anticipation
- Motivation
- Communication
- Confirmation

Introducing Movement and Physical Activities:

- **Allow time for exploration.**
- **Demonstrate.** Demonstrate movement/equipment and allow tactile modeling
- Perform the activities **together.** (coactive movement/imitation)
- **Follow the individual.** Student centered
- **Add rhythm** (turn-taking)
- **Use residual hearing and vision** to aid in learning
- Provide **confirmation and feedback**
- Match movement with a particular **object or symbol**

Include Common Language

- Be able to identify objects
- Make choices and have control
- Build meaningful vocabulary
- Based on the child's level
- Link language to objects/equipment
- Link objects/equipment to movement, game, or activity
- Gain further knowledge about same and different

Additional strategies include

- Pre-Teaching
- Motivation
- Guidance: Hand under Hand

Developing Appropriate Physical Activities and Movement- Provide adaptations when needed

The Game/Activity

- Reduce repetitions or slow the pace
- Change the rules of the game
- Change the objective of the game
- Change the number of players
- Decrease time of activity or add rest periods
- Increase tactile cues
- Add guidance or a leader
- Use partners
- Increase chances
- Provide visuals

Playing Area/Environment

- Area larger/smaller
- Add visible boundaries
- Increase/decrease the height/width of the goals
- Orient the individual to the activity area

Playing Object

- Larger or smaller
- Softer or harder
- Heavier or lighter
- Texture of the object
- Audible or bright
- Increase the size of the target

The Players

- Change the role of the players
- Limit or add responsibility
- Modify demands on the student
- Decrease competition
- Relationships

Information Needed about Physical Education:

The Four Components of a High-quality Physical Education Program

- Opportunity to Learn
- Meaningful Content
- Appropriate Instruction
- Student and Program Assessment

Why is Quality Physical Education Important?

Quality physical education programs help all students develop:

- Health-related fitness,
- Physical competence,
- Cognitive understanding,
- Positive attitudes about physical activity
- So that they can adopt healthy and physically active lifestyles.

Learning Experiences that Meet Developmental Needs.

- Quality physical education programs provide learning experiences that improve mental alertness, academic performance, and readiness and enthusiasm for learning in our nations' youth.

(Source: www.SHAPEAmerica.org)

Individuals with Disabilities Education Improvement Act, 2004: Physical Education

- The Individuals with Disabilities Education Act (IDEA), Public Law 108-466 (2004), states that physical education is a required service for children and youth between the ages of 3-21 who qualify for special education services because of a specific disability or developmental delay.

“Physical Education means the development of:

(A) Physical and motor fitness; (B) Fundamental motor skills and patterns; and (C) Skills in aquatics, dance, and individual and group games and sports (including intramural and lifetime sports); and (ii) Includes special physical education, adapted physical education, movement education, and motor development.”

“(3) Specially-designed instruction Means adapting, as appropriate to the needs of an eligible child under this part, the content, methodology, or delivery of instruction- (i) To address the unique needs of the child that result from the child's disability; and (ii) To ensure access of the child to the general curriculum, so that he or she can meet the educational standards within the jurisdiction of the public agency that apply to all children.”

Individual Education Program (IEP):

If specially designed physical education is needed

- IDEA mandates APE goals and objectives in IEP
 - Current level of performance
 - Measurable goals and objectives
 - Placement options
 - Assessment procedures
 - Accommodations or adaptations

Physical Education Options

- Least Restrictive Environment
 - Where a student can work **successfully** and **safely** on the general curriculum and individual goals

- Continuum of Placements and Services
 - General Physical Education
 - With consultation and accommodations
 - Integrated Physical Education
 - Peer tutors, small group
 - Adapted Physical Education
 - Small group, modified curriculum, one-on-one

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**Friday, July 31, 2015
Breakout Session #19: 2:15 - 3:15pm
Schaumburg H**

**Expect ExtraOrdinary: Using Vision,
Planning, and Community to Expand
Possibilities**

**Aubrie and Michele Westmaas
Expect ExtraOrdinary, Inc.**

Presenter Information:

Aubrie Westmaas was initially diagnosed with CHARGE syndrome. After attending every CHARGE Conference for her first ten years, her diagnosis was changed to Kabuki syndrome in 2007.

Since Aubrie's birth, Michele Westmaas has worked for several state advocacy organizations and was awarded Guide By Your Side 2011 Parent Achievement Award. As Central/Southern Illinois Organizer for Illinois Association of Microboards and Cooperatives, she facilitates person-centered planning sessions, supports families to organize personal networks, and empowers individuals to find their place in their communities.

Determined to enjoy regular opportunities with her peers, Aubrie has participated in a variety of community activities, performing arts camps and classes, has been a record cookie-selling Girl Scout, takes voice lessons and is part of the high school drama club, Quiz Bowl team, marching band, and chorus. She was named 2014 Illinois Statewide Transition Student of the Year.

Aubrie says, "People with disabilities need to be treated ordinary. But we do have some extras." Michele and Aubrie seek to empower all people with extra challenges to live extraordinary lives.

Presentation Abstract:

Individuals with CHARGE syndrome face a myriad of physical and sensory challenges. However, society's limiting view of people with disabilities can be the greatest barrier to achieving their ultimate potential. Aubrie and Michele share their experience and perspective to empower participants to envision a future full of possibilities, share their dream with others, plan for positive outcomes, and purposefully make their place in the community.

Expect ExtraOrdinary: Using Vision, Planning, and Community to Expand Possibilities

CHARGE Syndrome Conference

July 2015

Aubrie and Michele Westmaas

www.ExpectExtraOrdinary.org

Everyone CAN be ExtraOrdinary!

“People with disabilities
need to be treated ordinary
but they do have some
extras.”

Aubrie Westmaas

2

Meet Aubrie...



Aubrie belongs –

- In class
- In band
- In drama
- In chorus
- In church
- In Girl Scouts

....Everywhere her peers
belong!

3

**What if she'd been born
generations ago?**



This is
Margaret
as a baby.

4



By the time Margaret was 2 years old, she was not progressing at a typical rate.



At age 2, Margaret was sent to Dixon Developmental Center. Her siblings never knew she existed.

What if...



A new life...



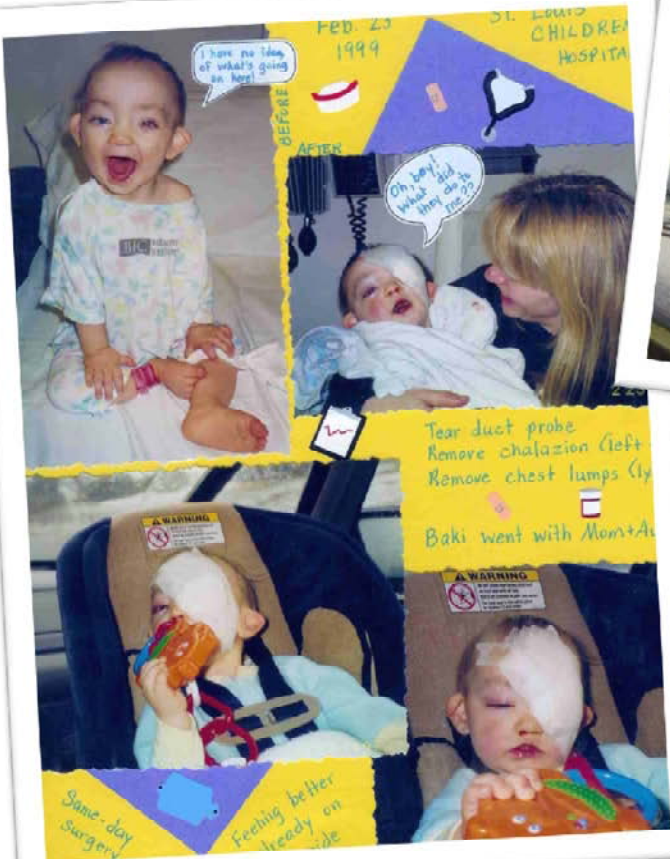


What would you do?

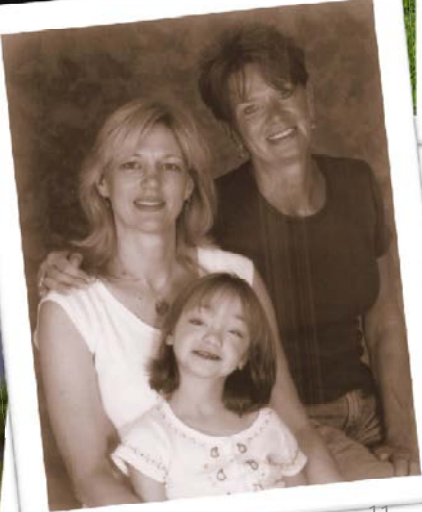
***Search
“Parallels in Time”
to learn more
about disability
history.***



Surgeries and therapies...



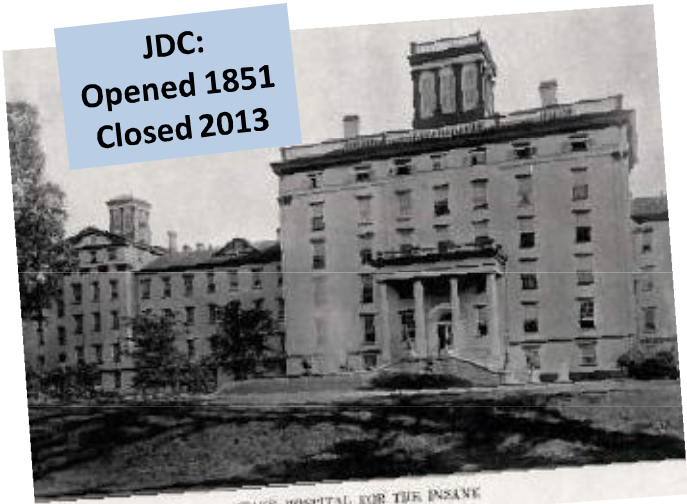
Family...







We've come a long way...
but we have a long way to go...



JDC:
Opened 1851
Closed 2013

Feb. 2013:
Robert Saylor
wanted to see the
movie again and
wouldn't leave the
theatre. He died
when police
intervened with
force.



Last Year: Mollie Cates
was not allowed to join
the local gym with her
family because she has
Down syndrome.

Last Year: Bonnie
Burch, Oregon 3rd
grader, must leave
school grounds for
her tube fed lunch.





**Breaking
Barriers!**



15

Progress Toward Community Inclusion

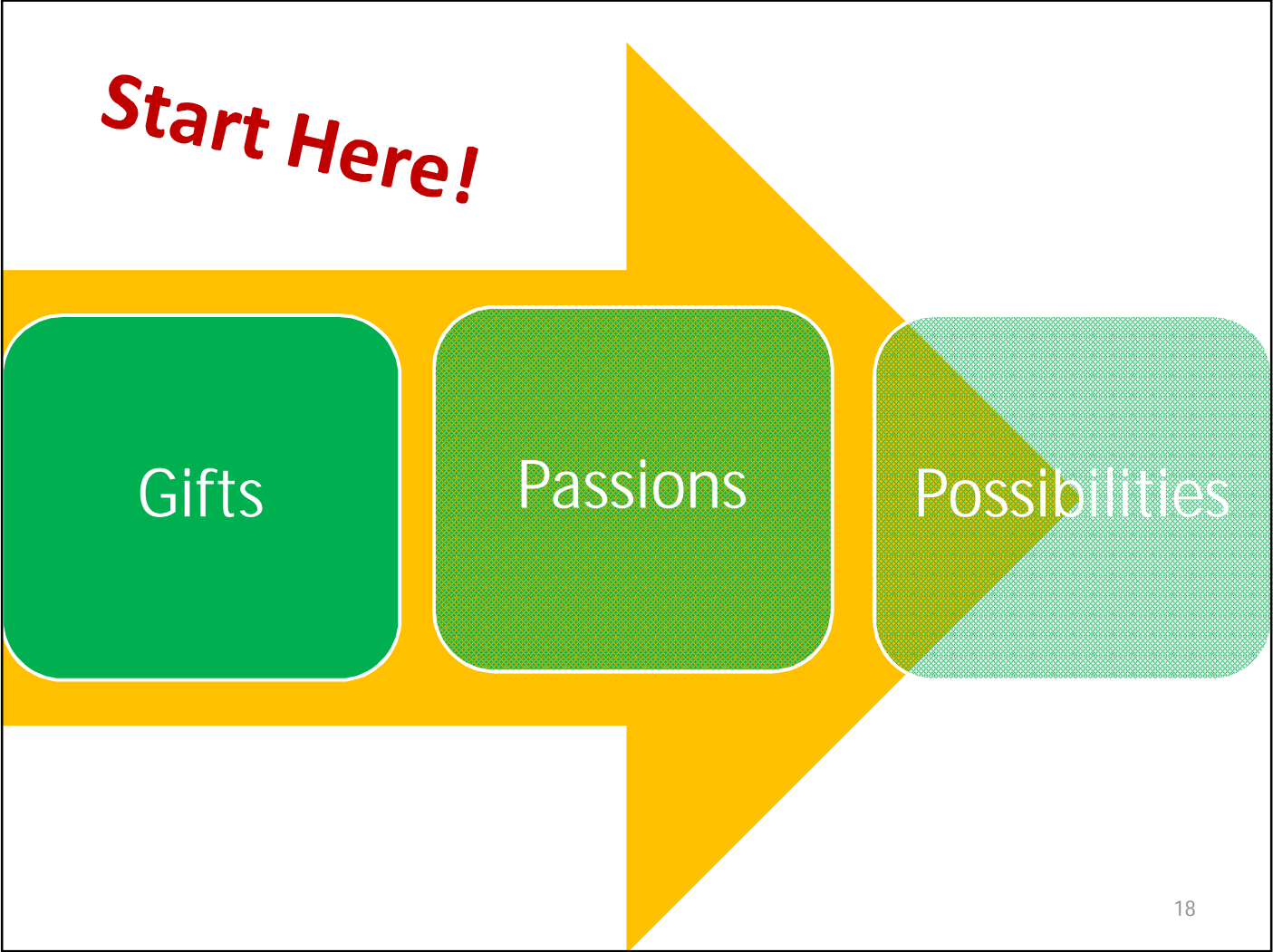
- Federal Medicaid regulations include specific requirements for home and community based services.
- Employment First is a national priority to invest in integrated community employment for individuals with significant disabilities.
- Person Centered Planning is replacing traditional forms and filling 'slots'.

16

Everyone IS ExtraOrdinary!

Aubrie said,
“Mom, it’s not about that
I’m extraordinary.
Everyone needs to know that
they are extraordinary too!”

17



To Support Big Dreams...

DESTINATION	HOW TO GET THERE
Nowhere	Road
Anywhere	Midnight Train
Georgia	Midnight Train
Clarksville	Last Train
Heaven	Stairway
Hell	Highway
Hotel California	Dark Desert Highway

you have
to know
which
road to
take!

What's The Vision?



No Bubble-Bursting!

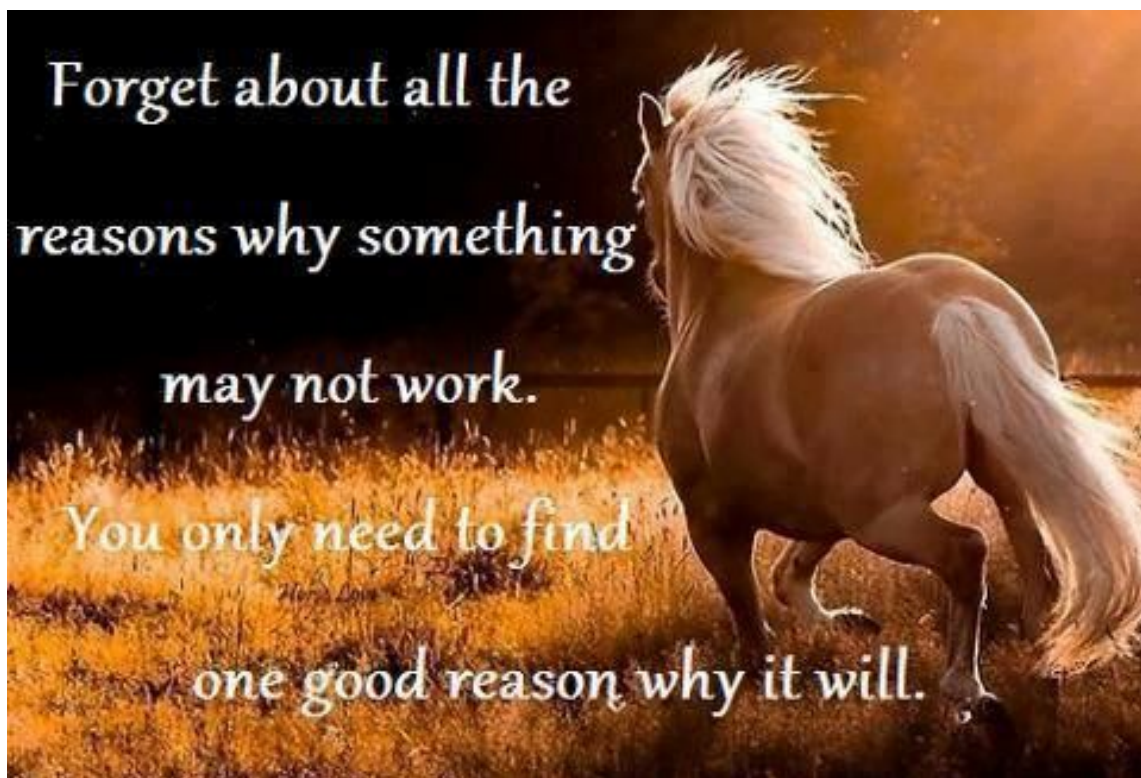


21

No more “Can’t”...



Danger of Low Expectations



23

"She Won't Get Anything Out of It"

Emma's Hope Book

On the topic of Shakespeare's Romeo and Juliet

Teacher: "Give me two or three words to describe 16 year-old boys."

Emma: "Acne-faced with bravado to mask their insecurity."

Teacher: "What's Benvolio's plan to cheer Romeo up in Act 1, Scene 1?"

Emma: "He doesn't consider women to be without replacement and suggests Romeo easily find another."

24

Wanted: Encouragers

[We have a surplus of critics already, thanks.]

25

Dignity of Risk




SpiritualityandPractice.com

Hope and Fear
cannot occupy
the same space
at the same time.
Invite one to stay.

— Maya Angelou

27

Far too many
people
have no idea
what they
can do

A person in a blue suit is walking on a thin tightrope. The tightrope is stretched across the top ring of a large, metallic blue anchor. The anchor is positioned in the center of the image, with its flukes pointing downwards. The background is a light gray gradient.

because all
they've
been told
is what they
can't do.

-Zig Ziglar

Ziglar.com

28

**“What will it
take to make
it possible?”**

29

**“How can this
person participate
in a
meaningful way?”**

30

**“How can this
person make a
meaningful
contribution?”**

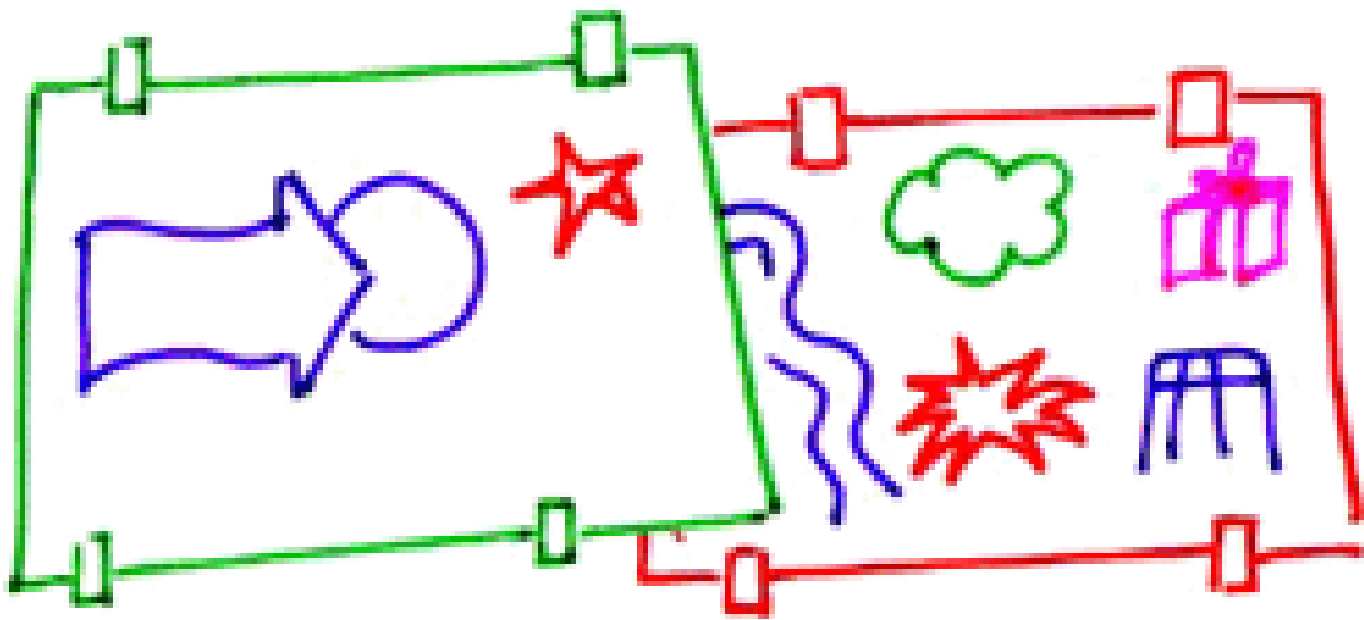
31

Planning for the Future



PATH: Planning Alternative Tomorrows with Hope
A person centered planning tool used by
Illinois Association of Microboards and Cooperatives.

Person Centered Planning



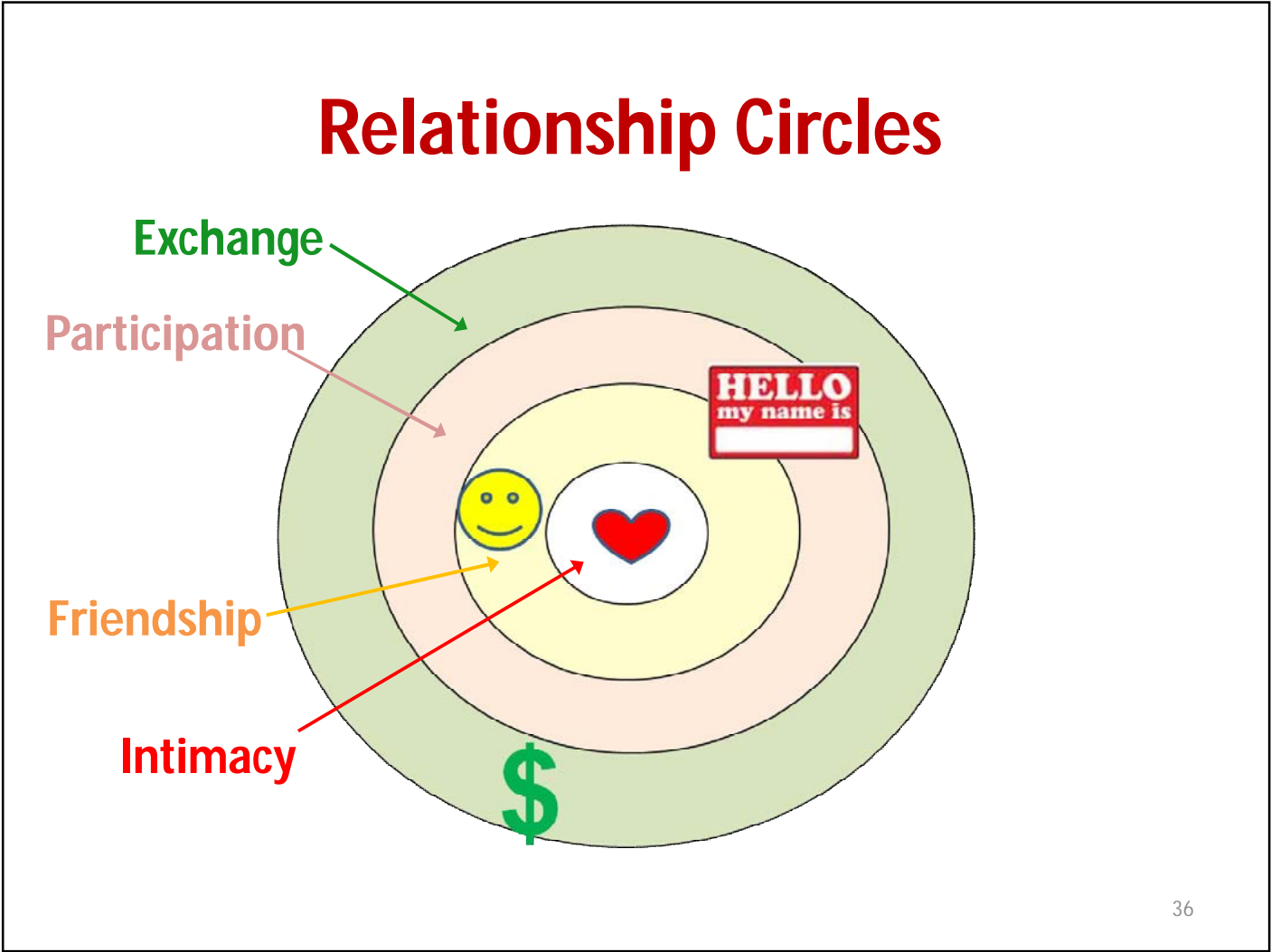
CALL IT A CLAN, CALL
IT A NETWORK, CALL
IT A TRIBE, CALL IT
A FAMILY.
WHATEVER YOU CALL
IT, WHOEVER YOU ARE,
YOU NEED ONE.

JANE HOWARD WWW.VERYBESTQUOTES.COM

34

Build Connections





Some Questions to Get Started

- What does your best life look like?
- Listen for the values and themes underneath the dream.
- What are your passions and gifts?
- Who are others who share your passions?
- Where are the 'sweet spots' where your gifts are appreciated?
- How can you build relationships with people who share your passions in places where your gifts are appreciated?

37

If you are a Person with CHARGE...

- Identify and express your vision.
- Learn and practice self-advocacy.
- Take the lead in planning meetings
- Take responsibility.
- Get to know new people and go new places.
- Find the 'sweet spots' in your community.
- Share your gifts and talents.
- *Expect ExtraOrdinary!*

38

If you are a Parent...

- Support self-advocacy and independence.
- Carry the dream.
- Be a cheerleader, encourager, coach.
- Let the protector role slowly fade...
- Facilitate connections and community inclusion.
- *Expect ExtraOrdinary!*

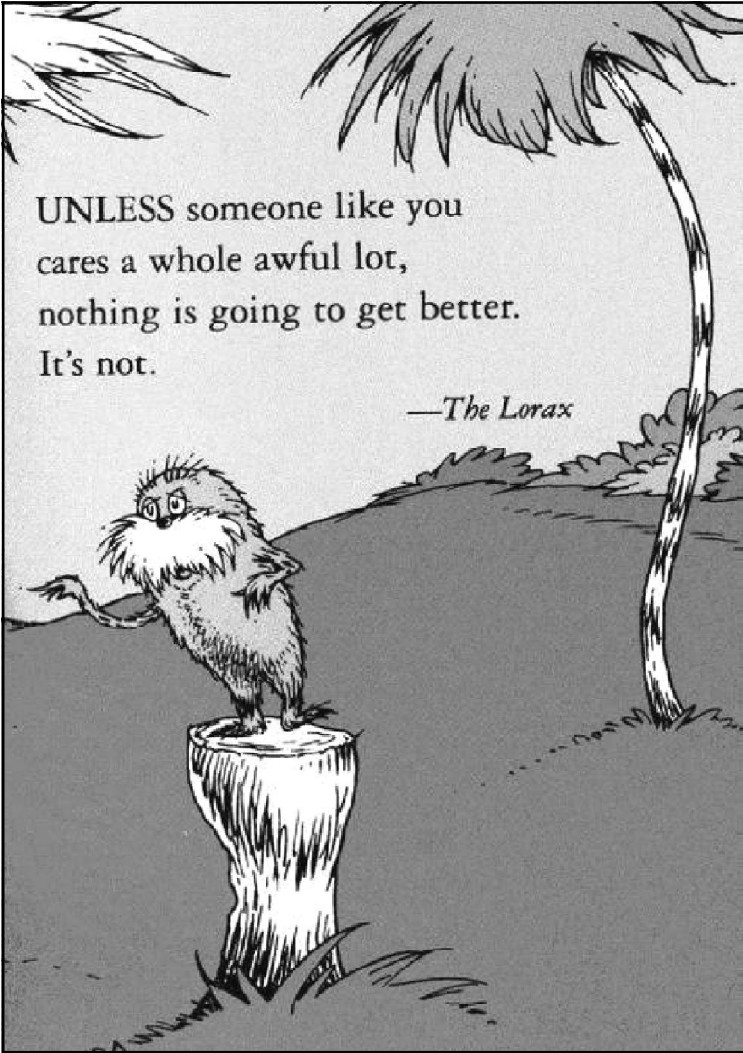
If you are a Professional...

- Respect the dream.
- Provide guidance and support.
- Model respect and equality.
- Add self-determination and self-advocacy skills to goals.
- Facilitate connections & community inclusion.
- *Expect ExtraOrdinary!*

40

Make Room for Magic





**Be an
ExtraOrdinarian!**

The Time to Start is Now!

“Someday is not
a day of the week.
You’ve got
to make a plan.”

~ DR. PHIL

www.parentingspecialneeds.org

Art Courtesy © Sergey/Pesterv/photoxpress.com

43

“IT ALWAYS SEEMS
IMPOSSIBLE
UNTIL IT’S DONE.”
- NELSON MANDELA



**Expect
ExtraOrdinary!!!**

**Resources & links related to topics
discussed can be found at
www.ExpectExtraOrdinary.org**

44

Expect ExtraOrdinary: Using Vision, Planning, and Community to Expand Possibilities

CHARGE Syndrome Conference
July 2015
Aubrie and Michele Westmaas
www.ExpectExtraOrdinary.org

Everyone CAN be ExtraOrdinary!

"People with disabilities
need to be treated ordinary
but they do have some
extras."

Aubrie Westmaas

2

Meet Aubrie...



Aubrie belongs –

- In class
- In band
- In drama
- In chorus
- In church
- In Girl Scouts

....Everywhere her peers
belong!

3

What if she'd been born generations ago?



This is
Margaret
as a baby.

4



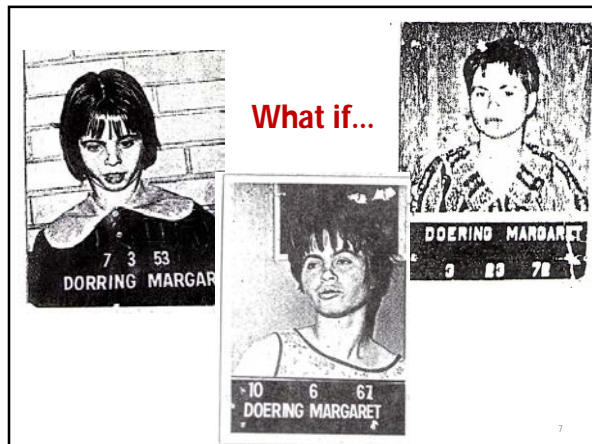
By the time
Margaret was 2
years old, she was
not progressing at
a typical rate.

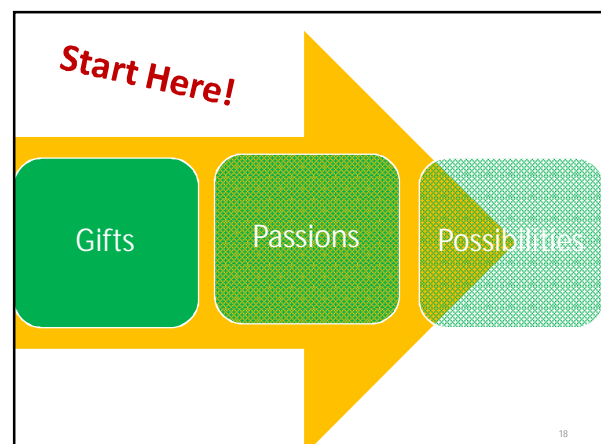
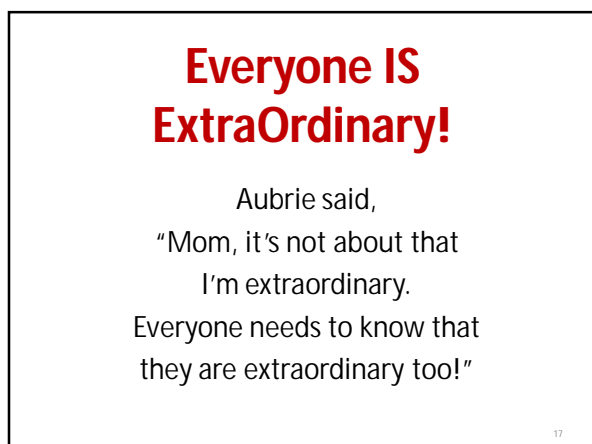
5



At age 2, Margaret was sent
to Dixon Developmental
Center. Her siblings never
knew she existed.

6





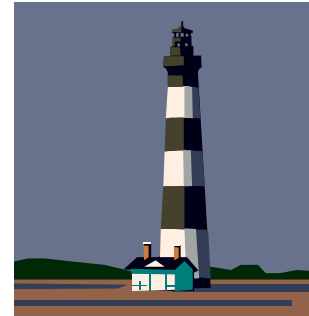
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**you have
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19

What's The Vision?



20

No Bubble-Bursting!



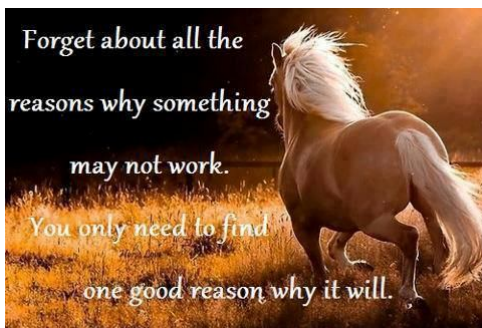
21

No more "Can't"...



22

Danger of Low Expectations



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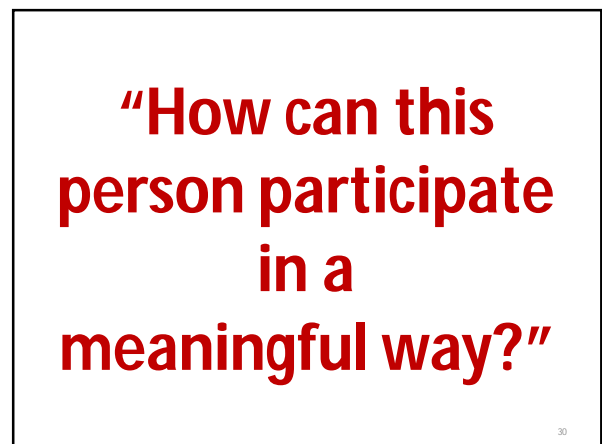
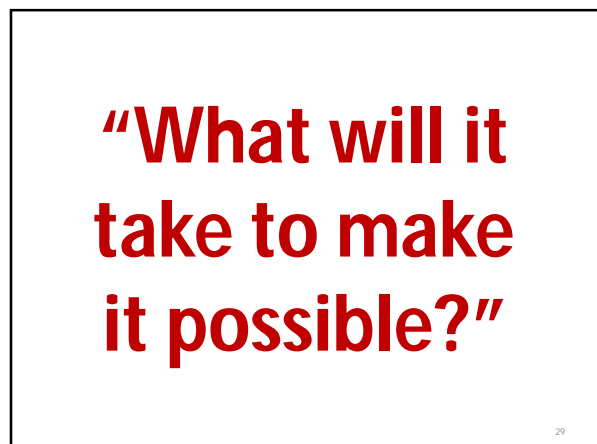
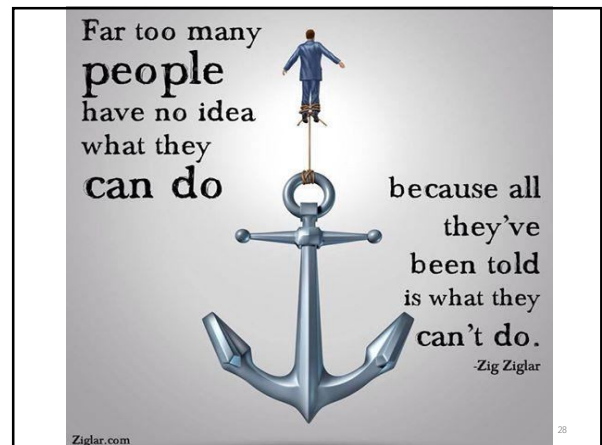
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Teacher: "What's Benvolio's plan to cheer Romeo up in Act 1, Scene 1?"

Emma: "He doesn't consider women to be without replacement and suggests Romeo easily find another."

24



“How can this person make a meaningful contribution?”

31

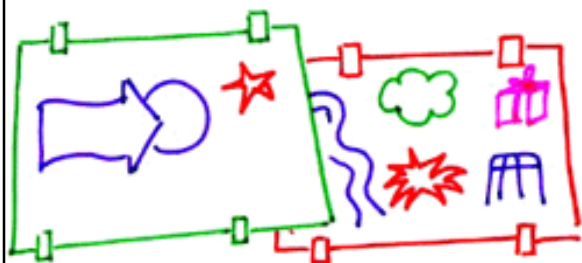
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32

Person Centered Planning



33

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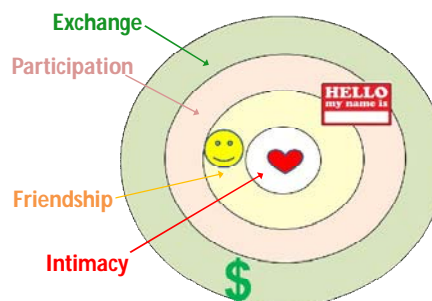
34

Build Connections



35

Relationship Circles



36

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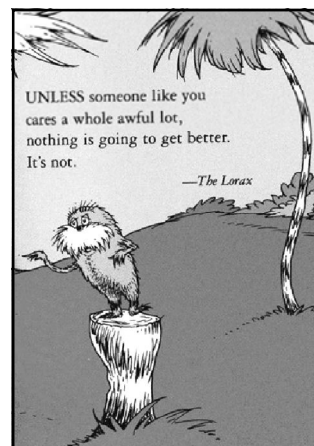
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- Model respect and equality.
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- Facilitate connections & community inclusion.
- *Expect ExtraOrdinary!*

40

Make Room for Magic



41



**Be an
ExtraOrdinarian!**

42



Additional resources related to topics discussed can be found at www.ExpectExtraOrdinary.org. Email us at ExpectExtraOrdinary@gmail.com. Find us on Facebook at www.facebook.com/ExpectExtraOrdinary.



**Friday, July 31, 2015
Breakout Session #20: 3:30 - 4:30pm
Schaumburg A-B**

**Psychiatric Issues in CHARGE
Syndrome**

**John Julian, M.D.
Consultant, Perkins School for the Blind**

Presenter Information:

John Julian, M.D., specializes in child and adolescent psychiatry and also specializes in developmental disabilities. He has consulted to the Perkins School for the Blind for 12 years and worked with many children who have CHARGE syndrome. He is affiliated with Massachusetts General Hospital.

Presentation Abstract:

Parents and professionals often have concerns about OCD, anxiety, aggression, self-injurious behavior, medications and more. During this one-hour question and answer session, attendees will have the opportunity to discuss these issues or other psychiatric issues with Dr. Julian, who has consulted to the Perkins School for the Blind for 12 years and worked with many children who have CHARGE.



**Friday, July 31, 2015
Breakout Session #21: 3:30 - 4:30pm
Schaumburg C-D**

Homemade Tube Feedings

**Carrie Ek, RD, LDN, MBA
Advocate Children's Hospital-Park Ridge, IL**

Presenter Information:

Carrie has worked at Advocate Lutheran General/Advocate Children's Hospital for over 20 years and has been involved with: eating disorders/addictions, adult cardiac, diabetes and general medical. She has worked with pediatric patients some inpatient, but primarily outpatient nutrition counseling for the last 15 years. Carrie works with children who have a wide variety of medical needs including: underweight/overweight, tube feedings, food allergies and those who are medically complex. She also works in cystic fibrosis, eosinophilic esophagitis (EOE), and celiac clinics and is the coordinator of the pediatric celiac center.

Presentation Abstract:

Homemade or blenderized tube feedings are an alternative to prepared formulas. This will be a discussion of benefits of using "whole foods" and the potential problem areas. Nutrition needs for specific ages and for kids with Charge syndrome will be discussed in detail. I will also touch on mixed (not put in a blender) options.

Home made Tube Feedings Charge Syndrome

Carrie Ek, RD, LDN, MBA
Pediatric Nutritionist



Nutrition Issues for Charge Syndrome

GERD

Growth hormone
deficiency: 15%

Short stature: 70%

Low muscle tone: 90%

Sensory/feeding
difficulties

Swallowing difficulties

Osteoporosis

Heart disease

Failure to thrive due to
chronic illness

Chronic constipation

Congenital heart disease
and need for surgery

Respiratory problems

Nutrition for Children

Goal is to Support Growth!



Protein 12-20%

Carbohydrate 50%

Fat 30%

Fluid/water

Multivitamin with iron for age

Calcium & Iron

Tolerance??

= **Calories**

How much does the child need?

AGE	CALORIES/kg	PROTEIN/kg	CALCIUM	IRON mg/day	VITAMIN D IU/day
0-6mos	108	2.2	800	6-10	400
6mos- 12mos	98	1.6	800	10	600
1-3 yrs	102	1.2	800	10	600
4-6yrs	90	1.2	1000	10	600
7-10yrs	70	1.0	1000	10	600
11-14yrs	50-55	1.0	1300-1500	15	600
15-18yrs	40-45	.8-1.0	1300-1500	15	600

Fluid needs are usually equal to calorie needs
“catch-up weight gain” add 10-50%, sometimes more
Pounds /2.2=kg

Food Sources

Protein: meats, eggs, dairy

Carbohydrate: fruit, rice, pasta, juice, milk

Fat: oils, butter

Iron: meats are best source

Calcium: 1 cup milk or yogurt=300mg

Vitamin D: Not many good food sources: Sunsh

Fortified: Milk 100 I U per 8 oz

Soy Milk 120 I U per 8 c

Yogurt 40 I U per ½ cup

Natural: Fatty fish (salmon) 300-1,300 I U/4 oz,

Eggs 25 I U/yolk

Cod liver oil 450 I U/tsp



Underweight Children

Calories are always the priority

Total calories=carbohydrate + protein + fat

Add 10-20% additional calories

Infants: concentrate formula

Toddlers usually 100-300 extra calories/day

Older kids 300-500 extra calories/day

Lower calorie foods: vegetables/fruits are low priority

Detailed food history!

How to add calories for weight gain?

Eat meals and snacks on a regular schedule.

Do NOT wait until hungry to eat.

Eat every 2-3 hours. Eat three meals and two-three snacks every day.

Make every bite count. Pack as many calories as possible into each bite!

Add **butter**, **margarine** or **oil** to foods wherever possible in cooking or at the table.

Sour cream

Grated cheese

Half and half

Cream

Mayonnaise

Salad dressing

Whipping cream

Avocado

Eggs

Gravies

More calories=weight gain. Read labels for calorie content and know your calorie goals.



Tube Feedings



Bolus feedings: usually over 30 minutes to one hour given via syringe

Continuous Feedings: small amounts given over most of the day through a pump



Combination? Bolus during the day and continuous at night

Oral feeding? Some bolus feedings and some meals by mouth

Pediatric formulas

General formulas:

30 kcal/oz standard

- Pediasure over 1 year (Nutren Junior)
 - Need 1 liter to meet RDA needs for 1-8 years old
 - Need 1.5 liters to meet RDA needs for >9
 - With and without fiber, 1.0 and 1.5

Specialty formulas:

- Peptamen Junior (1.0 and 1.5): Semi-elemental
- Pediasure Peptide (1.0 and 1.5): Semi-elemental
- Elecare Junior elemental
- Neocate Junior: elemental
- EO28 Splash: elemental juice

Tube Feeding Formula with Real Food Ingredients

The Only Tube Feeding Formula Containing Real Food Ingredients for Children Ages 1-13

1.0 cal/ml

Water, Corn Syrup, Green Pea and Green Bean Puree (Water Dehydrated Peas and Green Beans), Chicken Puree (Water Dried Chicken), Peach Puree (Water Peach Puree Concentrate), Sodium Caseinate (from Milk), Cranberry Juice (Water Cranberry Juice Concentrate) and less than 2% of Canola Oil, Medium Chain Triglycerides (from Coconut and/or Palm Kernel Oil), Partially Hydrolyzed Guar Gum*, Calcium Phosphate, Potassium Citrate, Hydroxylated Soy Lecithin, Choline Chloride, Maltodextrin, Salt, Sodium Citrate, Sodium Ascorbate (Vitamin C), Magnesium Oxide, Carrageenan, Potassium Hydroxide, Taurine, Alpha Tocopheryl Acetate (Vitamin E), M-Inositol, Ferrous Sulfate (Iron), Zinc Sulfate, L-Carnitine, Natural Flavor, Calcium Pantothenate, Niacinamide, Vitamin A Palmitate, Vitamin K₁ (Phytonadione), Vitamin D₃ (Cholecalciferol), Manganese Sulfate, Thiamine Mononitrate (Vitamin B₁), Pyridoxine Hydrochloride (Vitamin B₆), Riboflavin, Citric Acid, Copper Sulfate, Beta Carotene, Folic Acid, Biotin, Potassium Iodide, Chromium Chloride, Sodium Molybdate, Sodium Selenate, Vitamin B₁₂ (Cyanocobalamin).



Can be adapted for children.



Homemade Tube Feeding??



Can child tolerate bolus feedings?

G-tube?

Does the family have time, energy and motivation?

Medically stable?

Discuss with medical team?

Tube size? Should be 14 French or greater

Why choose to make your Tube Feeding?

- Variety
- Less additives/preservatives
- Feed your own child
- Real food: whole foods
- Possible better digestion
- Better management of food allergies
- It does not have to be all or nothing: it is ok to still use some formula.
- Less expensive
- Food allergies

Risks of Homemade Tube Feedings

Inadequate nutrition

Contamination: uncooked foods or unclean equipment

Tube clogging: blend until smooth: LIQUIFEY !

Monitor weight and growth!



Safety Tips for Homemade formulas

- Keep formula in refrigerator until right before feeding
- Never leave formula at room temperature for more than two hours.
- Thaw all foods in refrigerator, not room temperature.
- Store cooked fruit/veg/grains in fridge for 2-3 days, 6-8 months in freezer
- Store meats 1-2 days in fridge, 1-2 months in freezer
- Avoid "danger zone" 40-140 degrees: bacteria growth
- Keep hands and all equipment(utensils, cutting boards) VERY CLEAN



Blended Foods

- Good blender
- Proper Food Sanitation
- Better to prepare individual types of foods separately and then blend right before giving
- Store each food separately and blend together before giving
- Give at room temperature (68-72°)
- Always flush with water before and after feeding
- Lots of liquid: juice, broth, water, milk



What to blend?

Think in terms of basic food groups:

Fruit/vegetables: 4-5 servings/day: ½ cup= 1 serving

Grains : rice, oats, couscous, potatoes

Protein: legumes, meats of all kinds, tofu, cooked eggs

Dairy: milk, yogurt

“plant based milks”: rice, coconut, almond are VERY LOW
IN PROTEIN

Fats: oils of any kind

Water

Vitamins/minerals: liquid or crushed chewable

Don't put in a tube amounts a child would
not normally eat

Should be similar to typical foods for age

Back up Food

- Baby foods
- Blended or pureed soups
- Compleat Pediatric Formula
- Real food blends
- Liquid Hope formula
- It is ok to do some formula:
vacations/busy times



© POLYPACK containers | www.fourpack.it

Age	Fruits(Cups)	Vegetables (Cups)	Grains(ounces)	Meats & Beans(ounces)	Milk(Cups)	Oils(tsp)
2-3	1	2	3	2	2	3
4-8	1-1½	1½	4-5	3-4	2	4
9-13(females)	1½	2	5	5	3	5
9-13 (males)	1½	2½	6	5	3	5
14-18 (females)	1½	2½	6	5	3	5
14-18 (males)	2	3	7	6	3	6

Recipes: 1 cal/ml or 30 cal/oz
1014 calories, 45 gms protein

12 oz 2% milk

½ banana

2oz meat

4oz prune juice

1 egg, scrambled

1/8 avocado

½ cup brown rice

1 tablespoon olive oil

½ cup green peas

1 tbsps Honey

¼ cup spinach

1/8 teaspoon table

¼ blueberries

salt

Recipe: 1.5 cal/ml or 45 cal/oz
1508 calories, 53 gms protein

8oz apple juice

6 oz prune juice

5oz meat

1 egg, scrambled

½ cup rice

¼ cup green peas

½ cup carrots

1/8 cup spinach

½ banana

¼ avocado

2 tablespoons
vegetable oil

1/8 cup creamy peanut
butter

3 tablespoons honey

1/8 teaspoon table
salt

Oley foundation: www.oley.org

Mealtime Notions: www.mealtimenotions.com

Food for Tubies: www.foodfortubies.org

Liquid Hope: www.functionalformularies.com

Compleat: www.nestlenutrition.com

www.feedingtubeawareness.com

www.realfoodblends.com NOT a complete formula

Carrie Ek, RD, LDN
Pediatric Nutritionist

(847) 723-7181
carrie.ek@advocatehealth.com





**Friday, July 31, 2015
Breakout Session #22: 3:30 - 4:30pm
Schaumburg West**

**Cochlear Implants in children with
CHARGE Syndrome**

**Elizabeth Tournis, Au.D. & Nancy Young, M.D.
Ann & Robert H. Lurie Children's Hospital of
Chicago**

Presenter Information:

Beth Tournis is the Coordinator of the Cochlear Implant team at the Ann & Robert H. Lurie Children's Hospital of Chicago (formerly Children's Memorial Hospital). Beth joined the cochlear implant team at Lurie Children's in 2003. Prior to her involvement with cochlear implants she specialized in auditory evoked potentials and working with multiply involved hearing impaired children. She has worked with children with CHARGE syndrome since 2003. In addition to presenting at the 9th International CHARGE Syndrome Conference, she presented a talk regarding cochlear implants and this population at the 12th International Conference on Cochlear Implants and Other Implantable Auditory Technologies. Beth holds an Au.D. from Pennsylvania College of Optometry and an M.A. from Northwestern University.

Presentation Abstract:

Cochlear Implantation provides deaf children with the opportunity to have useful hearing that will assist them in achieving their communication and educational goals. The Lurie Children's Cochlear Implant Program, has been in the forefront of providing implants to diverse candidates including those with CHARGE syndrome. In our experience many of these children derive tremendous benefit from an implant. Our presentation will include case presentations with outcomes data demonstrating the impact of cochlear implants on this population.



**Friday, July 31, 2015
Breakout Session #23: 3:30 - 4:30pm
Schaumburg E-F**

**Quality Not Quantity – Thoughts on
Communication, Behaviour, Play and
Passions**

**Rob Last
CHARGE Syndrome Association of Australia
and New Zealand**

Presenter Information:

Rob Last is a special education teacher who has a long history working with children who have sensory disabilities, particularly in hearing and vision. Since the mid 1980's he has been involved with children who have CHARGE syndrome and their families. He and a group of parents established the CHARGE Syndrome Association for Australia and New Zealand in the late 1980's. Rob has been a presenter at many of the CHARGE Syndrome International Conferences since the first in St Louis in 1993 and the first CHARGE Syndrome Conference in Australia and New Zealand held in Sydney 1994. He presented at the CHARGE Syndrome Conference in Germany in 2012. Rob's career has been as an Early Childhood Educator with the Royal Institute for Deaf and Blind Children in Sydney and the Royal Victorian Institute for the Blind and Vision Australia in Melbourne, Australia.

He continues his work with the CHARGE Syndrome Association of Australia and New Zealand as a Director for the state of Victoria and as Director of Outreach for Australia and New Zealand. Rob has the pleasure of meeting and being in contact with families from all over Australia and New Zealand as well as the CHARGE international community.

Presentation Abstract:

The presentation will focus on early communication and behaviour, address the importance of establishing a meaningful communication system for children with CHARGE and the strategies that may be available for this. Play strategies will be demonstrated through video, photos and discussion. Passions/obsessions as educational opportunities will be discussed and illustrated through video and photos. We will discuss the importance of appropriate early intervention strategies to ensure positive outcomes. Case studies and photos will support this discussion. 'Meltdowns' will be discussed as a form of communication, why these may occur and strategies for managing these. Communication systems will be highlighted as well as the supports available in the acquisition these.

HANDOUT FOR ATTENDEES

12th International CHARGE Syndrome Conference
Chicago, USA, 30 July – August 2, 2015

Quality not Quantity
Thoughts on:
Communication, Behaviour, Play and Passions

Rob Last

Director of Outreach, Director for Victoria
CHARGE Syndrome Association of Australasia
Teacher of the Deaf
Early Childhood Educator
Email: roblast@inet.net.au

Introduction

Conferences are filled with highly experienced experts, these being parents and professionals and those who have CHARGE.

So I hope to offer some new and old thoughts on communication, behaviour, play and passions.

My primary focus throughout my career has been communication; communication with babies, children, teenagers and adults.

I believe it is one of the keys to breaking through, to find the person.

I also mean all kinds of communication, simple to complex and in it's many forms.

Nikki

Not long ago I participated in a Skype meeting with a family I hadn't met. It was a really useful meeting as I realised many of the things I've been saying for years are still relevant.

I met Megan; mother of Nikki, who at this time was 3 and 1/2 years old and some members of her cochlear implant team, Nikki had received a cochlear implant about 18 months previously.

The meeting was to discuss the impact of CHARGE syndrome on Nikki's development.

The all too familiar CHARGE scenario was described, multiple hospitalizations, surgeries, a range of interventions, all completely overwhelming for Nikki and her family.

More recently things were just starting to settle down with less hospitalisations and illnesses.

Nikki was described as having been withdrawn and anxious and was now 'emerging from her withdrawn behaviour' with everyone around her being very excited by the changes.

As we know many things may happen to children with CHARGE without warning or preparation.

Educators familiar with children with dual sensory impairment strive to educate the medical and therapy teams in strategies to alert the child as to what is about to happen. They often do this verbally ('I'm going to give you an injection now') but don't add in touch cues, gestures, pictures, drawings, anything that will help prepare the child for what is about to happen.

Nikki needed to understand what was going on around her, what was happening to her, what was going to happen, would it be painful, Nikki needed information meaningful to her to ease the stress of her already chaotic and confusing life.

Nikki was described as being 'anxious' and I was asked is anxiety typical in children with CHARGE.

'Anxious' is a word I hadn't often used before to describe behaviour in children with CHARGE.

I was surprised I hadn't used it more in this context.

Yes of course children, teenagers, adults with CHARGE are anxious.

Why would they be anxious?

They need to know:

- what is about to happen
- when will it happen
- that it needs to happen now
- then what is going to happen after that
- and after that, after that ...

Other terms are also used such as:

- obsessive compulsive disorder (OCD)
- autism spectrum disorder (ASD)
- repetitive behaviours
- attention deficit disorder

- tics
- self-harm
- non-compliance
- tantrums
- meltdowns.

We often talk about how all the behaviours we see in children with CHARGE are perhaps exhibited by everyone; it's just that people with CHARGE really amplify these behaviours.

I remember in one of Tim's presentations a few years ago he had a slide of the things that define Obsessive Compulsive Disorder.

I must tell you I scored really well on that OCD screening test.

Order, organisation and tidiness have a really important purpose for me. It keeps my world orderly, predictable and easy to access.

I think those words predictable, orderly and easy to access apply to many of those with CHARGE.

Meltdowns

We talk about Meltdowns a lot.

It seems to me that one of the contributing elements would be anxiety.

In the early years

- What are you doing to me?
- What are you going to do to me?
- I don't understand what's happening
- That hurt
- I feel sick

In the later years

- I want it now
- I want it to happen now
- You are not listening to me
- You do not understand what it is I want

- You didn't understand what I meant
- You just didn't understand
- You haven't explained it to me and I'm unsure as to what is happening or what is expected of me
- Will it happen now?
- What will happen next?
- What will happen after that ... then after that?

These anxieties may lead to

- Distrust
- Fear
- Anxiety
- Outbursts
- Refusal
- Withdrawal

At that Skype meeting I explained how 'Meltdowns' are not restricted to age or development. I've seen 'meltdowns' in babies, children, teenagers and adults. From those with high support needs to those who are independent, mainstream educated and employed ... the whole spectrum.

What I've observed over the years is that nearly all of those who have CHARGE have functional vision. They may be legally blind but function extremely well visually.

I've always felt we must use all senses as a means of providing meaningful information, even though we know all senses are affected.

Therefore using visual language and touch cues to support spoken language is of great importance to me.

So for parents, educators, therapists and doctors it's our challenge to make every effort to make their world more predictable, more ordered, more organised and tidier, using all possible means to achieve this.

- what will happen now
- what will happen next
- what will happen after that
- then after that and so on

to reduce distrust, fear and anxiety.

I digress, back to Communication

At that Skype meeting I reiterated my belief that it's our job is to help make sense of this chaotic, unpredictable world for the baby, child, teenager and adult.

In talking to Megan (Nikki's Mum) about communication we discussed communication as a complete entity.

I often hear the focus being speech as communication and secondly sign language. Speech is of course the most desirable as it's the mother tongue of the home and the world. The aim of speech being the most desirable outcome is an appropriate aim.

Parents and professionals always strive to provide the best possible audition through: amplification, hearing aids, cochlear implants, bone aids, bone anchored hearing aids, FM systems.

I also maintain we need to add all types of 'visual language': sign language, key word signing, gestures, body language, facial expression, pointing, mime, real objects, symbolic objects, photos, drawings, symbolic drawings (boardmaker, proloquo2go) and reading and writing.

Multimodal communication.

About Learning Sign Language

I acknowledge that acquiring a new language like American Sign Language (ASL) is a big challenge.

Here are a few suggestions to assist in this process.

- use teachers of the deaf fluent in sign language
- use signing dictionaries
- use DVD's teaching sign language
- seek out members of the deaf community
- seek out CODA's (Children Of Deaf Adults)
- attend classes/courses
- attend events with the deaf community
- find a neighbour, friend of a friend, someone who is fluent in sign language
- choose voice off times to rely on signs language only

Play with Younger Kids

Of course play is also communication.

Parents report that bonding and attachment is severely interrupted when they have a child with CHARGE. One of the challenges is building a trusting relationship through offering interactions and communication that are meaningful, that will make sense, that will be understood, that will be fun, that will be interesting and will be rewarding and worthwhile.

Nikki is only just embarking on this journey.

Her parents and therapy team have progressed gently and cautiously. This has come from an understanding as to why her early experiences have caused her to withdraw into that inner place, that place where she feels safe.

The adults have engaged in careful and gentle encouragement to enable Nikki to trust and engage in the world outside her body.

They began by joining Nikki in her world.

Heiko and Jonas

In this presentation I show three movie clips of a Dad, Heiko, and his son, Jonas. The movies shows them engaged in relaxed and pleasurable play.

Heiko uses simple strategies to engage Jonas in play.

The movie is in German so it is interesting to notice how much is understood without understanding spoken German, how much the visual communication explains what is going on.

(Movies – Heiko and Jonas)

So what was noticed about play in the movies.

Movie 1

- Heiko gains Jonas's attention
- He prompts turn taking
- He rewards imitation
- He pauses while Jonas looks at the camera.
- Rewards again with 'Bravo' and 'clap, clap'
- He allows looking away
- He uses speech, gesture, signs and more
- It was also interesting and fun

Movie 2

- Jonas is well supported on his dad's body
- Dad is also comfortable
- Dad follows Jonas's interests
- Allows time for Jonas to process information
- Responds to Jonas's request for a repeat
- Dad uses animated, interesting and rewarding facial expressions

Movie 3

- This one depicts more adult directed play
- Heiko takes his time to ensure Jonas understands the game
- He revises the steps to ensure Jonas has every chance to succeed
- He allows Jonas to succeed without adult intervention
- Success is Jonas's reward

Heiko used

- Speech
- Sign language
- Facial expression
- Singing
- Body language
- Gestures
- Mime
- Touch cues
- Symbolic objects
- Real objects
- Photos
- Drawings

To engage in successful play the adult must

- Follow the child's lead
- Capitalize on interests
- Provide interesting choices
- Break tasks into achievable steps
- Provide developmentally appropriate tasks
- Encourage and support self-discovery
- Provide meaningful communication

- Plan for success
- Experiential learning is often the best learning

And it's a good idea for the adult to rediscover their inner child

PLAY - Play is Children's Work

A parent reminded me to mention that often professionals engage in play without explaining what it is they are trying to achieve and what they are looking for.

Teachers and therapists understand that play is children's work and know that through play an assessment of abilities can be achieved. They note gaps in abilities and use play to teach to those gaps.

In the early years it is following the interests of the child in play which progresses into adult/teacher directed play in the later years.

It should always be:

- Stimulating
- Rewarding
- Challenging
- Meaningful
- Enriching
- Experiential
- Fun

Play for Older Kids

Passions/Obsessions

We all have them. We are much more content when we are able to engage in our passions/obsessions.

Using passions/obsessions to support learning is good teaching.

These may be through

- Sean and Bailey – Lifts
- Trent - Technology
- David – Horse riding
- Sarah – Horse riding
- Phillip - Tapestries
- Belinda - Travel
- Ellen – Facebook
- Sean – Caravans

- Julia – Sorting and Organising
- Mitchell – Football and Spinning
- Sophie – Basketball

Use these behaviours to:

- Educate – for example; language building, reading, writing, maths, science, geography, social skills, self-help skills, organisational skills, planning skills and more
- To establish and develop friendships
- As a hobby
- For pleasure
- To provide relaxation
- To enhance self esteem
- As exercise
- To further encourage communication
- As a possible career plan

AND

- Focus on the passion
- Plan ways of engaging with the passion
- Involve friends in the passion
- Find others with the same passion
- Record through photos, filming and writing
- Have fun
- Remember experiential learning is the most powerful.

Take Home Messages

- Communication means auditory and visual expressive and receptive language
- Be thoughtful and thorough in preparing for routines and events
- Use play for learning and social outcomes
- Use passions/obsessions for learning and social connections ... and maybe a career opportunity

Reading tips

Four Great Books

- 'An Exceptional Fellow' A Father's Story - Svein Olav Kolset
To purchase: <http://anexceptionalfellow.com/>
- 'Far From The Tree' - Andrew Solomon
Available from Amazon and The Book Depository
- 'CHARGE Syndrome' - Hartshorne, Hefner, Davenport, Thelin
Available from Amazon
- 'Why I Am Me' - Ward, Patterson and Levett
Purchase at this conference or from Australian/NZ CHARGE website:
<http://www.chargesyndrome.org.au>

Families as Resources

- Parents, siblings and grandparents
- Each and everyone here is a resource
- Find out who has similar issues
- Access each other via email, phone, social media, face-to-face
- Be a pro-active resource to others
- Notice with whom you feel a connection
- Swap contact details
- Use each other as needed

Other Human Resources

- All the speakers
- All the parents and grandparents
- All those who have CHARGE syndrome

Thank you

Rob Last



**Friday, July 31, 2015
Breakout Session #24: 3:30 - 4:30pm
Schaumburg G**

**Defense mechanisms: Immunological
and adrenal function in CHARGE
syndrome**

**Monica T.Y. Wong, MD, PhD student
University of Groningen, University Medical
Center Groningen**

Presenter Information:

Monica is graduated from the medical school of the University of Groningen. She has worked a couple of years as a resident in the department of Obstetrics and Gynecology and in the Intensive Care Unit. Currently, Monica is working as a PhD student at the department of Genetics in the group of prof. Conny van Ravenswaaij. Her main research project is on immunology and adrenal function in CHARGE syndrome. In addition, she is involved in projects on novel gene identification by next generation sequencing and developing growth charts for CHARGE syndrome.

Presentation Abstract:

Children with CHARGE syndrome have problems in many organ systems. This means that they are more vulnerable to illness. The immunological and adrenal system can prevent children from becoming ill. We show that otitis and airway infections, including pneumonia, often occur in children with CHARGE syndrome. Their immune system shows decreased numbers of certain immune cells and reduced responses to vaccination. However, adrenal function is almost always normal in children with CHARGE syndrome.

“Defense mechanisms: Immunological and Adrenal function in CHARGE syndrome”

About the presenter



My name is Monica Wong and I am a PhD student in the group of Prof. Conny van Ravenswaaij-Arts at the department of Genetics of the University Medical Center Groningen, the Netherlands. My main research project is on immunology and adrenal function in CHARGE syndrome.

Introduction to the study

What is the immune system and why is it important to explore this in CHARGE syndrome?

The immune system is an important defense mechanism of the body against infections by bacteria or viruses. The first barrier is formed by the skin and the mucosal membrane coating the inside of the nose, mouth and intestines. Most bacteria and viruses are blocked by this first barrier. When a bacteria or virus is able to cross this barrier and enter the tissues or bloodstream, the immune cells become active (see figure 1).

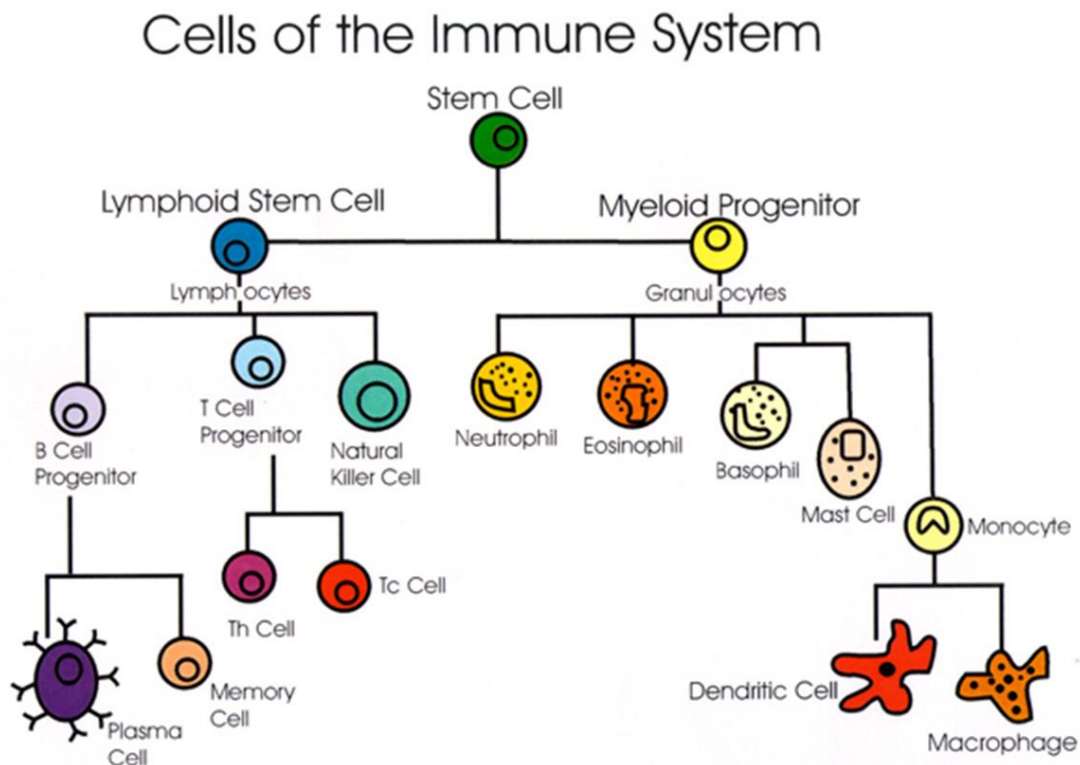


Figure 1. Cells of the immune system. All immune cells were explored in this study.

There are many types of immune cells and each type is needed for the immune system to function normally. The immune cells work together to detect and kill the bacteria or viruses for which a very delicate communication between the different cells is needed. Abnormalities in the immune system, such as decreased cell numbers or function, will lead to increased susceptibility to infections.

It is known that children with CHARGE syndrome have frequent infections, particularly in the upper airway (sinusitis and otitis media) and lungs (pneumonia). These infections are partially due to the anatomical problems, such as cleft palate and ear deformities, and swallowing difficulties due to cranial nerve abnormalities. However, abnormalities in the immune system might also contribute to these frequent infections. In literature, abnormalities in the immune system have been described in several CHARGE patients and they resemble the immunological abnormalities seen in patients with 22q11.2 deletion syndrome. This is not surprising since these two syndromes show overlap in other clinical features as well, such as heart defects, cleft palate, and ear deformities. But unlike 22q11.2 deletion syndrome, the immune system in CHARGE syndrome has never been studied in a systematic way. To better understand the frequency and type of abnormalities in the immune system, we have extensively explored the immune system of children with CHARGE syndrome. This knowledge will benefit the management of recurrent infections and therefore minimize the burden risk in these children.

What is the adrenal function and why is it important to explore this in CHARGE syndrome?

Another important defense mechanism of the body is the adrenal function. The adrenal glands are lying on top of the kidneys (see figure 2).

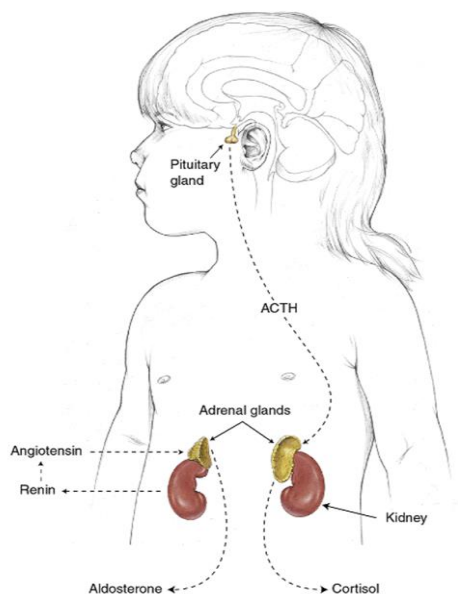


Figure 2. Anatomy of the adrenal glands

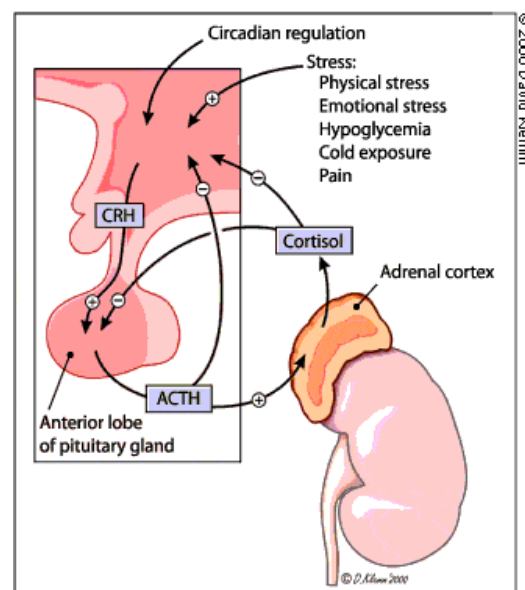


Figure 3. Regulation of the release of cortisol

The adrenal glands produce the hormone cortisol, important for your body to deal with stressful events. These stressful events can be emotional stress, such as anxiety, but also physical stress, such as a serious infection or an operative procedure (see figure 3). Subtle insufficiency in cortisol production by the adrenals will not present with overt symptoms. But if left unnoticed in an acute stressful event, it can lead to life-threatening situations.

Children with CHARGE syndrome deal with a lot of stressful events, such as frequent infections and several surgical procedures. It is therefore important to know whether adrenal insufficiency is part of the syndrome, especially since unexpected mortality has been described in several CHARGE patients.

What have we done in our study?

Participants

Children with CHARGE syndrome, between the age of 20 months and 17 years, were recruited from the Dutch CHARGE clinic. Only children with a genetically confirmed diagnosis, thus a mutation in the CHD7 gene, were eligible to participate. All children completed a questionnaire on infectious history.

Immunological laboratory tests

The immune cells and their function were assessed by laboratory tests in blood samples taken by all children. All immune cells were counted and the immune responses of the B-cells and T-cells (see figure 1) were tested.

Adrenal function test

The adrenal function was assessed by the low-dose ACTH test. During this test the adrenals were stimulated to produce cortisol. An inadequate level of cortisol indicated insufficient adrenal function. This had to be confirmed by a second test, the glucagon stimulation test.

What have we found in our study?

Frequent infections

24 children with CHARGE syndrome were included in our study and all children had a history of (recurrent) infections. Otitis media (16 children, 67%) and pneumonia (7 children, 29%) were most prevalent, for which 7 children (29%) received prophylactic antibiotics. 18 (75%) children needed hospital admissions for reasons related to infectious diseases, including insertions of ear tubes.

Immunological abnormalities

Three main abnormalities were observed:

1. Decreased numbers of T-cells.

12 children (50%) had decreased numbers of T-cells, probably due to decreased output from the thymus, an organ involved in forming functional T-cells. Thymus abnormalities have been described in CHARGE patients as well as in patients with 22q11.2 deletion syndrome.

2. Incomplete formation of memory B-cells.

8 children (33%) had B-cells which seemed impaired in their formation into normal memory B-cells. Insufficient T-cell help to B-cells might be an explanation for this abnormality.

3. Reduced immune responses to vaccines.

19 children (83%) had insufficient specific antibodies (reduced immune responses) to vaccines given in their childhood. This might be due to the impaired formation of functional memory B-cells that are involved in the production of specific antibodies.

Adrenal insufficiency is not common in CHARGE syndrome

We could test the adrenal function in 23 children. According to the low-dose ACTH test, 7 children were suspected to have adrenal insufficiency. However, only 1 child had a confirmed diagnosis of adrenal insufficiency after the second test. So, adrenal insufficiency seems not to be a common feature in CHARGE syndrome.

What are the implications of our results in the care for CHARGE patients?

Immunology

We found frequent infections in combination with a high percentage of immunological abnormalities. Further research in more children with CHARGE syndrome is needed to confirm our results and to develop evidence-based guidelines to improve the management of recurrent infections. Nonetheless, we would recommend to perform specialistic immunological laboratory tests in children with persistent infections needing prophylactic antibiotics. It may be worthwhile to give these children a booster vaccination.

Adrenal function

The adrenal function seems not to be affected in children with CHARGE syndrome. However, our results need to be confirmed in a larger group of children. For now, there are no indications for testing the adrenal function in children with CHARGE syndrome.

Acknowledgements

The following people were involved in conducting the study:

- Prof. Conny van Ravenswaaij-Arts, clinical geneticist, UMCG
- Dr. Elisabeth Schölvinck, pediatrician infectious diseases and immunology, UMCG
- Dr. Gianni Bocca, pediatrician endocrinology, UMCG
- Dr. Annechien Lambeck, medical immunologist, UMCG
- Dr. Mirjam van der Burg, medical immunologist, Erasmus MC
- Dr. Sacha la Bastide-van Gemert, statistician, UMCG
- Lianne Hogendorf, laboratory technician medical immunology, UMCG

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Contact information

For further information or questions, please do not hesitate to contact me.

You can send an e-mail to: charge@umcg.nl



**Friday, July 31, 2015
Breakout Session #25: 3:30 - 4:30pm
Schaumburg H**

**Conociendo los derechos básicos de
Educación Especial.
*Getting to know the basic rights about
Special Education***

**Clara Berg
New York Deaf-Blind Collaborative
Myrna Medina
California Deaf-Blind Services**

Presenter Information:

Myrna es Mexicana del estado de Durango, tiene dos hijos, Deloris de 20 y Norman de 18 años, sordo-ciego, quien fue el motivo de entrar en este medio. Myrna tiene 15 años de trabajar como Especialista Familiar para CDBS, entre otros compromisos, pertenece al grupo de Birth to Five Vision Network como Parent Liason desde el 2007; Centro de Niños y Padres, como coordinadora del grupo de padres.

Clara oriunda de Uruguay tiene 3 hijos. Su segundo hijo Kenny (34) es sordo-ciego debido a un nacimiento muy prematuro. Para aprender más acerca de sordo-ceguera comenzó trabajando como voluntaria en el Proyecto de Sordo-Ciegos de NY, en el que ahora ocupa su posición de Especialista de Familias por más de 25 años. Clara fue miembro fundador de dos organizaciones para padres, en 1988 el New York Parent Network con el que sigue muy involucrada, y en 1994 el National Family Association for Deaf-Blind, del cual es su actual Presidente.

Presentation Abstract:

Para familias de habla hispana y que tienen niños con el Síndrome de CHARGE, así como también para el equipo educacional y otros especialistas que son parte del equipo (OT, PT, TVI, enfermera, etc.) es de extrema importancia que estén bien informados de los derechos que tienen los niños para recibir una educación libre y apropiada. A veces por falta de comunicación o traducción apropiada, los padres no están al tanto de los derechos educacionales que están garantizados bajo las leyes estatales y federales. Esta va a ser una sesión interactiva para aclarar dudas o temores que tengan las familias acerca de derechos educacionales básicos para sus hijos.

For Spanish speaking families who have children with CHARGE Syndrome, as well as the educational teams and other specialists who typically serve them (OT, PT, TVI, nursing staff, etc.), it's extremely important to be informed about the rights the children have to receive a free and appropriate education. Sometimes because of lack of communication or appropriate translation, parents are not aware of the educational rights their children are guaranteed by state and federal laws. This is going to be an interactive session to clear the doubts or fears that families might have about basic educational rights for their children.



12th INTERNATIONAL CHARGE SYNDROME CONFERENCE

THE SKY'S THE LIMIT

CHICAGO 2015



The contents of this presentation were developed under a grant from the US Department of Education, # H326T130031. However, those contents do not necessarily represent the policy of the US Department of Education, and you should not assume endorsement by the Federal Government. Project Officer, Jo Ann McCann.



CONOCIENDO LOS DERECHOS BASICOS DE EDUCACION ESPECIAL

CLARA BERG, ESPECIALISTA DE FAMILIAS, NYDBC
MYRNA MEDINA, ESPECIALISTA DE FAMILIAS, CDBS

JULY 2015



The contents of this presentation were developed under a grant from the US Department of Education, # H326T130031. However, those contents do not necessarily represent the policy of the US Department of Education, and you should not assume endorsement by the Federal Government. Project Officer, Jo Ann McCann.

APRENDIENDO EL PROCESO DEL IEP, SUS LEYES Y TIEMPOS



Myrna Medina & Clara Berg

Agenda

Aprendiendo el proceso del IEP, sus leyes y tiempos

- ▶ Definición de Educación Especial
- ▶ Derechos y Responsabilidades según las Leyes
- ▶ Derechos de los padres de familia
- ▶ Servicios relacionados
- ▶ Proceso del IEP
- ▶ Participación de los padres de familia
- ▶ Un repaso y sugerencias para los padres

Cómo se define “Educación Especial”

5



- ▶ La educación especial es instrucción especialmente diseñada para responder a las necesidades únicas del niño con discapacidades.
- ▶ La instrucción puede ocurrir en el aula, en el hogar, en un hospital o una institución.
- ▶ La educación especial también puede incluir otros servicios relacionados (ej. terapia de lenguaje, visión, audición, movilidad, etc.)
- ▶ La educación especial puede empezar desde el nacimiento y hasta los 22 años de edad, sin costo para las familias.

Derechos y Responsabilidades de Acuerdo a las Leyes

▶ A nivel Federal:

- ▶ IDEA
- ▶ Procedural Safeguards
- ▶ No Child Left Behind
- ▶ Section 504

▶ A nivel Estatal:

- ▶ Reglamentaciones
- ▶ Leyes
- ▶ Pólizas y Estatutos

▶ A nivel Local:

- ▶ Reglamentaciones
- ▶ Pólizas (éstas pueden diferir de escuela a escuela).

IDEA

7

(Individuals with Disabilities Education Act)

Acta de Educación para Individuos con Discapacidades

- ▶ El Sistema de Educación Especial está basado en la ley Federal de Educación Especial llamada IDEA
- ▶ IDEA en combinación con leyes estatales de Educación Especial protegen a los estudiantes con discapacidades que son elegibles y garantizan un Plan de Educación Individualizado (IEP) diseñado para complementar sus necesidades únicas.



LOS *SEIS* PRINCIPIOS DE IDEA

8

1. Cero Rechazo
2. Identificación y Evaluación no Discriminatoria
3. Educación Pública Apropiaada Gratuita (FAPE)
4. Entorno Menos Restringido (LRE)
5. Derechos de Procedimiento Debido Legal (*Due Process*)
6. La Participación del Padre de Familia y el estudiante en la toma de decisiones.

LOS *DERECHOS* DE LOS PADRES BASADO EN LAS GARANTÍAS DE PROCEDIMIENTO DEBIDO LEGAL DE ACUERDO A IDEA

9

- ▶ **A participar:**
 - ▶ Referencia a Educación Especial
 - ▶ Participación en el desarrollo del IEP
 - ▶ Informarse
- ▶ **A recibir notificación previa por escrito:**
 - ▶ Cualquier cambio en el IEP
 - ▶ En su Lenguaje Nativo
 - ▶ Cualquier inicio o rechazo de petición



LOS *DERECHOS* DE LOS PADRES BASADO EN LAS GARANTÍAS DE PROCEDIMIENTO DEBIDO LEGAL DE ACUERDO A IDEA

10

▶ A dar consentimiento:

- ▶ Evaluaciones
- ▶ Cambios de servicios

▶ A rehusar consentimiento:

- ▶ Evaluación
- ▶ Ubicación de programa

- A que le proporcionen una evaluación que no discrimine
- A recibir evaluaciones educativas independientes.

LOS *DERECHOS* DE LOS PADRES BASADO EN LAS GARANTÍAS DE PROCEDIMIENTO DEBIDO LEGAL DE ACUERDO A IDEA

11

- A tener acceso a los expedientes educativos.
- A permanecer en el mismo programa si no hay un acuerdo sobre ubicación. Hasta que la disputa se resuelva.
- A que le den audiencia (*hearing*) si hay desacuerdos sobre el IEP.
- A recibir mediación.
- A presentar una queja contra el distrito escolar. Si creen que el distrito escolar ha violado la ley.

LOS *DERECHOS* DE LOS PADRES BASADO EN LAS GARANTÍAS DE PROCEDIMIENTO DEBIDO LEGAL DE ACUERDO A IDEA

12

- A ser informado sobre la disciplina de la escuela y ubicación alternativa. Se refiere a reglas de expulsión y suspensión.
- A ser informado sobre las políticas de atender escuelas privadas. Los distritos escolares son responsables de identificar, localizar y evaluar estudiantes con discapacidades que están matriculados en escuelas privadas por sus padres. Sin embargo, las escuelas no están obligadas a proporcionar educación especial o servicios relacionados. Aún así algunas escuelas privadas y estudiantes puede que reciban servicios del distrito escolar.

IEP SU DEFINICIÓN Y PROPÓSITO

PLAN DE EDUCACIÓN INDIVIDUALIZADO (*INDIVIDUALIZED EDUCATIONAL PLAN*)

- ▶ Es un plan de educación individualizado hecho por escrito, en el cual se describe el programa educacional del estudiante.
- ▶ El propósito del IEP es preparar al estudiante para que tenga una vida adulta lo más independiente y productiva posible.

Findings, IDEA 2004 ©(5)(A)



PROCESO DEL IEP

- Identificar área o áreas de necesidad del estudiante;
- Evaluar/Valorar las necesidades;
- Determinar el criterio de elegibilidad;
- Desarrollar el IEP;
- Observar el progreso;
- Revisar y actualizar el IEP;
- Finalizar la Educación Especial

Servicios Relacionados

(Related Services)

- ▶ A los estudiantes con discapacidades se les debe proveer con lo que ellos requieran para que aprovechen su programa de educación especial.
- ▶ Es necesario recordar que la educación para los niños con discapacidades incluye el área académica, así como también instrucción adecuada para que vivan de forma independiente.



Servicios Relacionados

(Related Services)

16

- Visión (*VI / TVI*)
- Maestra de Sordos e Impedimentos auditivos (*DHH / HES*)
- Terapia de Lenguaje (*Speech*)
- Orientación y Movilidad (*O&M*)
- Terapia Física (*PT*)
- Terapia Ocupacional (*OT*)
- Educación Física Adaptada (*APE*)
- Intérprete (*Interpreter*)
- Interventor (*Intervener*)
- Tecnología Asistida (*Assistive Technology*)
- Escuela de Verano (*12 months program*)
- Manejo de Conducta (*Behavior management*)
- Consejería (*Counseling*)
- Enfermera (*Nurse*)
- Servicios Recreativos (*Recreation*)
- Fondos para discapacidades de baja incidencia (*low incidence funds*)
- Transporte (*Transportation*)
- Instrucción en el hogar (*home instruction*)

Servicios Relacionados

(Related Services)

- ▶ **Instrucción individual o instrucción en grupo**
- ▶ **Tiempo de Instrucción**
- ▶ **Tiempo adicional de terapistas para incluir en el IEP:**
 - ▶ Para proveer consultación con el equipo educativo del estudiante
 - ▶ Para reuniones de equipo
 - ▶ Para entrenamiento de equipo

OTRAS AREAS IMPORTANTES DEL IEP 18

- Identificación
- Evaluación/es
- Elegibilidad
- Equipo educativo (*IEP team*)
- Trabajando en conjunto

IDENTIFICACIÓN

19

▶ Reconocer las necesidades del estudiante (Niño/jóven adulto)

- ▶ Doctor
- ▶ Terapeuta
- ▶ Maestro
- ▶ Padre

▶ Solicitar la evaluación/valoración:

- ▶ Por escrito
- ▶ Mencionar las posibles áreas de necesidad
- ▶ Es una carta formal muy breve

Se identifican las áreas de necesidad

EVALUACIÓN

20

- ▶ **Noticia Anticipada por escrito** (*Prior written notice*)
- ▶ Tipo de evaluación/valoración:
 - ▶ Inicial
 - ▶ Re-evaluación
 - ▶ Independiente (*IEE*)
- ▶ **Consideraciones especiales:**
 - ▶ Evaluadores
 - ▶ Métodos apropiados

Tiempo: 30 días hábiles para valorar y recibir el reporte

EVALUACIONES

De tipo funcional:

- Comunicación
- Visión y/o audición
- Parte académica
- Competencias sociales
- Destrezas de vida diaria
- Ocio y recreación
- Destrezas motoras finas y gruesas
- Orientación y movilidad
- Destrezas vocacionales e intereses

Informales:

- A través de entrevistas
- Evaluaciones de portafolio
- Revisando tareas de su hijo y su trabajo en la clase

PREGUNTAS ANTES DE LA EVALUACION 22

1. **Quién va a realizar la evaluación?**
2. **Cuál es su formación y experiencia?**
3. **En qué ambiente se realizará?**
4. **Qué áreas se evaluarán/valorarán?**
5. **Qué pruebas específicas o partes de las pruebas se utilizarán y por qué?**

ELEGIBILIDAD

23

- ▶ Entendiendo y usando el resultado
- ▶ Criterio y categoria de elegibilidad.

EQUIPO EDUCACIONAL (*IEP TEAM*)

24

- Padre y madre (pueden traer un consejero u otro padre de familia)
- Estudiante (si es apropiado)
- Maestros de educación especial, y de educación general (si es apropiado)
- Proveedores de Servicios Relacionados
- Representante del Distrito:
 - Que conozca los recursos y la manera de utilizarlos
 - Que tome decisiones
- Otros expertos

TRABAJANDO EN CONJUNTO

Los padres y maestros/profesionales tienen la meta de trabajar conjuntamente como iguales para desarrollar un plan educacional apropiado.

- El padre (y el estudiante) tienen una participación activa
 - Usan sus habilidades
 - Usan sus derechos
 - Aceptan su responsabilidad
- Cooperacion:
 - Trabajan en conjunto para desarrollar un programa educacional significativo

FIRMAS

26

- Una vez terminado el IEP, Ud. tiene el derecho de leer detenidamente el documento en ese momento, o llevárselo a su casa y leerlo con calma;
- Una vez terminado el IEP, Ud. tiene 14 días para firmarlo o rechazarlo;
- Si Ud. no lo firma después de los 14 días se considera como aceptado y se implementa;
- Si Ud. rechaza el IEP, tiene 10 días hábiles para que se lleve acabo una reunión de conciliación entre la escuela y Ud.

PERIODOS DE TIEMPO

27

- **Petición para una evaluación del estudiante:**
 - Formal, por escrito y describiendo áreas de necesidad;
 - Sin tiempo establecido, sólo razonable (10 días hábiles para responder)
- Una vez firmado el consentimiento para la evaluación:
 - 30 días hábiles del día que se firmó el consentimiento para hacer la evaluación y recibir reporte
- Una vez firmado el IEP
 - 14 días para aceptar o rechazar el IEP

DESACUERDOS Y DIFERENCIAS

28

Cuando hay diferencias y desacuerdos hay maneras de solucionarlos:

- ▶ **Métodos informales:**

- ▶ Reuniones con el personal de la escuela

- ▶ **Métodos formales:**

- ▶ Conciliación o consejería
 - ▶ IEP facilitado
 - ▶ Mediación
 - ▶ Procedimiento debido legal (*due process*)
 - ▶ Quejas formales si la escuela no está acatando las leyes de educación especial



NEGOCIAR ES UNA FORMA SALUDABLE DE LLEGAR A UN ACUERDO

- ▶ Padres son los expertos en la vida de sus hijos y no los pueden ayudar a triunfar solos!
- ▶ Padres no necesitan estar siempre de acuerdo con lo que se les ofrece
- ▶ Padres pueden tener preferencias o/y hacer sugerencias
- ▶ Padres pueden decidir lo que es apropiado
- ▶ Padres pueden tener mayores expectativas que otros

SIEMPRE Y CUANDO

***HAYA COMUNICACION ABIERTA CON RESPETO, CONFIANZA
E IGUALDAD EN LAS OPINIONES MUTUAS***



5 REGLAS DE ORO PARA UNA SALUDABLE NEGOCIACION

1. Escuche MAS, hable MENOS
2. Haga preguntas para clarificar las perspectivas y las posiciones del otro lado
3. Contar una historia reduce resistencia. Haga su solicitud contando la historia de su hijo
4. Escape la formalidad. Encuéntrense en lugares distintos. Si la situación se vuelve tensa, traiga comida que tenga buen aroma
5. Trate a todos con respeto

TRANSICION A LA VIDA ADULTA

31

- Se inicia a los 14 años o Noveno grado (*9th grade*)
- Se basa en los intereses del estudiante y habilidades:
 - Ir al colegio o algún entrenamiento especializado
 - Trabajar
 - Vida adulta independiente
 - Recreación & Socialización
 - Participación en la comunidad

CULMINACION

- ▶ **Culminación de Educación Especial**
- ▶ **Cuando ya no es elegible**
- ▶ **Por causa de la edad**
- ▶ **Graduación**

1. **Enrolar al estudiante en Educación Especial**
Si sospecha de alguna posible área de necesidad
2. **Identificación del área de necesidad**
Reportes médicos, observaciones de familia o escuela
3. **Evaluación apropiada**
Dar consentimiento de Evaluar
Por un profesional experto en el área
En su lenguaje natal
En su entorno natural
4. **Desarrollar un IEP**
Basado en las necesidades del estudiante



1. Miembros del equipo del IEP

Padres, Maestro de Educación Especial, Terapistas, Representante del Distrito, evaluadores, estudiante, Maestro de Educación regular (si es apropiado)

2. Evaluaciones apropiadas

Para justificar los servicios relacionados

3. Servicios relacionados

Indicando duración y cantidad de veces por semana

Incluir tiempo para que los proveedores de servicios puedan entrenar y consultar con el equipo



Los derechos de la familia

- Las reuniones del IEP se llevan a cabo una vez por año.
- Ud. puede pedir otra reunión si desea cambiar algo.
- Al recibir notificación de la reunión, confirme lugar, día, hora y participantes
- Si Ud. no responde o confirma, la reunión se puede llevar a cabo sin Ud.
- Pida copias de todos los reportes antes a la reunión. Hágalo por escrito
- Si las personas que tienen que tomar decisiones no están presentes el día del IEP, solicite de hacer otra reunión cuando estén disponibles.



Los derechos de la familia

- Ud. puede firmar el IEP en el momento si está completamente de acuerdo o llevárselo a su casa para leerlo detenidamente y después firmarlo.
- Si no firma el nuevo IEP, el anterior sigue en vigencia
- Ud. tiene derecho a traer con Ud. un familiar o consejero que esté familiarizado con las necesidades de su hijo
- Solicite un intérprete si lo considera necesario
- Ud. es un integrante del equipo. Su opinión cuenta en la preparación, repaso y modificaciones del IEP



Ud. conoce a su hijo mejor que nadie, describa:

37

- **Sus fortalezas**

En que ámbito sobresale, cualidades, lo que le gusta, lo que le hace feliz, ...

- **Sus debilidades**

Lo que le incomoda, lo que lo frustra, su sensibilidad a bajas expectativas, ...

- **Su nivel de funcionamiento**

En diferentes medio ambientes, condicionado a la gente que lo rodea, ...

- **Elementos necesarios para mejor aprendizaje**

Consistencia de personal, iluminación, amplificación, proximidad al pizarrón, repetición, material adecuado, tecnología, ..



- Escriba una lista de ideas antes de la reunión
- Hable con la cabeza, no con el corazón
- Mantenga una actitud de colaboración y apoyo
- Haga que sus expectativas sean realizables (reales)
- Sea específico en lo que su hijo necesita para progresar
- Asegúrese que sus preguntas sean contestadas adecuadamente
- Como buen integrante del equipo hable de “nosotros” y no “yo”
- Ofrezca su colaboración cuando sea necesaria
- Esté alerta al comportamiento de otros, no se deje intimidar
- Comparta su idea del presente, y sus esperanzas para el
- mañana



ESTILOS DE COMUNICACIÓN

- **Pasivo**

No habla, se disculpa, dice que si aún cuando no está seguro o no quiere

- **Agresivo**

Habla mucho, quiere salirse con la suya, no escucha, amenaza

- **Seguro de si mismo**

Sabe lo que quiere pero está dispuesto a escuchar otras alternativas



ESTILOS DE COMUNICACIÓN

40

- Sus palabras son irreversibles
- Sea respetuoso, no interrumpa
- Hay mensajes hablados y mensajes corporales, esté alerta al movimiento de su cuerpo
- Cuando hable, mire a los ojos de los otros participantes
- Mantenga un tono de voz bajo y amistoso
- Escuche atentamente, concéntrese en la conversación, no en la respuesta
- Si no entiende algo, pida que se lo repitan



EXPRESIONES EN INGLES E INICIALES

41

- ▶ IDEA – Individuals with Disabilities Education Act
- ▶ FAPE – Free Appropriate Public Education
- ▶ LRE – Less Restrictive Environment
- ▶ IEP – Individual Education Plan
- ▶ TVI – Teacher for the Visually Impaired
- ▶ HES – Hearing Education Services
- ▶ EI – Early Intervention
- O & M – Orientation & Mobility
- PT – Physical Therapy
- OT – Ocupacional Therapy
- APE – Adapted Physical Education
- ----
- SPEECH - Lenguaje
- Hearing – Terapia de audio
- DUE PROCESS – Procedimiento Debido Legal

RECURSOS

42

- ▶ PACERS – <http://www.pacer.org>
 - ▶ IDEA - <http://www.idea.ed.gov>
 - ▶ Disability Rights California – <http://www.Disabilityrightsca.org>
 - ▶ NICHY (información obtenida en previos records)
 - ▶ Parent to Parent NYS – <http://www.parenttoparentnys.org/>
 - ▶ FAPE –
<http://www.parentcenterhub.org/repository/definicion-fape/>
 - ▶ LRE - <http://www.parentcenterhub.org/repository/definicion-lre/>
 - ▶ DICAPTA – <http://www.dicapta.com>
- “Isabel Necesita tecnología de Asistencia”
- <http://www.youtube.com/watch?v=7710wToSm8M>

MUCHAS GRACIAS POR SU PARTICIPACION

43

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Can't Stomach This: a discussion on gut motility in CHARGE syndrome including data from recent research.



Dr. Kim Blake, Professor Pediatrics
IWK Health Centre and Dalhousie University
kblake@dal.ca



Chicago, Illinois USA



CHARGE Conference 2009



Objectives

1. Outline the common gastrointestinal (GI) problems in CHARGE syndrome including gastroesophageal reflux, abdominal pain and constipation.
2. Identify the professionals and treatments that are available for the range of GI problems.
3. Present the clinical and basic science research that is taking place at Dalhousie University, Nova Scotia Canada (2013/14) to answer some of the eating and GI motility issues

Let's Rate Your CHARGE's Eating Difficulties Over the Years

OR

Someone you Know with CHARGE Syndrome

0	1	2	3	4
None	A little (reflux, choking, no G or J tubes)	G or J Tube, less than 12 months	G or J tube feeding more than 12 months	Extreme difficulties, one of the biggest problems

CASE HISTORY

4 Major & 3 Minor

MAJOR

C - Coloboma [Left Eye].

C - Choanal Atresia [Right].

C - Cranial Nerves [VII (Right), VIII, IX, XI].

C - Characteristic Ears [Severe SNHL].

MINOR

C - Cardiac - aberrant subclavian artery, bicuspid aortic valve.

C - Characteristic CHARGE face.

D - Developmental delay – balance, expressive speech.



Age 9 Months

Blake et al 1998 CHARGE Association - An update and review for the primary Pediatrician.

Hidden Structural Problems

CASE HISTORY

- Feeding Issues
- Severe renal hydronephrosis
- Abnormal temporal bones

Cochlear implant , age 2 ½

Nissens fundoplication and tonsillectomy, age 3



Eating Issues

- Poor sucking and swallowing
- Velopharyngeal in-coordination
- Gastroesophageal Reflux (GER)



Dobbelsteyn C, Blake KD. 2005. Early Oral Sensory Experiences and Feeding Development in Children with CHARGE Syndrome: A Report of Five Cases. Dysphagia. Vol : 89-100.

MacKenzie at the 2011 CHARGE Conference



MacKenzie had her G-Tube removed prior to the conference and was feeling sick to her stomach and worried that she couldn't vomit.

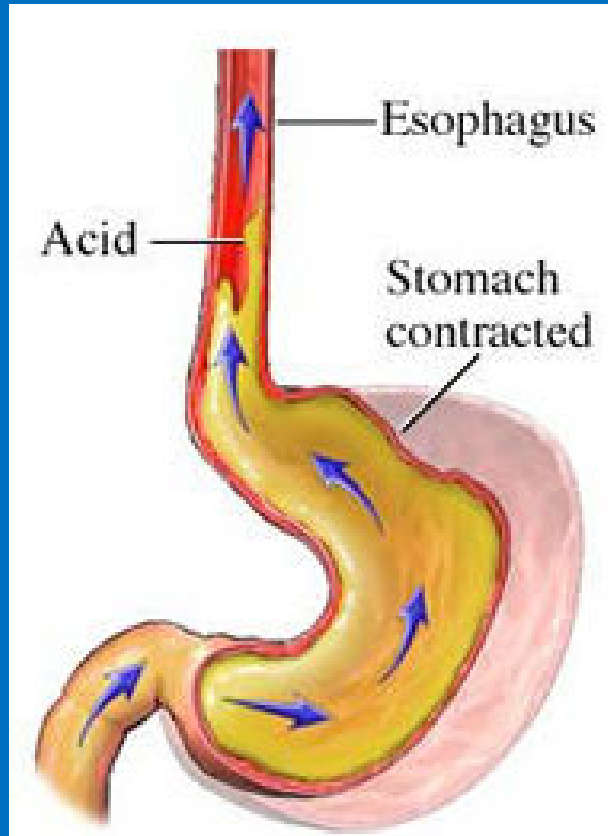
Feeding Question #1

"My 2 year old has been getting more picky and will not eat lumps. We never needed a tube but she's losing weight and now has regular hiccups. She was on ranitidine (Zantac) as an infant but we weaned her off this."

The family doctor feels that this is just the terrible two's and not to worry.

Cindy Dobbelsteyn, et al. Feeding Difficulties in Children with CHARGE Syndrome: Prevalence, Risk Factors, and Prognosis. Dysphagia. 2008 Vol. 23, No. 2, p. 127

Gastroesophageal Reflux

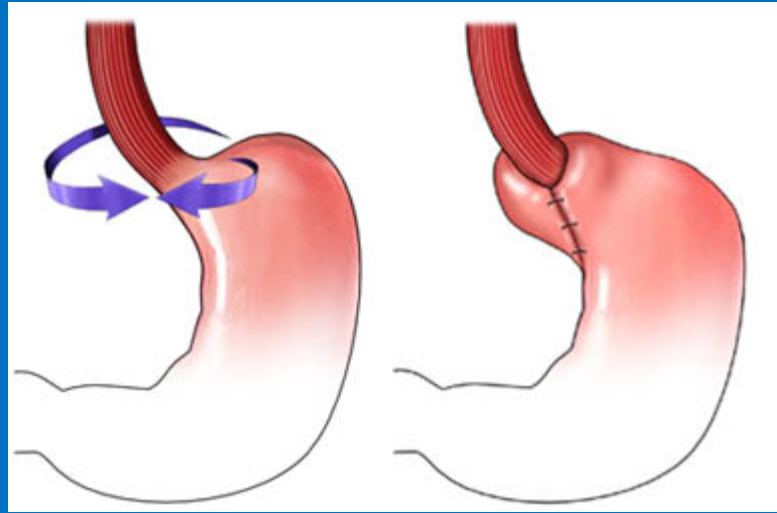


Treatments for Gastroesophageal Reflux (GER)

1. Behavioral treatment – raising the bed, small frequent meals, limiting foods that promote reflux such as tomatoes, meat, chocolate.
2. Medical management
 - Ranitidine 8mg/kg per day in 2-3 divided doses (for babies 3-4 divided doses)
 - Prevacid (lansoprazole)- 1-2 mg/kg per day at the beginning of the day (occasionally twice a day)
 - Domperidone (Motilium) – 4 times a day before meals (watch for side effects)
 - Cisapride (Propulsid) special authorization

Also consider cow's milk protein intolerance

Fundoplication - Surgery



<http://uvahealth.com/>

But is the problem more than just reflux?

Feeding Question #2

“How can she Eat a Hot Dog in 3 Seconds Flat?”



Ate quickly and swallowed without chewing

The Cranial Nerves and Swallowing

Sensory IN



IX Glossopharyngeal – Taste

V Trigeminal – sensation in the palate, upper lip, jaw, mouth, and tongue.

Motor OUT

X Vagus – Swallow, visceral

XI Spinal Accessory – moves head and shoulders, laryngeal muscles

IX Glossopharyngeal – Salivation and swallow

V Trigeminal – Muscles of mastication (chewing)

XII Hypoglossal – moves tongue

Feedback Loop

Cranial Nerves

These guys direct the traffic & run the show

Name	What It Does
I Olfactory	Smell
II, III, IV, VI	Eye control
V Trigeminal	Chewing, sensory for facial regions; sensations in the sinuses, the palate and the upper lip, the jaw, mouth and tongue.
VII Facial	Facial movements, taste, salivation
IX Glossopharyngeal	Taste, salivation, swallow; some visceral
X Vagus	Phonation, swallow; important visceral
XI Spinal Accessory	Moves head & shoulders; laryngeal muscles
XII Hypoglossal	Movement of the tongue

How Many of You Have CHARGE's with Suspected Cranial Nerve Problems?

No

1

2

3

More

CHARGE hands up



Feeding Question #3

After gastrostomy removal some children cram their mouths with food, why?

- Oral hyposensitivity
- Need for substantial amount of food in mouth before bolus preparation occurs



Two friends having lunch

Experiences with Feeding and Swallowing Mouth Over-stuffing and Pocketing of Food



Alex & MacKenzie at the IWK, 2015

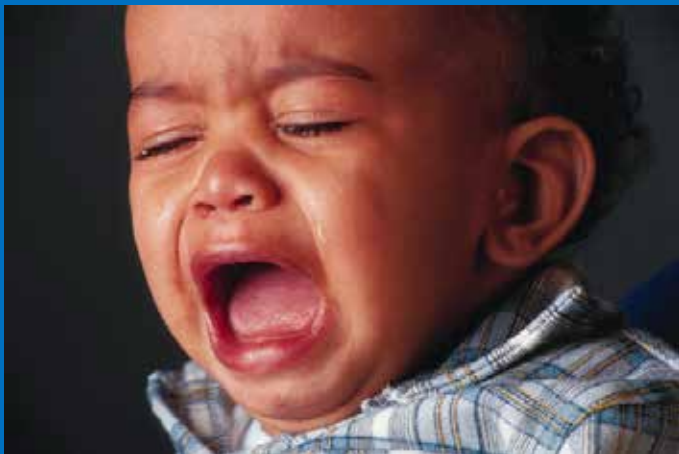
Mouth Over-Stuffing and Food Pocketing

- Parents of children with CHARGE syndrome
- Children over-stuff their mouths or pocket food in their cheeks when they eat
- 45 minute interview & Feeding/Swallowing Impact Survey
- Interviewed 16 parents of children aged 2 – 32 years old

IWK Study 2015 - 2016

Parent's Highlighted Issues

- Increased risk of choking
- Have to have someone with the child when eating
- Increased time to finish eating
- Speech and feeding therapy has not addressed mouth over-stuffing or food pocketing
- Over stuffing can begin at any age



Things to Avoid

- Chewable or solid foods
- Really hard vegetables and fruit
- Dry food (e.g. a muffin)
- Bread and pasta
- Putting too much food on their plate at one time



Parent's Tips & Tricks

- Remind to chew and swallow and finish what's in their mouth – then take more from plate
- Use a water or liquid chaser while eating
- Use favorite foods as incentives to eat other foods
- Serve food textures that work well (e.g. purees)
- Have puree and solid food options at the same meal
- Cut food into really tiny pieces
- Use a smaller spoon



Parent's Tips & Tricks cont'd

- Have your child eat with you at the normal table
- Use an iPad or TV show to distract while eating
- External pacing / therapist input
- Give one item / one bite at a time



Two Main Themes

Sensory

“Yes, often I have her come home from school on the bus and I find bits of whatever she’s had for snack at school in her cheeks.”

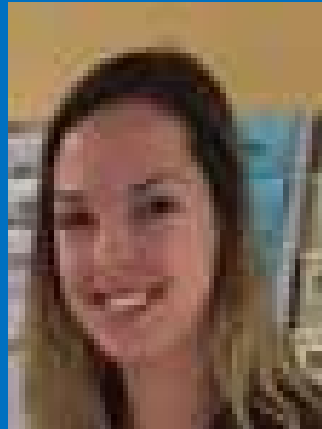
Psychological

“Because she is too smart for her own good, giving her a water chaser...is ineffective because she swallows the water around the food”

Please Visit Alex at Her Poster

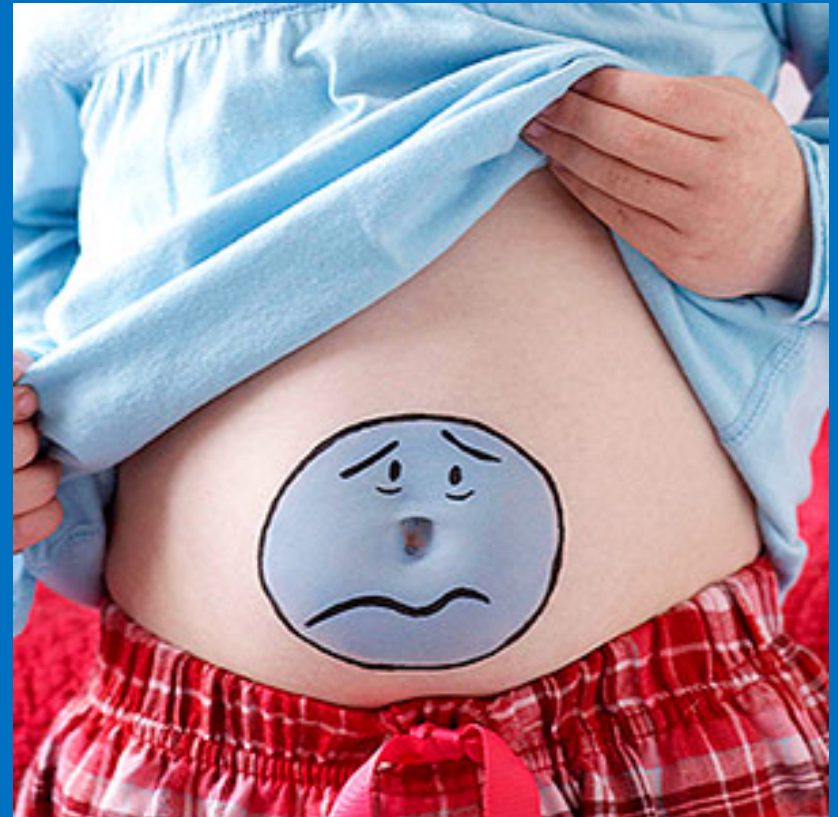
OR

email Alex at dalmed.charge@gmail.com



Abdominal Pain

- Reflux
- Bloating
- Difficulty with digestion
- Abdominal migraine
- Constipation
- Non organic



Treatment Suggestions

- Triggers for migraine
- Venting G-Tubes
- Massage
- Diet
- Motility agents

Experience with Feeding and Gastrointestinal Motility in Children with CHARGE Syndrome



Meghan & Kim at the Research in Medicine (RIM) Presentations at Dalhousie University 2015

Questionnaires

- Structural abnormalities
- Motor impairment
- Oral sensory impairment
- Delayed oral feeding
- Reflux
- Bloating
- constipation



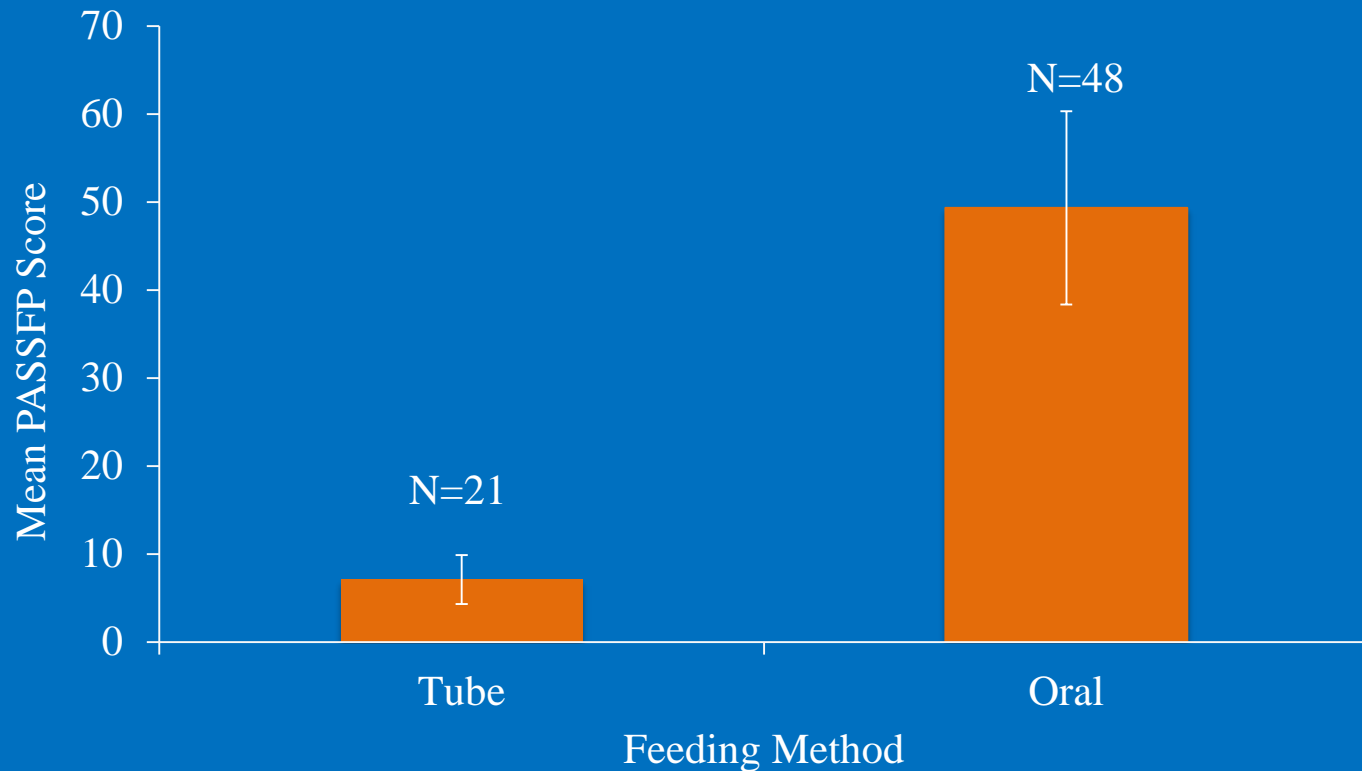
Questionnaires + Short Answer Questions

- Questionnaires include:
 - Demographic and CHARGE characteristics
 - Pediatric Assessment Scale for Severe Feeding Problems ©IWK
 - PedsQL™ Gastrointestinal Symptoms Scale
- Short Answer Question Topics:
 - Reflux and vomiting
 - Bloating
 - Constipation
 - Transition from tube feeding to oral feeding
 - Three major feeding/motility challenges

Preliminary Results

- Participants: 73 completed
 - Age range 1-18y (avg. 7.76 y), 60% Female, 40% Male
 - Age of diagnosis: in utero – 2 years
 - Country: North America (31), Europe (27), NZ/AUS (9), Asia (1), Unknown (2)
 - Gene CDH7
 - Positive 66%
 - Negative 9%
 - Not tested 25%

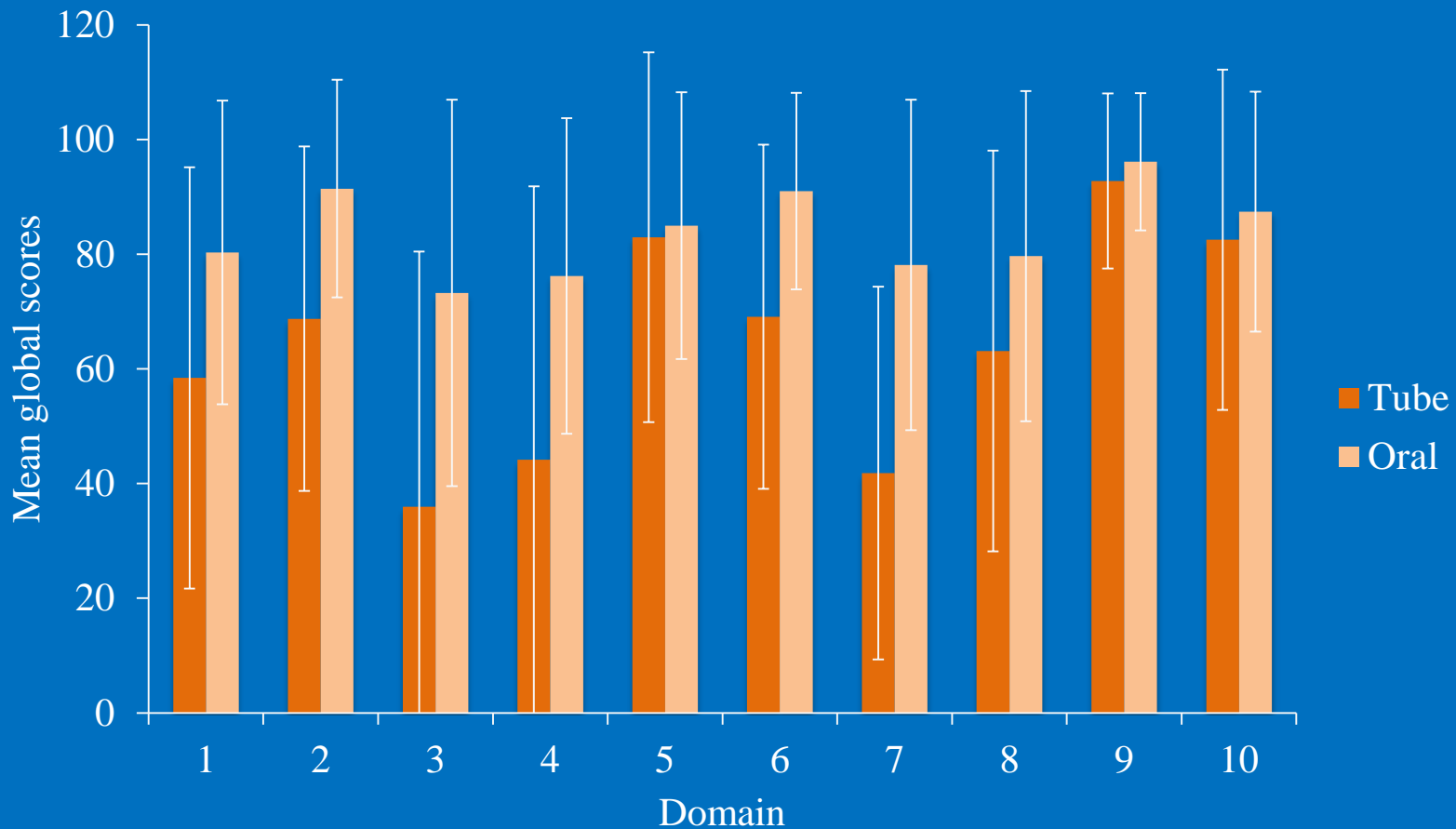
Pediatric Assessment Scale for Severe Feeding Problems (PASSFP)



Lower score indicates more severe feeding difficulties (range 6-61)

PedsQL Gastrointestinal Symptoms Scale

Lower score indicated greater GI symptoms

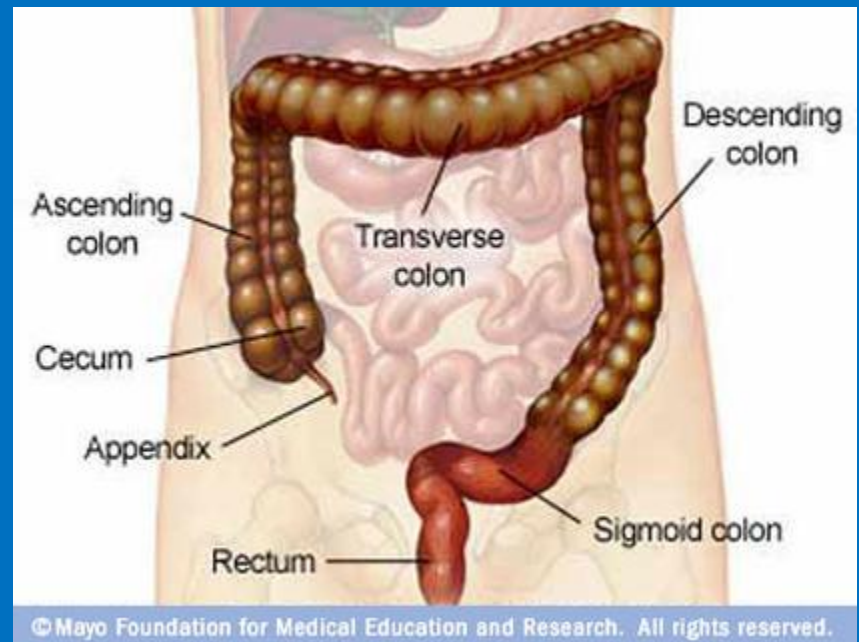


Domain: 1 Stomach Pain
2 Discomfort when eating
3 Trouble swallowing
4 Food and drink limits
5 Heartburn and reflux

6 Nausea and vomiting
7 Gas and bloating
8 Constipation
9 Blood in poop
10 Diarrhea

Constipation – Big Issue

More data to be presented at
the Conference



Prevention / Treatment for Constipation

Prevention:

- Fluids
- Exercise
- Behavioural therapy
- diet



Treatment:

- Polyethylene glycol / MiraLAX
- PEG
- Senocot
- Behavioural techniques

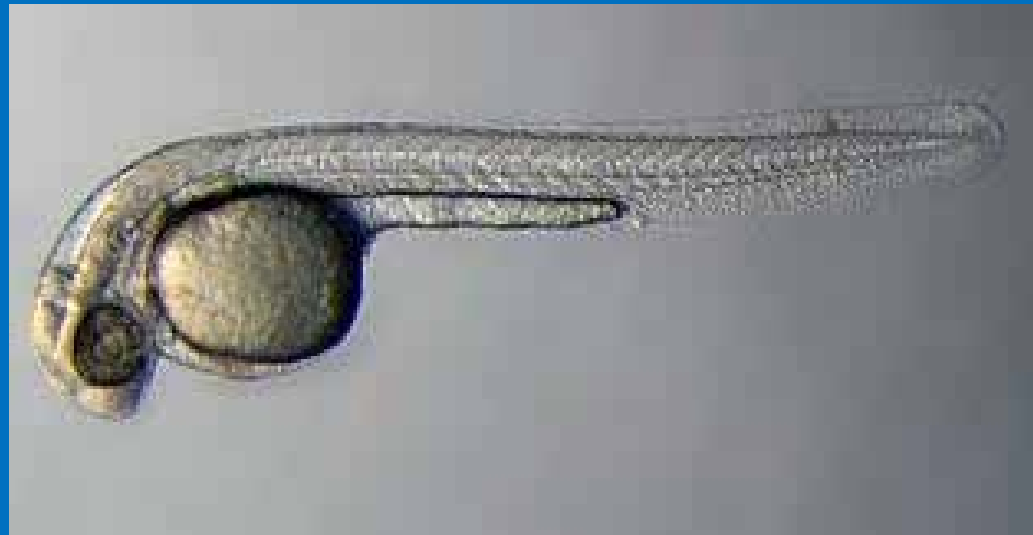
Modeling CHARGE Syndrome in Zebrafish: A Look at the Innervation and Function of the Gastrointestinal System



Kellie Cloney presenting at the Dalhousie Research in Medicine (RIM) 2015. Award for Outstanding Platform Presentation.

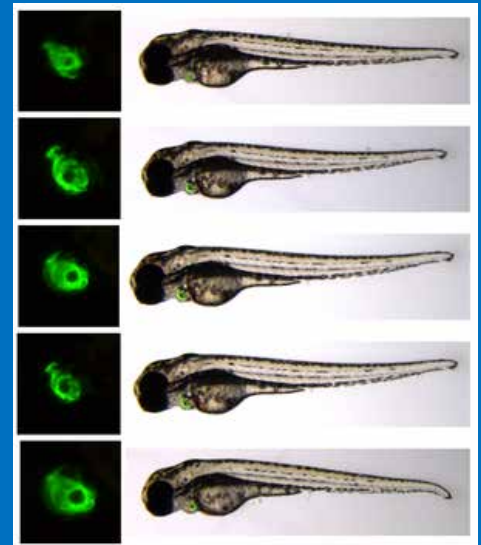
The Zebrafish

- Zebrafish make an excellent model organism to study rare pediatric single gene diseases because:
 - Conserved genetics
 - Ease of genetic manipulation
 - Embryonic transparency
 - Rapid development



Zebrafish and CHARGE

- CHD7 gene is conserved in the zebrafish
- CHD7 knock down has demonstrated the following physiological effects in the zebrafish¹:
 - Dysmorphic heart
 - Smaller eyes
 - Curvature of the body axis
 - Disruption in the number, organization, and patterning of the cranial nerves (mainly V, VII, and X)



Research at IWK 2014 - 2016

- Teaming up with Dr. Berman, who has expertise in modeling rare diseases in zebrafish, we are exploring three main areas of CHARGE syndrome:
 1. Gut motility and function:
 2. Heart anomalies and genetics
 3. Cranial nerve anomalies

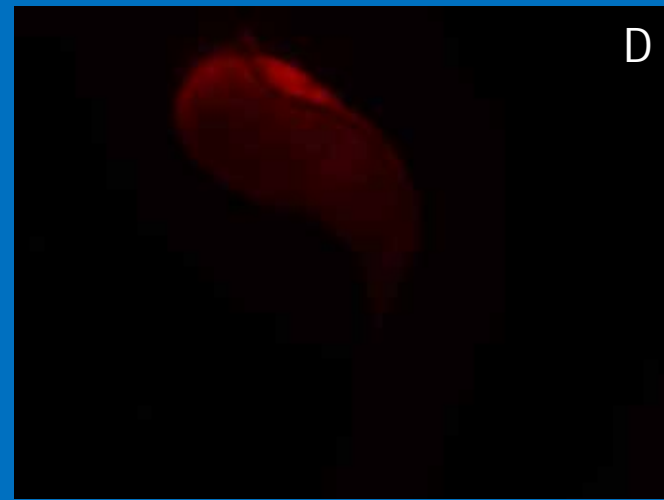
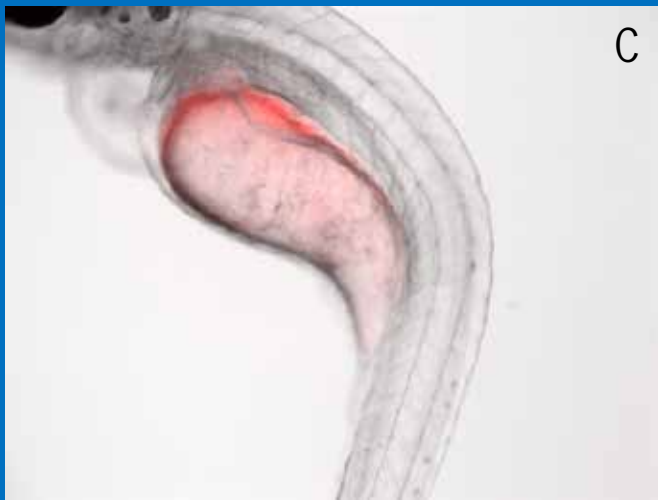


Objective 1: Nile Red Motility Study

Results from Kim

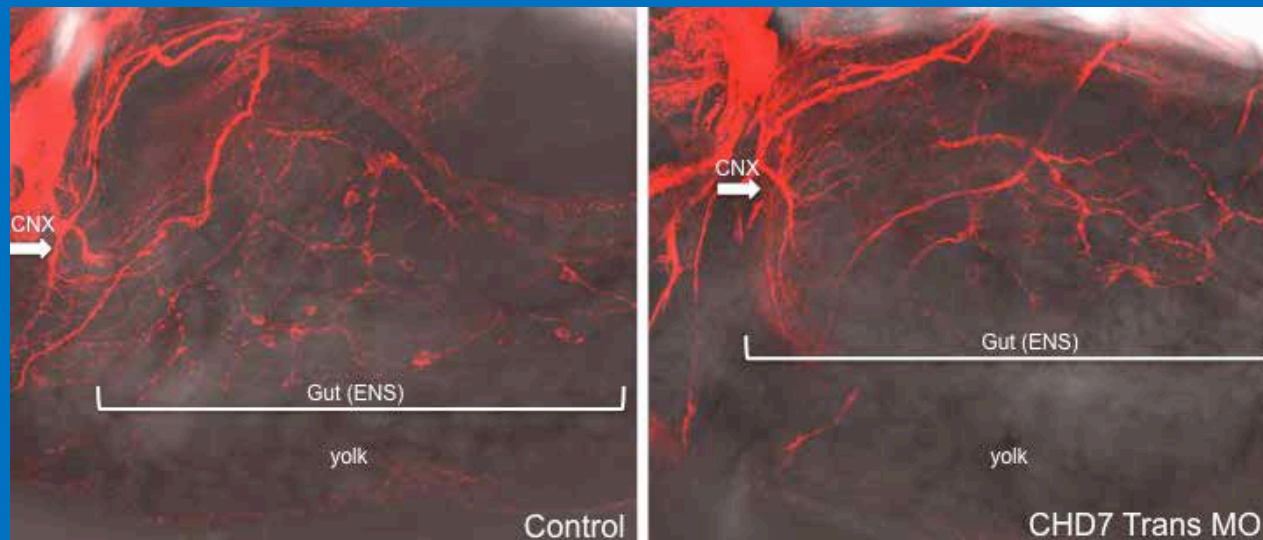


Nile Red Motility Study – CHD7 Morpholino



Immunohistochemistry

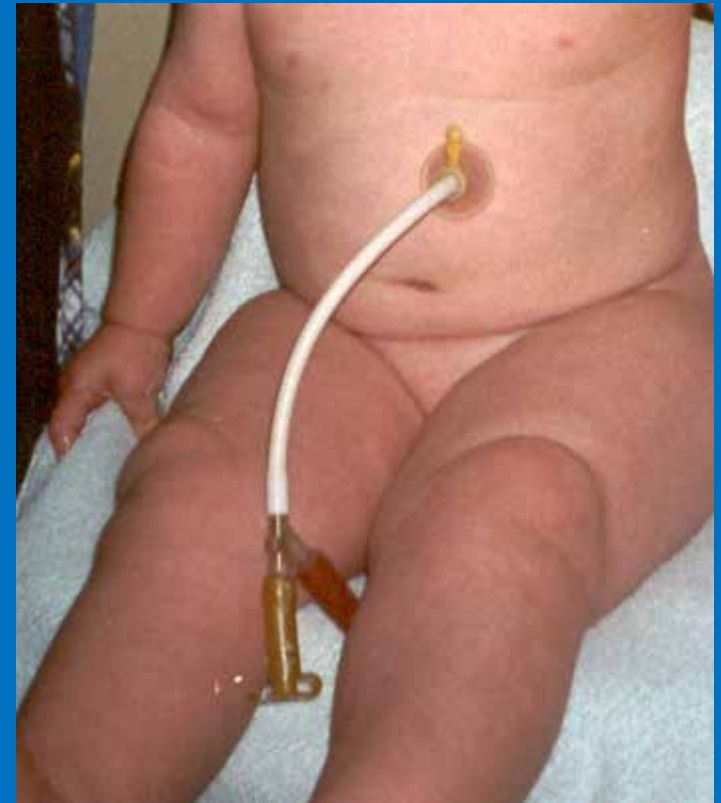
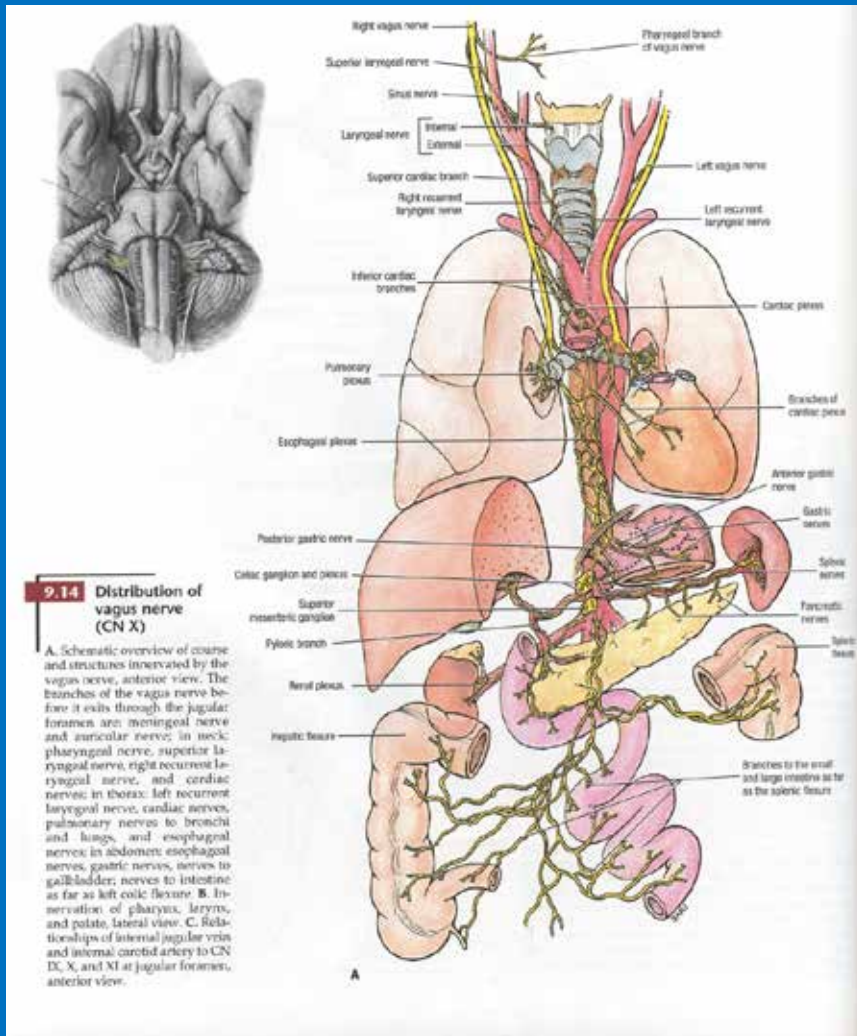
- Early results demonstrate changes in the enteric innervation of the gastrointestinal track.
- Changes in the ENS could lead to altered gut motility



How will our Research Affect Individuals with CHARGE Syndrome

- More emphasis on the gastrointestinal system (gastroenterologist feeding team)
- Treatment to enhance motility of the gut
- Therapists with an understanding of the over-stuffing and pocketing phenomenon

From the Zebra Fish Study we are Closer to Proving that the Vagus Nerve is Abnormal in CHARGE Syndrome



Cranial Nerve X Vagus

Thank you!

To Our Young
CHARGE
Researchers
and You!



Questions: