



11th International CHARGE Syndrome Conference
Scottsdale, Arizona 2013

11th International CHARGE Syndrome Conference Friday, July 26, 2013 Program Handouts



Category: Family Support

**Friday
Breakout Session #1:1 10:45-11:45
Palomino 1 & 2**

**Presentation Title
“Sexuality Issues for Children and
Adolescents with CHARGE”**

**Kasee Stratton, Ph.D.
Kennedy Krieger Institute/Johns Hopkins
University School of Medicine**

Presenter Information:

Dr. Kasee Stratton has been researching and working with children and young adults who have CHARGE for the past 7 years. She is a previous student of Dr. Timothy Hartshorne. Dr. Stratton's primary research and clinical interests include: reducing challenging behaviors, teaching appropriate adaptive skills, identifying non-vocal pain behaviors, and reducing the pain experience for individuals with CHARGE. Kasee is currently finishing her Post-Doctoral Fellowship at the Johns Hopkins University School of Medicine and the Kennedy Krieger Institute. She plans to continue her work with CHARGE following completion of her fellowship and hopes to open a CHARGE clinic in the near future.

Presentation Abstract:

This presentation is important for individuals of ALL AGES. The presentation will highlight the need to establish appropriate social and safe behaviors early (during childhood) regarding the developmental of sexuality. Research will be presented on the need for sexuality education and direct teaching due to high rates of sexual violence against individuals with developmental delay. Topics will include physical examinations, identifying body parts, teaching modesty, how to shower, menstruation, masturbation, and resources for sex education. This presentation has been designed specifically for individuals with CHARGE.

**3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013**

Sexuality and Young Adult Concerns for Individuals with CHARGE

11th International CHARGE Syndrome Conference

Kasee Stratton, Ph.D.
Post-Doctoral Fellow
Kennedy Krieger Institute
Johns Hopkins School of Medicine

Outline

- Importance of prevention and education
- Consideration of short- and long-term goals
- Identifying body parts and gender
- Identifying boundaries
- Hygiene and Personal Care
- Menstruation
- Masturbation
- Sex Education
- Odds and Ends

Where do we begin? And WHY?

"Not teaching a child about sexuality and how to express natural urges in a safe and appropriate manner denies a big part of what is essentially human." (Moss & Blaha, 2001).

Embarrassing moment or teaching moment

- Children with DB and DD do NOT learn incidentally about sexuality issues.

Importance?

- Multiple studies indicate that children who are Deaf know less about sexuality issues than hearing peers (Getch et al., 2011)
- 2.2 times higher for sexual abuse
- Often repeated and will go unreported (Stinson, Christian, & Dotson, 2002)

References: National Center for Abuse and Neglect

Sexual Abuse Research

Males

- About 13.9% vs. 3.7% reported lifetime sexual violence
- 32% victims of sexual assault


Females

- 26.6% vs. 12.4% experienced lifetime sexual violence
- Abuse estimated to range from 33% to 83%
- 83% victims of sexual assault (less than half seek treatment or legal support)


References: American Journal of Preventive Medicine 2000, 15 (1); Stinson, Christian, & Dotson, 2002; Murphy & Ellis, 2006

Goals

- Begin teaching EARLY!
- 6 month
- 1 year
- 5 year
- Is the location (environment) appropriate?



NAMING BODY PARTS



Body Parts

- Use daily teaching when appropriate
- Begin Early
- Be Specific
- *Recommendations:*
 - Games: “Is it a boy or a girl?”
 - Books, magazine clippings

















ESTABLISHING PERSONAL BOUNDARIES

Personal Boundaries

- Establish a pattern of behavior.
 - More difficult to change highly established behaviors
 - Identify examples of safe and not safe hands
- Model Modesty
 - REQUIRE modesty from teachers, physicians, etc.
 - Opportunities for practice
 - Teach for the environment




Establishing Boundaries

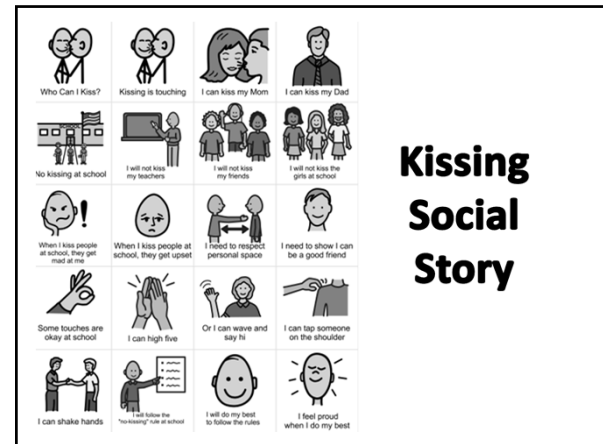
- State specific people who can see the child without clothing
- Who can assist with baths?
- Examples of private and not-private
- Prep for physical examinations

 Being naked means wearing no clothes NOT my friends!	 Who can see me naked? Who can see my underwear? Who can see my private parts? NOT other students!	 Sometimes my doctor needs to see my body NOT other grown-ups!	 It's okay for Mom or Dad to see me naked! My body is private
 I can be naked in my bedroom. It is private.	 I can be naked in my bathroom. It is private.	 But I should close the door first	 Being naked is private
 No one at school should see my underwear or my private parts!	 I don't take off my clothes at school	 I don't pull my shirt up	 I don't pull down my pants
 Sometimes I need to fix my underwear	 Sometimes my clothes feel uncomfortable	 I can ask to go to the restroom	 At school I can fix my clothes by myself in the restroom

Safe Hands

- Use a consistent vocabulary
- Forewarn your educators
- Perseverative touch; redirect when able and address with a formal behavior plan



Personal Boundaries

- How do introduce yourself to others
- Name
- Place out hand
 - "Count down" handshake

HYGIENE AND PERSONAL CARE



Hygiene and Personal Care

"A critical component of social and sexual maturity is attaining independence in basic self-care tasks." -AAP

- Intervention Options: direct instruction, formalized cues, and reinforcement system
- Clothing: Acceptable to peers and socially appropriate?
- Add to your child's yearly goals at school

Showering/Bathing

- Assistance- *Did you ask for permission?*
- Use of soap
- Forgotten parts
 - Neck
 - Back
 - Feet
- Caring for Stoma site and other medical necessities



Hygiene Interventions

"First Impressions"

- Male and Female Versions
- Hygiene
 - Importance
 - Shower, Shampoo, Soap
 - "5-minute shower"
 - Toilet Hygiene
 - Self-exams (testicular and breast)
- Social Stories; Laminated Task Lists

<http://www.stanfield.com/products/social-life-skills/first-impressions/>

MENSTRUATION

Menstruation









- Sanitary Pad Compliance
 - Practice, Practice, Practice!
 - Visual scripts (placement and changing)
 - Reinforcement
 - Hygiene
- Choices
 - Sanitary pad type
- Add to IEP







Menstruation








- Prepping for School
- Medications
- Keep record of periods

- Teaching Tool: "Janet got her period."

<http://www.stanfield.com/products/family-life-relationships/other-family-ed-programs/janets-got-her-period/>

 <p>Once, I was a baby.</p>	 <p>Big girls and women have a period that comes once each month.</p>
 <p>Then, I was a little girl.</p>	 <p>When I have my period, I will see blood on my underwear.</p>
 <p>Now, I'm a big girl.</p>	 <p>I will need to wear a sanitary pad inside my underwear so that blood does not get on my clothing.</p>
 <p>Soon, I will be a woman.</p>	 <p>When I have my period, the blood flow will last about 5 - 7 days.</p>

 <p>I will need to change my pad several times during the day.</p>	 <p>If I start my period at home, I can tell Mom or Dad.</p>
 <p>I will go to the bathroom, wrap the dirty pad in toilet paper, and put it in the trash.</p>	 <p>They will go to the bathroom with me.</p>
 <p>I will put a new, clean pad on my underwear.</p>	 <p>At home, my Mom and Dad will help me.</p>

	If I start my period at school, I will quietly tell Ms. Clinton or Ms. Randall		When I have my period, it will be very important to wash between my legs every day to stay clean.
	I will talk quietly because this is a private matter. Friends and other teachers do not need to know.		When I have my period, I might feel a light pain in my tummy. If I do, I will tell my Mom or Dad.
	I will have pads in a bag in my backpack. I will get my bag from my backpack.		That light pain in my tummy is called cramps. My Mom or Dad can give me some medicine if I need it.
	We will quietly walk to the nurse's suite. I can use the nurse's bathroom when I have my period.		

Examinations

- The American Academy of Pediatrics supports having a trusted caregiver present for examinations if the individual provides consent to do so (Murphy & Ellis, 2006)

Birth Control

- Discuss questions and concerns with your child's physician
- Caution: antiepileptic medications decrease the effectiveness of oral and implanted contraceptives (Murphy & Ellis, 2006)
- Injectable contraceptive can effectively minimize or eliminate menstrual flow
 - Prolonged use linked to bone density loss in healthy adolescent females, which may not reverse completely after discontinuation of the medication (Murphy & Ellis, 2006)

MASTURBATION

Masturbation

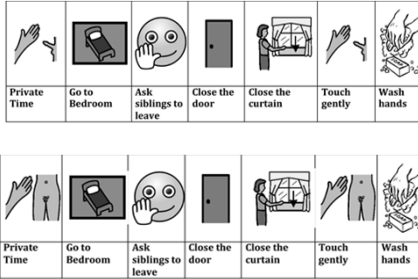
- Prerequisite Skill #1:
 - Does your child understand the concept of "wait"?
 - “Later when we get home”
- Prerequisite Skill #2:
 - Teach public versus private

Masturbation

- LOCATION, LOCATION, LOCATION
 - Redirects; Use of a timer
 - Social Story; Visual Script
 - Reinforcement Systems!
 - Be cautious of items used for humping
 - Gentle touch
- Video Modeling:
 - Males: “Hand Made Love: A guide...”
 - Females: “Finger Tips: A guide...”

(Diverse City Press: Diverse-city.com)

Masturbation



SEX EDUCATION?

Sex Education

- Education and intervention for sexuality allows the individual to knowledgeable to make informed decisions to promote sexual identity and sexual safety (Getch et al, 2001)
- Consult with your IEP team early

Final Notes

- Begin education early
- Physicians and educators must know that sexuality education is primary part of your child's goals
 - National Consortium on DB: "Intro to Sexuality Edu."
- Instructional strategies may require the use of concrete materials and tactual exploration.

Additional Resources

- Sexuality and Developmental Disability:
 - A Guide for Parents, from the Alberta, Canada web initiative teachingsexualhealth.ca, which was developed by educators and health professionals. The website offers up-to-date, evidence-based information and strategies for teachers and educators in four main areas: How To Teach, Lesson Plans, Teacher's Lounge and Resources.
- Introduction to Sexuality Education for Individuals Who Are Deaf-Blind and Significantly Developmentally Delayed—from the National Consortium on Deaf-Blindness.
 - This is much like a curriculum and provides good background and some specific content. While it is written about children and teens who are Deaf-Blind, it is also an excellent general disabilities resource. The chapters include an introduction, guidelines for developing a process for intervention and instruction, and topics such as modesty, appropriate touch and personal boundaries, menstruation, masturbation, sexual health care, sexual abuse, and a resources section. 126-pages

References

- Getch, Y.Q., Branch, D.L., Fitz-Gerald, D., & Fitz-Gerald, M. (2001). A rationale and recommendations for sexuality education in school for students who are Deaf. *American Annals of the Deaf*, 146, 401-408.
- Hingsburger, D., & Tough, S. (2002). Healthy sexuality: Attitudes, systems, and policies. *Research & Practice for Persons with Severe Disabilities*, 27, 8-17.
- Stinson, J., Christian, L., & Dotson, L.A. (2002). Overcoming barriers to the sexual expression of women with developmental disabilities. *Research & Practice for Persons with Severe Disabilities*, 27, 18-26.
- Moss, K., & Blaha, R. (2001). Introduction to Sexuality Education for Individuals Who Are Deaf-Blind and Significantly Developmentally Delayed. DB-LINK.
- Murphy, N.A. & Elias, E.R. (2006). Sexuality of children and adolescents with developmental disabilities. *Pediatrics*, 118, 398-403.
- National Information Center for Children and Youth with Disabilities (Sexuality Education for Children and youth with Disabilities): <http://nichcy.org/pubs/newsdig/nd17txt.htm>
- Sexuality Education for Youth with Disability or Chronic Illness A Resource List: <http://www.med.umich.edu/gourin002/toping/00000000.htm>

Contact Information

Kasee Stratton, Ph.D.
Post-Doctoral Fellow
Kennedy Krieger Institute
Johns Hopkins School of Medicine
stratton@kennedykrieger.org
strat1kk@cmich.edu

Soon to be joining Mississippi State University



Category: Education

Friday
Breakout Session #1:2: 10:45-11:45
Palomino 3

**Strategies for Involvement in Physical
Education & Balance Activities for All**

Dr. Lauren Lieberman
Distinguished Service Professor
SUNY College at Brockport

Dr. Pamela Haibach
Associate Professor
SUNY College at Brockport

Presenter Information:

Lauren Lieberman, Ph.D. is a Distinguished Service Professor in the area of Adapted Physical Education at the State University of New York College at Brockport. She has taught children who are Deafblind at Perkins and is the founder and director of Camp Abilities a sports camp for children who are visually impaired, blind or deafblind. Her area of expertise is Inclusion, and physical activity and sport for children who are visually impaired, blind or deafblind. She has written seven books and many research articles in this area. Dr. Lieberman along with Dr. Pamela Haibach co-direct the recently established The Institute for Movement Studies for Individuals who are Visually Impaired at The College at Brockport. This Institute includes research in the area of motor development, physical education , and physical activity for children with CHARGE Syndrome.

Presenter Information:

Pamela Haibach, Ph.D. is an Associate Professor at the State University of New York College at Brockport. She has conducted research focusing upon children and adolescence with visual impairments and blindness for 8 years and has conducted research and published on balance in children with CHARGE Syndrome. Her area of expertise is motor development and motor learning and has published a first of its kind textbook joining these two fields entitled *Motor Learning and Motor Development* through Human Kinetics Publishers. Recently, she served as the chair of National Association for Sport and Physical Education (NASPE) Motor Development & Learning Academy. She also serves as a reviewer for adjudicated journals in the fields of motor behavior, aging, and visual impairment.

Presentation Abstract:

Physical Education is a direct service and is required by law for every child. Appropriate physical education when implemented correctly can make a major difference in the lives of all children especially children with CHARGE Syndrome. In a recent study at The International Charge Syndrome Conference it was found that many children with CHARGE Syndrome do have physical education, yet many parents were not satisfied with the educational experience provided. Physical Education provided appropriately can offer educational opportunities in all nine areas of The Expanded Core Curriculum. It was clear that the most satisfied parents expressed the need for the paraeducator to be trained for physical education and for the child to have experiences in both inclusive and separate classes during the week.

The purpose of this presentation is to share with parents and families the components of a physical education program that will enhance the lives of children with CHARGE Syndrome. The components shared will be 1) The variables necessary to make the best placement decision, 2) The importance of Pre-teaching before each unit, 3) How all nine areas of the Expanded Core Curriculum can be infused into physical education naturally, 4) Training components for paraeducators, 5) Training components for peer tutoring, 6) Instructional strategies (including the use of interpreters if necessary), 7) Resources to share with the physical education team. The resources will include books, videos, web sites, equipment, products, recent research findings, and summer programs that may be available.

The last part of the presentation will be a discussion time where parents can share their positive and negative experiences with each other and we as a group can discuss what is working and what we want to change in the future. Our hope is to come up with 3-4 points to add to FamilyConnect (through AFB).

PHYSICAL EDUCATION INSTRUCTIONAL STRATEGIES FOR CHILDREN WITH CHARGE SYNDROME: RESEARCH TO PRACTICE

National CHARGE Syndrome Conference, Scottsdale, AZ

Lauren J. Lieberman Ph.D.
llieberm@brockport.edu

Pamela S. Haibach, Ph.D.
phaibach@brockport.edu

Overview of Presentation

- What is Adapted Physical Education?
- Research Results from 2011 CHARGE conference
- What should the children learn?
- What should be involved in training?
- How do we get children moving at home?
- Questions



What is Adapted Physical Education?

- APE is physical education modified to meet the needs of children with disabilities.
- APE can be delivered in an inclusive class, a modified environment or a segregated class
- Each child must be provided PE in the least restrictive environment for them.



Children with CHARGE should be self-determined!

Adapted Physical Education



- Adapted Physical Education is the service provided NOT the placement

Position Statements

• Placement in Physical Education

- AAHPERD.org
- AAPAR
- APAC
- Position Statements



• CEC Position Statement on Physical Education for Children with VI

- Division on Visual Impairment



Results of Study on PE and Children with CHARGE

- Physical Education placement affects children's success and parental satisfaction
- Children with support staff such as a teachers aid, paraeducator or intervenor have a more successful experience
- Communication methods used in classes
- Successful units:
 - swimming, scooters, bowling, fencing, t-ball, dancing, rock climbing, floor hockey, field hockey and gymnastics
- Difficult units:
 - fundamental motor skills



What else do we know?

- Children with CHARGE Syndrome- benefit from appropriate placements and 1:1 instruction (Lieberman, Haibach, & Schedlin, 2012)
- Paraeducators need to be trained (Lieberman, & Conroy 2013)
- Children with visual impairments and deadblindness benefit from pre-teaching (Conroy, 2012)
- Children who are deadblind must have specific communication strategies implemented into lessons (Arndt, Lieberman, & Pucci, 2004)

What should they learn?

- The same curriculum that their aged peers learn. But, may need modifications to the rules, equipment, environment and instruction.



Preteaching

- Preteaching must occur before each new unit and must be well planned
- It is especially important in inclusive settings
- Pre-teaching can be provided by the PE teacher, APE teacher, O & M, TVI Intervenor, Paraeducator, peer tutor, parents or siblings



Tactile Sports Boards

Beep Baseball

Goalball

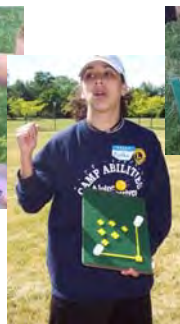


Tactile Sports Boards

Beep Baseball



Tactile Sports Boards



Utilizing Support Staff

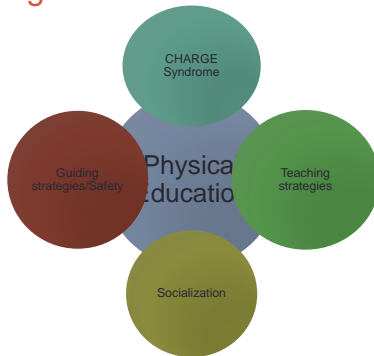
ROLE	TRAINING
<ul style="list-style-type: none"> • Peer tutors • Paraeducators • Intervenor • SSPs 	<ul style="list-style-type: none"> • Must be trained on deafblindness • Must practice skills learned and activities • Must learn appropriate communication modes • Appropriate positioning is key for everyone involved!

What should be involved in the training?

- Description of PE?
- The needs of the child
- Their role
- Teaching Techniques
- Feedback techniques
- Assessment techniques
- Behavior needs



Training Paraeducators



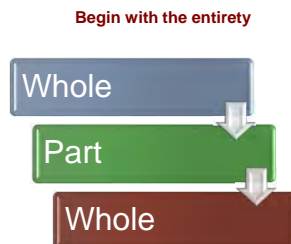
Expanded Core Curriculum

- Independent living skills
- Recreation and leisure activities
- Assistive technology
- Self-determination
- Career education
- Orientation and mobility
- Compensatory or functional academics
- Sensory efficiency skills
- Social interaction
 - ECCADVOACACY.org



Instructional Strategies

- Whole-part-whole
- Physical guidance
- Tactile modeling
- Task analysis
- Communication techniques
- *Will be part of Motor development curriculum through APH



Whole-Part-Whole example

- Explain entire sport of bowling
- Teach component parts
 - Getting the ball
 - Finding the placement on the deck
 - Executing the skill of a roll
- Put it all together again



Tactile modeling

- Allow student to feel instructor or a peer go through motion
- Explain goal ahead of time
- Explain how it fits into larger picture
- Document all physical assistance

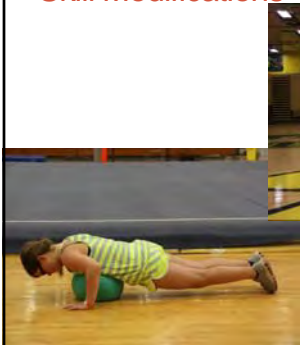


Physical guidance

- Have instructor or peer move the student through a motion with either full assistance or just a tap of a knee or elbow
- Explain ahead of time
- Document all physical assistance



Skill Modifications



Task analyze

- Break skills down into component parts and repeat necessary components with cues
- Be consistent
- Use universal cues

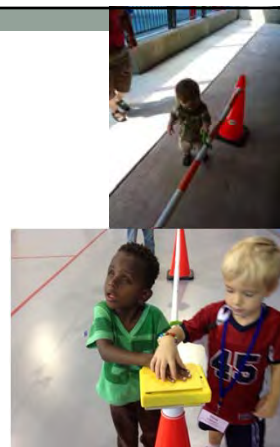


Promote Independent Movement



Equipment for "Count me In" Kit

- APH Sound ball (7.5 inches)
- Intermediate basketball
- Beep-T-ball
- Wiffle ball w bells
- 30-Love Tennis Ball
- batting tee
- aluminum bat
- bean bags (2)
- 28 inch orange cones (2)
- Telescoping safety pole
- Slide ring
- loop rope
- carabiner
- 52 feet of guidewire
- Motivator switch (2)



Modified Equipment



Fetzer Institute Grant

- Motor development video to promote instruction of young children (3-10) with visual impairments
 - Follows the MD curriculum
 - Can be viewed in entirety or as short instructional clips
- Paraeducator Video
 - Paraeducator training video for physical education
 - Includes information on visual impairment
 - Perkins.org e-learning web site



Facilitating Movement

- Activities in PE
 - Movement activities
 - Scooterboard
 - Jump rope
 - Parachute
 - Relay activities (guidewire)
 - Obstacle courses
 - Dance
- Outside
 - Playgrounds
 - Parks
 - Pools
 - Ice skating



Facilitating movement continued

- exploring the playground
 - Swing
 - Slides
 - Monkey bars
 - Bridges
 - Rock walls
- floor time motor games
 - Balls
 - Tug of war
 - Scooters
 - Bolsters
 - Wedge mats
 - Etc.



How do we get young children moving at home?

- Movement games in the house
 - Forts
 - Pillow games
 - Ball rolling
 - Kicking, gross motor activities
 - bowling
- In the yard
 - Locomotor activities
 - Exploration of flowers and plants
 - Visiting neighbors
 - Swings



Expect Achievement!

Do not just expect participation



Modify Equipment

- Add sound
- Add texture
- Brighter balls
- Softer balls
- Balls on strings
- Larger balls
- Add tactile boundaries
- Add bright boundaries
- Lower baskets



Modify Rules

Allow Intervenor peer tutor or 1:1 aid for support and communication

Bat off a tee

Guide runner

No defenders in open sport games

Slow down the game such as scooters or carpet square games

Other?



Modify the Environment

- Ensure clear boundaries
- Modify lighting accordingly
- Decrease excessive sounds
- Increase tactile cues
- Use a multisensory teaching approach
- Other?



Role Models

- Cody Colchado-
 - Paralympic Power lifter
 - Deaf-blind

www.coachcody.org

Who is *YOUR* role model?

Celebrate Successes!

We need to celebrate the successes our children experience as it occurs and cherish what we accomplish!



Promote The Full Potential of Each Child!





Category: Medical/Genetics
(Grant Research Updates)

Friday
Breakout Session #1:3: 10:45-11:45
Palomino 4& 5

**Epigenetic regulation of neural
crest cell development by
Brg1 and Chd7**

Ching-Pin Chang, M.D.,Ph.D.
Stanford University

Presenter Information: CHARGE syndrome, which includes congenital defects in the cardiac outflow tract, is caused by CHD7 mutation. Our studies of Brg1, a chromatin-remodeling factor, demonstrate a molecular interaction between Chd7 and Brg1 to control mammalian fetal heart development. Brg1 and Chd7 are chromatin-regulating factors that structure the epigenome to program gene expression. Within neural crest cells, Brg1 partners with Chd7 on the promoter of *PlexinA2* to program its expression, which is essential for guiding neural crest cells to the heart to control the development of cardiac outflow tract. In addition, Brg1 is necessary for maintaining neural crest cell pool for heart development. These studies thus uncover a new layer of regulation related to the pathogenesis of CHARGE syndrome.

Presentation Abstract: Development of the cerebral vessels, pharyngeal arch arteries (PAAs) and cardiac outflow tract (OFT) requires multipotent neural crest cells (NCCs) that migrate from the neural tube to tissue destinations. However, little is known about how mammalian NCC development is orchestrated by gene programming at the chromatin level. Here we show that Brg1, an ATPase subunit of the BAF chromatin-remodeling complex, is required in NCCs to direct cardiovascular development. Mouse embryos lacking *Brg1* in NCCs display immature cerebral vessels, aberrant PAA patterning, and shortened OFT. Brg1 suppresses an apoptosis factor *Ask1* and a cell cycle inhibitor *p21^{cip1}* to inhibit apoptosis and promote proliferation of NCCs, thereby maintaining a multipotent cell reservoir at the neural crest. Also, Brg1 supports *Myh11* expression for NCCs to develop into mature vascular smooth muscle cells of cerebral vessels. Within NCCs, Brg1 partners with a chromatin remodeler Chd7 on the *PlexinA2* promoter to activate *PlexinA2*, which encodes a receptor for semaphorin to guide NCCs into the OFT. Our studies thus reveal a new role of Brg1 and its downstream pathways in the survival, differentiation, and migration of the multipotent NCCs, critical for mammalian cardiovascular development.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013



Category: Medical/Genetics
(Grant Research Updates)

Friday
Breakout Session #1:3: 10:45-11:45
Palomino 4 & 5

**The Cerebral Cortex in
CHARGE Syndrome**

Robert F. Hevner, MD, PhD
Credentials & Organization
Professor, University of Washington and
Seattle Children's Research Institute

Presenter Information:

Dr. Hevner is a pediatric neuropathologist at Seattle Children's Hospital. His lab studies development and malformations of the cerebral cortex, using mice as a model system. Dr. Hevner obtained his B.S. in Cellular and Molecular Biology (with High Honors) from the University of Michigan, and M.D. and Ph.D. degrees from the Medical College of Wisconsin. He completed residency in Anatomic Pathology at Brigham and Women's Hospital, and fellowship in Neuropathology at Stanford University. He then performed postdoctoral research at UCSF. He has been at the University of Washington since 2000, and at Seattle Children's Research Institute since 2008.

Presentation Abstract:

CHARGE syndrome is caused in most cases by mutations in *CHD7*, a gene that is highly expressed in many areas of the developing brain, including the cerebral cortex. The cerebral cortex may develop abnormally in CHARGE syndrome, contributing to cognitive and behavioral problems in some affected individuals. To investigate this possibility, we have studied mice with *Chd7* gene mutations to evaluate cortical development. The cortex in these mice indeed shows multiple anomalies of cortical gene expression during development, including defects of neuronal differentiation and cell migration. Both projection neurons (excitatory neurons with long axons) and interneurons (inhibitory neurons with short axons) are affected. Our findings suggest that cortical development may be perturbed in CHARGE syndrome and contribute to disease symptoms.



Category: Medical/Genetics
(Grant Research Updates)

Friday
Breakout Session #1:3: 10:45-11:45
Palomino 4 & 5

**Identification of Molecular Markers to
Predict Auditory Neuron Function for
CHARGE Syndrome**

Kelvin Y. Kwan
**Assistant Professor of Cell Biology &
Neuroscience**
Rutgers University

Presenter Information:

Kelvin Kwan is an Assistant Professor in the Department of Cell Biology and Neuroscience at Rutgers University. His lab is located in the Rutgers University Stem Cell Center. Dr. Kwan's lab is interested in regenerating the sensory and nerve cells of the inner ear. He is working closely with the Rutgers University Cell and DNA Repository, the largest university based cell and DNA biobank, to generate induced pluripotent stem cells from archived cells.

Presentation Abstract:

Encased in a bony labyrinth, the cochlea residing within the inner ear allows us to discriminate and hear complex sounds. Hair cells in the cochlea are the sensory cells that convert sound into neural signals, which are then relayed to the brain by auditory neurons. Patients with CHARGE are frequently affected by sensorineural hearing loss resulting from hair cells or auditory neuron dysfunction. Currently, the only treatment for hearing loss is the use of a cochlear implant or a hearing aid. A major factor for auditory prosthesis candidacy is a functional auditory nerve. Since auditory neuron impairment can vary dramatically in CHARGE, it would be ideal to determine functional activity of neurons from individuals. However, direct recording from the auditory nerve is invasive and difficult because the inner ear is small, encased in bone and difficult to access. Instead, we have established a progenitor cell line that continually proliferates and can differentiate into auditory neurons. I propose to use progenitor-derived auditory neurons as a platform for identifying a panel of genes that correlates to auditory neuron function. In the future, when samples from patients with CHARGE become available, induced pluripotent stem cells (iPSCs) can be made. Auditory neurons generated from iPSCs from patients with CHARGE can be used to determine their candidacy for auditory prosthesis.



Category: Medical/Genetics
(Grant Research Updates)

Friday
Breakout Session #1:3: 10:45-11:45
Palomino 4 & 5

Presentation Title
Chd7 in neural crest-
mediated cardiac
development

Adam B. Stein, M.D.
Assistant Professor of Medicine
Dept of Medicine, Division of
Cardiology
University of Michigan

Presenter Information:

Adam B. Stein, M.D.
Assistant Professor of Medicine
Dept of Medicine, Division of Cardiology
University of Michigan

Presentation Abstract:

CHD7-mutation positive CHARGE patients display a range of clinical abnormalities including temporal bone defects, hearing defects, heart defects, craniofacial defects and choanal atresia. Although CHARGE Syndrome results in a seemingly diverse spectrum of congenital abnormalities, one unifying explanation is that the phenotypic traits result from abnormal neural crest cell (NCC)-mediated differentiation and/or migration. Cardiac NCCs (CNCCs) are a NCC population that migrate to the heart and great vessels where they are important for the development of the aorta and pulmonary artery from the pharyngeal arch arteries (PAAs) as well as the septation of the conotruncus into the ventricular outflow tract. We hypothesized that the absence of Chd7 in murine cardiac neural crest cells could recapitulate the congenital abnormalities seen in patients with CHARGE Syndrome. Using a Wnt1-Cre mouse model and a floxed Chd7 allele, Chd7 was conditionally deleted from neural crest cells (Wnt1-Cre) in vivo. We observed that a lack of Chd7 in Wnt1-Cre expressing neural crest cells does not compromise the ability of these cells to migrate and differentiate into normal cardiac structures. Thus, we conclude that Chd7 is not necessary for the development of neural crest-derived cardiac structures in our murine model.

People with CHARGE Syndrome often have congenital cardiovascular defects. Mutations in *CHD7*, the gene encoding chromodomain helicase DNA binding protein 7, have been identified in CHARGE Syndrome in a majority of cases. In an effort to understand more about this syndrome, scientists have created mice that have only one functioning copy of the *Chd7* gene. Mice with a loss of *Chd7* function display CHARGE-like phenotypes and are an excellent model of human CHARGE Syndrome. Several of the murine models with one functioning *Chd7* gene display congenital cardiac abnormalities. In patients with CHARGE Syndrome, observed congenital heart problems are likely a result of abnormal development of the conotruncal region (outflow tract- i.e. where the pumping chambers of the heart give rise to pulmonary artery and the aorta) and the great vessels (aorta and pulmonary artery).

During development, the conotruncal region and the great vessels are derived from several different populations of early progenitor cells. Neural crest cells (NCCs) are one developmental cell population that is necessary for the proper development of these cardiac structures. NCCs are an interesting cell

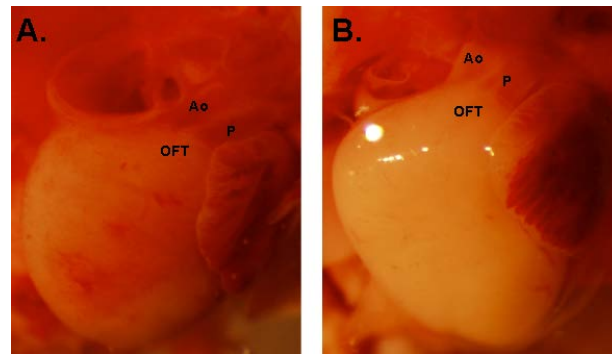


Figure 1. Grossly intact septation and structure of the outflow tract (OFT), aorta (Ao), and pulmonary trunk (P) in mice with *Chd7* still present in NCCs (Panel A) and with *Chd7* deleted in NCCs (Panel B).

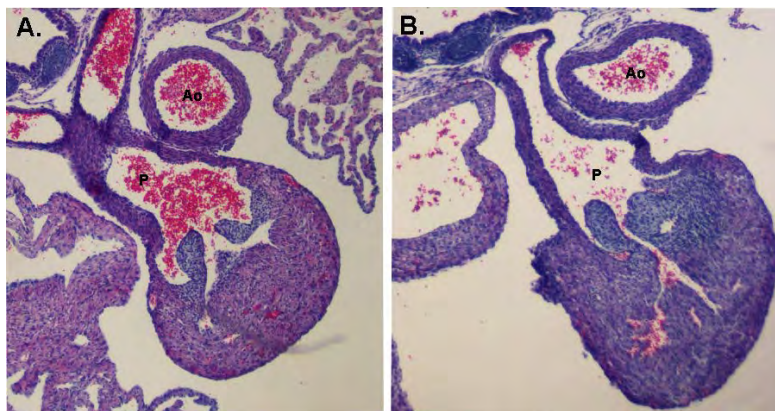


Figure 2. H&E staining of embryonic sections from mice with *Chd7* present in neural crest cells (Panel A) and in mice with *Chd7* absent from NCCs revealed intact septation and overall structure of the pulmonary trunk (P) and the aorta (Ao).

type that originates near the neural tube. NCCs migrate to many different parts of the developing embryo where they differentiate into a diverse array of tissues. Thus, it is plausible that many of the clinical features of CHARGE Syndrome may be a result of an inability of NCCs to migrate and differentiate into various tissues. Interestingly, it has been shown that one

group of NCCs, namely cardiac NCCs, migrate from the neural tube to the heart where they are important for the normal development of the heart and the great vessels. The objective of our study was to determine whether *Chd7* is important for NCCs to migrate to the heart region and successfully participate in the development of the outflow tract and the great vessels.

We created a unique mouse model in which we could breed mice to selectively delete *Chd7* (*Chd7^{fllox}*) from NCCs (*Wnt1-Cre*). We found that we were not able to generate viable pups that have *Chd7* deleted from the NCC population (*Wnt1-Cre:Chd7^{fllox/fllox}*). The pups died shortly after birth, and our observations revealed that the pups likely died due to abnormal brain development and oral palate defects that prevented them from feeding properly. In order to determine if mice without *Chd7* in the NCC population have abnormalities in the development of the conotruncal region and the great vessels, we studied at embryonic mice just before birth (e16 and later). As shown in figure 1, at embryonic day 18, we observed normal development of the outflow tract and normal septation of the aorta and pulmonary trunk. In order to further visualize the structure of the outflow tract, pulmonary trunk and aorta of these structures, we fixed and cut tissues from mice with and without *Chd7* in the NCCs. As shown in figure 2, staining revealed that the aorta and the pulmonary trunk were septated.

Thus far, our results suggest that *Chd7* deletion in NCCs using a *Wnt1-Cre* driver is not critical for the development of the outflow tracts and the septation of the pulmonary trunk and the aorta. We are currently looking at earlier time points to see if the development of the pharyngeal arch arteries is impacted by the deletion of *Chd7* in NCCs. We are also using other murine models to delete *Chd7* from a variety of early cell populations that participate in the development of the cardiac structures that are often impacted in patients with CHARGE Syndrome.



Category: Medical/Genetics

Friday
Breakout Session #1:4: 10:45-11:45
Palomino 6 & 7

A LOT TO SWALLOW
Understanding why individuals with
CHARGE Syndrome
are at high risk for choking:
Awareness, Recognition, and
Response

Dr. Kim Blake, MD, MSc, FRCPC
 Associate Professor of Medicine
 (Pediatrics)
 Dalhousie University, Halifax, NS, Canada

Kate Beals, OTR/L
 South Carolina Interagency Deaf-Blind
 Project
 South Carolina School for the Deaf and the
 Blind
 Spartanburg, SC, USA

Presenter Information:

Dr. Kim Blake is a Professor of Pediatrics at the IWK Health Centre in Halifax, Nova Scotia, Canada. She began her involvement with CHARGE 30 years ago at Great Ormond Street hospital in the UK. She published some of the earliest papers of CHARGE Syndrome and helped organize the UK family support group. At Dalhousie University in Halifax, she has continued her research on CHARGE, with focus on anesthesia, feeding, sleep and issues of the adolescent and adult with CHARGE. Kim has recruited local faculty and mentored many medical students in doing research on CHARGE Syndrome.

Kate Beals is an Occupational Therapist with 16 years of experience working with children. For the past five years she has worked with the South Carolina Interagency Deaf-Blind Project, providing training and support for the families and educational teams of children who have combined vision and hearing challenges, often with multiple and complex disabilities. Kate's areas of special interest include sensory integration, deaf-blindness, early communication development, and of course CHARGE Syndrome. Kate credits her 23-year-old son, who has autism, with being her best teacher.

Presentation Abstract:

Children who have CHARGE Syndrome often have difficulty chewing and swallowing, which causes them to be at high risk for choking. Parents and professionals who have a better understanding of how the process of chewing and swallowing operates will be better prepared to recognize and respond to this potentially life-threatening aspect of CHARGE Syndrome.

11th International CHARGE Syndrome Conference
Scottsdale, AZ, USA
July 25-28, 2013

A LOT TO SWALLOW

Understanding why individuals with CHARGE Syndrome are at high risk for choking:

Awareness, Recognition, and Response



Dr. Kim Blake, MD, MSc, FRCPC
Professor of Medicine (Pediatrics)
Dalhousie University, Halifax, NS, Canada

Kate Beals, OTR/L
South Carolina Interagency Deaf-Blind Project
South Carolina School for the Deaf and the Blind
Spartanburg, SC, USA

Session Objectives

- Learn about how chewing and swallowing work.
- Learn about where, how, and why chewing and swallowing can be problematic for children who have CHARGE Syndrome.
- Learn which professionals should participate in a multidisciplinary swallowing assessment for a child with CHARGE Syndrome.
- Become more aware of behavioral signs and signals that your child may be experiencing difficulty with swallowing.
- Begin making an emergency plan to use in case of a choking incident.

Don't be scared - just be ready.

Session Road Map

Awareness

- How do chewing and swallowing normally work?
- What nerves and muscles are involved?
- How do they interact?
- What goes amiss for children who have CHARGE Syndrome?
- What is the best way to assess chewing and swallowing functions for a child who has CHARGE Syndrome?

Recognition

- Can you tell if/when your child is experiencing oral feeding problems?
- How do you know?
- What signs and signals can you look for?

Response

- What do you do in the event of a choking emergency?
- Make a plan before you need it!

First Stop

"Eating and swallowing are complex behaviors involving volitional and reflexive activities of more than 30 nerves and muscles."

Awareness

Beware:
This presentation may make you hungry!

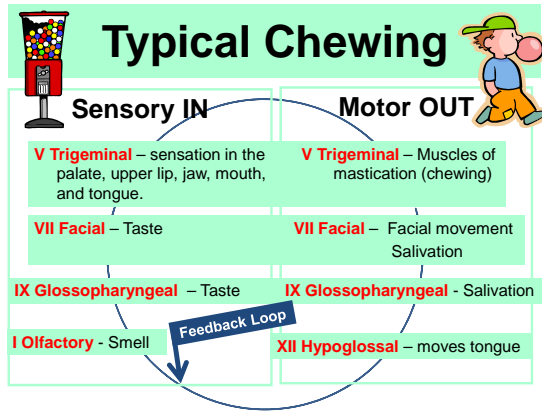
Koichiro Matsuo, DDS, PhD^a and Jeffrey B. Palmer, MD^{b,c,d}

Cranial Nerves

These guys direct the traffic & run the show.

Name	What It Does
I Olfactory	Smell
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

Typical Chewing



What about taste...

...and smell?



How do they work together?

An Experiment



NO PEEKING!

Typical 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

Awareness Activity

This guy makes it look easy...



...but eating is a lot more complicated than you might think.

When everything works

Eating is a fun, rewarding, and pleasant experience.



When everything doesn't work



Sometimes the **Cranial Nerves** don't work properly in individuals with **CHARGE Syndrome**.

And eating is not so much fun...

Name	If it doesn't work, this is what happens:
I Olfactory	Anosmia – inability to smell
V Trigeminal	Absent sensory response of face, absent corneal reflex, weakness of muscles of mastication decreased or absent sensation in the sinuses, the palate, upper lip, jaw, mouth, and tongue.
VII Facial	Facial paresis, compromised function of taste, tearing and salivation
IX Glossopharyngeal	Impaired gag reflex, uncoordinated swallowing...
X Vagus	Loss of gag reflex, uncoordinated swallowing; Abdominal pain from viscera; dysphonia
XI Spinal Accessory	Shoulder droop, winging of scapula, weakness of head turning (posture affects swallowing)
XII Hypoglossal	Deviation of tongue to affected side

Seriously, we'd rather have our G-tubes until this chewing and swallowing thing gets sorted out.



Yeah, we're cool.

Sorting it out: Research

DLHOUSE University

The Cranial Nerve Anomalies in CHARGE Association/Syndrome

Paediatrics and Child Health. Vol 8 (Sup B: 26B No. 38 May/June 2003

CMD Lawand¹, C Prasad², JM Graham, Jr³, KD Blake⁴

¹Medical Student Dalhousie University, ²Genetics & Metabolism, Department of Pediatrics, University of Manitoba, ³Director of Clinical Genetics & Dysmorphology, Cedars Sinai Medical Centre, UCLA, ⁴Department of Pediatrics, FWK Health Centre

Cranial Nerve Anomalies in CHARGE Syndrome

Population based cohort n=65
33 individuals with abnormal brain scans,
30 had abnormal Cranial Nerves:

- 92% demonstrated Cranial Nerve anomalies.
- 77 % more than one Cranial Nerve involved.

...and some of them affect chewing and swallowing!

Lawand CMD, Blake KD, Prasad C, Graham JM Jr. *The Cranial Nerve Anomalies of CHARGE Association/Syndrome (A/S)*. Paediatrics and Child Health. Vol. 8 (Supp B): 26B No. 38, May/June 2003.

Frequency of suspected CN Anomalies (study population)

*CN II involvement was not explicitly requested on the physician reporting form, but was indicated for some patients. Hence, this is a minimum frequency, and is likely higher in reality.

Apparent CN Involvement	Number Affected	Total Number Reported	Frequency (%)
At least one	60	65	92%
More than one	50	65	77%
CN II*	15	65	23%
CN V	34	46	74%
CN VII	28	58	48%
CN VIII (Vestibular)	19	31	58%
CN VIII (Cochlear)	48	55	87%
CN IX / X	46	56	82%

Likelihood of Specific CN Dysfunction, given dysfunction in any other CN

Then the Likelihood of:					
If:	CN V	CN VII	CN VIII (coch)	CN VIII (vest)	CN IX / X
CN V		20/34 = 59 %	27/34 = 79 %	14/34 = 41 %	29/34 = 85 %
CN VII	20/28 = 71 %		24/28 = 86 %	10/28 = 36 %	22/28 = 79 %
CN VIII (coch)	27/34 = 79 %	24/28 = 86 %		17/48 = 35 %	34/48 = 71 %
CN VIII (vest)	14/34 = 41 %	10/28 = 36 %	17/48 = 35 %		14/18 = 78 %
CN IX/X	28/46 = 63 %	22/46 = 48 %	34/46 = 74 %	14/46 = 30 %	

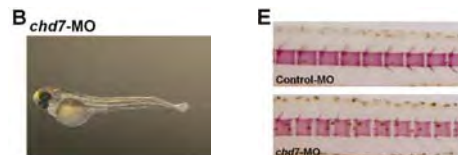
Summary of Cranial Nerve (CN) Findings in CHARGE Syndrome

- Apparent dysfunction of **Cranial Nerves** is more frequent and multiple.
- The extent and involvement of **Cranial Nerves** may reflect the clinical spectrum.
- **CN VII Facial Nerve**- is more frequently associated with other CN's
 - is seen in those individuals more severely affected.
- **CN V Trigeminal Nerve**- "muscles of mastication" affected in CHARGE.
- Structural brain malformations highly associated with CN.

Research Hot off the Presses!

Role of Chd7 in Zebrafish: A Model for CHARGE Syndrome. PLoS One. 2012;7(2):

Patten SA, Jacobs-McDaniels NL, Zaouter C, Drapeau P, Albertson RC, Moldovan F.
Sainte-Justine Hospital Research Center
Montreal, Quebec, Canada.



Recommendations

- Referral to a pediatric gastroenterology specialist in the neonatal period, as soon as the diagnosis of CHARGE is made
- Follow-up should involve a multidisciplinary feeding team
- Parents should be made aware of the potential for long-term feeding issues.

Dobbelsteyn C, Marche DM, Blake KD, Rashid M. 2005. *Dysphagia*. 2005, Vol. 20: 89-100.

Assessment

Who should be on the multidisciplinary feeding team?

- Speech language pathologist
- Occupational therapist
- Psychologist
- Dietician
- Sometimes a physiotherapist

Find a team or put one together.

Second
Stop

What was THAT?
What WAS that?
WHAT was that?

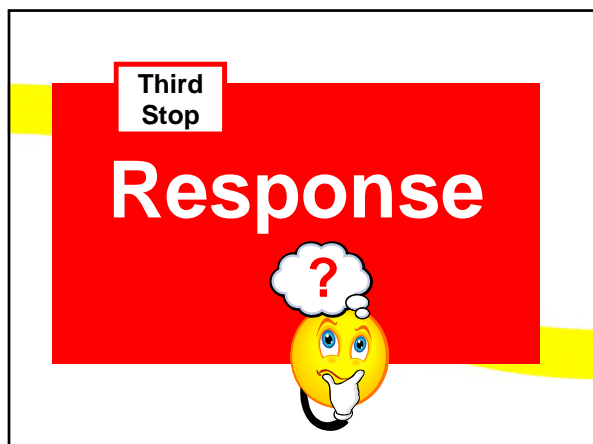
Recognition



Be on the lookout for...


- Cough, spluttering, choking
- Can't speak, but can sign
- **Red** in the face, **blue** in the face
- Labored breathing
- Abdominal breathing
- Clutches at throat or mouth

These are signs of distress
that indicate a blocked airway.



If my child is choking

- How will I know?
- What will I do first?
- Will I call anyone? Who?
- If I am not home, and someone else is caring for my child, does that person know what signs to look for, what to do, who to call?
- Is emergency information posted in a prominent place and easy to read?



Choking Intervention

~ What I need to know ~ What I need to do ~

ABC

A. Airway

B. Breathing

C. Circulation


OR

Cough it out.

Slap it out.

Squeeze it out.


1. Assess the situation
2. Encouraging the victim to cough
3. Back slaps
4. Abdominal thrusts
5. Modified chest thrusts
6. Finger sweeping



Family Action Plan

1. Make it.
2. Post it.
3. Use it.

Don't be scared – just be ready.



References and Resources

Anatomy and Physiology of Feeding and Swallowing – Normal and Abnormal
Koichiro Matsuo, DDS, PhD^a and Jeffrey B. Palmer, MD^{b,c,d}
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2597750/>

Dobbelsteyn C, Marche DM, Blake KD, Rashid M. 2005. Dysphagia. 2005, Vol. 20: 89-100.

Dobbelsteyn, Peacocke, Blake, et. al. Feeding difficulties in children with CHARGE syndrome: prevalence, risk factors and prognosis. Dysphagia
<http://www.livestrong.com/article/143185-cranial-nerve-mouth/#ixzz2RVQOEhLW>

Lawand CMD, Blake KD, Prasad C, Graham JM Jr. The Cranial Nerve Anomalies of CHARGE association/syndrome (A/S). Paediatrics and Child Health. Vol. 8 (Supp B): 26B No. 38, May/June 2003.

Patten SA, Jacobs-McDaniels NL, Zaouter C, Drapeau P, Albertson RC, Moldovan F. Role of Chd7 in Zebrafish: A Model for CHARGE Syndrome. Sainte-Justine Hospital Research Center Montreal, Quebec, Canada.

Rosenfeld-Johnson, S. Oral Placement Therapy for Speech Clarity and Feeding Skill Development. TalkTools, Charleston, SC

CHARGE Syndrome: A Management Manual for Parents
©2002 - Edited by Meg Hefner, M.S. & Sandra Davenport, M.D.
(Note the sections on Cranial Nerves and Feeding and Swallowing)

Questions? Ideas? Thoughts to share?

Session Evaluation





Thank you for coming!

11th International CHARGE Syndrome Conference
Scottsdale, AZ, USA
July 25-28, 2013





Category: Behavior

Friday
Breakout Session #1:5: 10:45-11:45
Palomino 8 & 9

**Behavior as self-regulatory adaptation,
or
“I can’t believe my child just did that!”**

Tim Hartshorne, Ph.D.
Central Michigan University

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:

Individuals with CHARGE often show odd, sometimes challenging, behaviors. These can lead to various psychiatric diagnoses. However, behavior is rarely random, and in fact humans actively attempt to adapt to their experience. The concept of self-regulation is a way to view “CHARGE behavior” as adaptation, and leads to avenues for intervention. This is the first of three presentations on self-regulation and intervention for behavioral challenges.

Behavior as self-regulatory
adaptation, or “I can’t believe my
child just did that!”

Tim Hartshorne
Central Michigan University

Typical 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 vocalization

How to make sense of it

- The kid has a syndrome!
- It’s pathological and should be eliminated
- It’s due to frustration and pain
- It’s communication
- It works for the kid

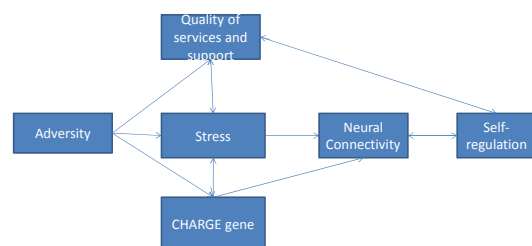
Not because they guarantee
success, but because they
serve a purpose

Self-regulation problems in CHARGE

- Rapid changes in arousal levels
- Melt downs
- Unfocused behavior
- Diagnoses
 - OCD – a way to reduce stimulation and exercise control
 - ADHD – a problem with regulating sensory and behavioral stimulation and focusing on a goal
 - Tic disorder – a stress response to lack of control over environment
 - Autistic-like behavior – the failure of regulation strategies, and the adoption of dysregulated behavior

Definition

The primarily voluntary
regulation of cognition, behavior,
emotion, and physiological
states for the purpose of goal-
directed actions



After Blair & Raver, 2012

Adversity

- Fragile health
 - Breathing problems
 - Multiple hospitalizations
 - Multiple surgeries with anesthesia
 - Multi-sensory impairment
 - Defects in major organs
- Nervous parents
- Sources of stress
 - Social relationships
 - School
 - Family
 - Abuse

Quality of Services and Support

- Lack of medical or specialist knowledge
- Needs multi-disciplinary medical and educational teams
- Parent-Professional relationships
- Lack of social support
- Parent and family resilience

CHD7 Gene

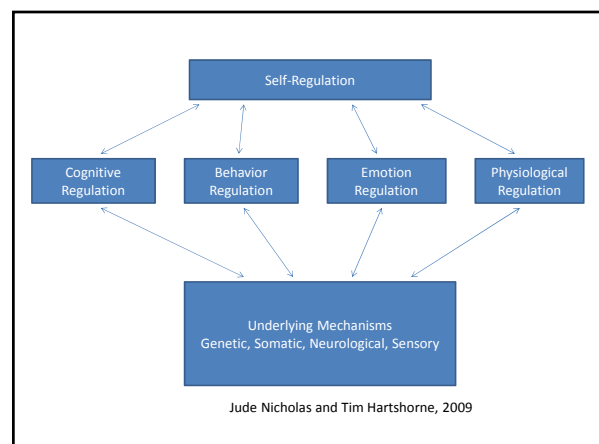
- Regulatory gene
 - Neural crest
 - Placode cells
- Multisensory impairment
- Major organs may be affected
- Vestibular functioning impaired

Stress

- Endocrine regulatory system
- Perception of adversity
- Availability of resources
- Response of professionals
- Response of family

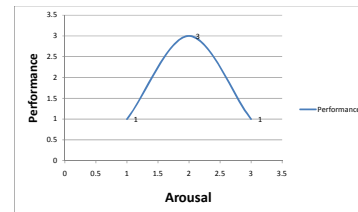
Neural Connectivity

- Prefrontal cortex and executive function
 - Reactive forms of learning and behavior
 - Reflective forms of learning and behavior
- Neuropsychological control over behavioural schemas
 - Routine control
 - Supervisory attentional system



Dunn Conceptual Model

Arousal of thoughts, behavior, feelings, sensations	PASSIVE Self-regulation Strategies	ACTIVE Self-regulation Strategies
Habituation	Non-reactive Tune it out	Sensation Seeking
Sensitization	Reactive to Stimuli	Sensation Avoiding



Self-regulation is used to manage arousal levels

With a regulatory disorder, child is challenged to manage
 Cognitive – unfocused vs. obsessive
 Behavior – hyperactive vs. hypoactive
 Emotion – reactive vs. passive
 Physiological – overwhelmed vs. underwhelmed

Self-regulation begins with a goal

- What do you want to have happen?
- What must you do to make it happen?

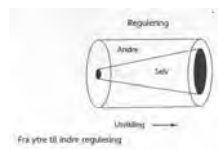
Study for an exam

- Cognitive
- Behavioral
- Emotion
- Physiological

Strategies?

Supporting self-regulation

- Because self-regulation skills are hard for children with significant disabilities to develop
- We have to provide the external support for what will become an internal self-regulatory process



Scaffolding

- The process of planning and organizing the activity of children so that they can execute a task that is beyond their current level of ability.

Components of Scaffolding

1. Identification of the problem to be solved
2. Focus activities on outcomes and goals
3. Frustration control
4. Reducing the complexity of the task
5. Marking critical relevant features
6. Modeling

The Shape Sorter



1. Problem Identification
2. Focus on outcomes
3. Frustration control
4. Reducing complexity
5. Marking features
6. Modeling

Examples

- Cognitive self-regulation
 - Break down larger goals into shorter (pie)
- Behavioral self-regulation
 - Feedback on reactions from others (consequences)
- Emotional self-regulation
 - Creating an environment for self-soothing
- Physiological self-regulation
 - Squeeze technique; hand on arm or leg

Summary

- Children with disabilities often have poorly regulated systems
- This is centrally related to stress, deriving from adversity, quality of supports, and genetics
- The child's attempts to self-regulate manifest as peculiar behavior, often labeled as challenging
- They will do better socially and academically if they can learn to self-regulate
- They can only develop self-regulation skills slowly while they experience a lot of scaffolding from the adults in their lives

Thanks to my Lab

- Maria Ramirez
- Andrea Larson
- Sarah Haney
- Kayla Hilyard
- Ben Kennert



www.chsbs.cmich.edu/timothy_hartshorne

Contact information

- Dr. Tim Hartshorne
 Department of Psychology
 Central Michigan University
 Mount Pleasant, MI 48859
 989-774-6479
tim.hartshorne@cmich.edu
www.chsbs.cmich.edu/timothy_hartshorne



Category: Medical

Friday
Breakout Session #1:6: 10:45-11:45
Palomino 10

**Communication Challenges in
 Charge Syndrome**

Daniel Choo, MD
 Cincinnati Children's Hospital Medical
 Center

Presenter Information:

Daniel Choo, MD, FAAP, FACS

Professor and Director, Division of Pediatric Otolaryngology Head and Neck Surgery

Cincinnati Children's Hospital Medical Center

University of Cincinnati College of Medicine

3333 Burnet Avenue, Cincinnati, Ohio. USA. 45229

(513) 803-4194 (phone); (513) 636-8133 (fax)

Presentation Abstract:

Children affected by Charge Syndrome (CS) pose a significant challenge to healthcare providers in terms of determining an optimal timeframe and sequence for managing communicational issues. Children are frequently affected by anomalies of the external, middle and inner ear that can cause a hearing loss and concomitant delays in auditory and verbal skill development. Compounding these delays are associated problems of the oral cavity or oropharynx (e.g. cleft lips and palates) or the upper airway that require multiple treatments or sometimes tracheostomy tube placement that can impair voice, speech and swallowing development as well. Adding additional layers of challenges to the management plans can be a multitude of other congenital medical issues (e.g. heart anomalies) that pose major health risks and need to take priority over communicational issues. These circumstances can readily create a scenario where aspects such as hearing, speech and language can get pushed onto the back burner for so long that communicational development is beyond the typical and optimal timeframes. With increasing experience in managing children with CS, greater predictability in terms of treatment outcomes and most importantly, better interdisciplinary communication amongst managing professionals, it is possible to create a comprehensive approach and strategy that comes close to an ideal management plan for complex children with CS.



Category: Medical and
Family Support

Friday
Breakout Session(s) #1-3: #7, #15 & #20
10:45-11:45; 1:00-2:00; 2:15-3:15
Dorado

**CHARGE 101: New family information
and orientation**

Meg Hefner, MS, Genetic Counselor
Sandra Davenport, MD, Medical
Genetics
Kim Blake, MD, Developmental
Pediatrics
Nancy Hartshorne, PhD, School
Psychology

Presenter Information: Together, this panel represents more than 100 years of experience and expertise with CHARGE syndrome (CS). Meg and Sandra started working together on a DeafBlind study in 1982 in Columbia, Missouri and were on the founding Board of the CHARGE Syndrome Foundation. Kim was on the original UK team and helped found the UK support group. She is now in Eastern Canada. Nancy has expertise as a mother of an adult son with CS, is a school psychologist and professor specializing in development in CHARGE, and lives in Michigan. In the last hour or so of the 101 sessions, we will be joined by “experienced” parents to add to the expertise in the room.

Presentation Abstract: As the Conference has grown, it has become more difficult for families coming to their first conference to get oriented – where to go, who to talk to, how to meet each other. CHARGE 101 is intended to make the process a little easier. CHARGE 101 is a series of presentations intended for families with new babies and/or new to Conference. It is a combination of an overview of CHARGE and an orientation to the rest of conference. Meg will cover diagnostic criteria/medical features of CS. Sandra will talk about how sensory deficits (especially hearing, vision and balance issues) affect early development. Kim will present information on some key medical issues in infancy, especially regarding breathing, sleep, and anesthesia. Nancy will talk about communication options. Along the way, we encourage participants to ask questions of the presenters and interact with one another. We will help guide families to other talks throughout the weekend that cover areas they are most interested in and point them to other experts to answer questions. The final hour of the 101 series will include “experienced parents” who can also answer questions and concerns. Please feel free to come and go as your needs and interests dictate.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013



Category: Medical/Genetics,
Family Support

Friday
Breakout Session #1:7: 10:45-11:45
Dorado

CHARGE 101: New families
What makes it CHARGE?
How do sensory deficits change
development

Meg Hefner, MS, Genetic Counselor
Sandra Davenport, MD, SensoriGenetics
Saint Louis University

Presenter Information:

Together, the CHARGE 101, 102 & 103 panel represents more than 100 years of experience and expertise with CHARGE syndrome (CS). Meg and Sandra started working together on a DeafBlind study in 1982 in Columbia, Missouri and were on the founding Board of the CHARGE Syndrome Foundation. Kim was on the original UK team and helped found the UK support group. Nancy has expertise as a mother of an adult son with CS and is a school psychologist specializing in CHARGE. In the 102 & 103 the panel will be joined by “experienced” parents to help answer all of your questions.

Presentation Abstract:

As the Conference has grown, it has become more difficult for families coming to their first conference to get oriented – where to go, whom to talk to, how to meet each other. CHARGE 101-103 is intended to make the process a little easier.

In the CHARGE 101 hour, Meg will cover diagnostic criteria and medical features of CHARGE – what are the clinical diagnostic criteria and what are the other features commonly seeing in babies and children with CHARGE. Then Sandra will talk more specifically about the sensory deficits in CHARGE (especially hearing, vision and balance issues) and how those differences affect early development, with emphasis on the communication bubble. This will set the stage for the 102 hour where Nancy will talk about communication options for children with CHARGE.

We encourage participants to ask questions of the presenters and interact with one another. We will also help guide families to other talks throughout the weekend that cover areas they are most interested in and point them to other experts to answer questions. Please feel free to come and go as your needs and interests dictate.

CHARGE SYNDROME DIAGNOSIS AND FEATURES

Meg Hefner, M.S.
Genetic Counselor
Clinical Associate Professor of Pediatrics
Saint Louis University School of Medicine

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

****Should NOT be used for diagnosis**

Epidemiology of CHARGE Syndrome

- 1 in 8,500-10,000 births
- Most often a new dominant mutation
 - Usually paternal in origin
 - Increased but low recurrence risk
 - Major gene identified in 2004 (CHD7)
 - There may be other genes/causes
- Mild end of spectrum is still emerging

Recurrence

- Empiric risk of
~1% for parents
with one child
with CHARGE
- 50% for children of
individuals with CHARGE
- <<1% for other family
members



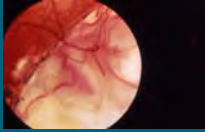
Current CHARGE Diagnostic Criteria

- Positive gene test (CHD7+)
- OR
- Clinical diagnosis
 - Major features
 - Minor features

CHARGE Syndrome Major Features (4C's)

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

Coloboma – cleft of the eye



Retinal Coloboma
Increased risk of retinal detachment with retinal coloboma



Iris Coloboma

Retinal coloboma causes upper visual field defects



Macular coloboma: central vision loss

- The child will look *above* the point of interest and not appear to be making eye contact



CHARGE Syndrome Major Features (4 C's)

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

Choanal atresia/stenosis

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



Baby with stents

Choanal stenosis

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



CHARGE Syndrome Major Features (4 C's)

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

Cranial nerves 9 & 10: Swallowing problems



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

Swallowing complication: Aspiration (Kim Blake will cover in hour 3)

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

Cranial Nerve # 7 Facial palsy



Unilateral – lopsided face



Bilateral – no facial expression

Cranial Nerve #1 Sense of smell

- Decreased or absent sense of smell
- Difficult to evaluate, but EXTREMELY common (>90%)
- Changes "taste"
- Implications for feeding
- Social implications
 - smelly feet
 - body odor
 - passing gas

CHARGE Syndrome Major Diagnostic Features (4 C's)

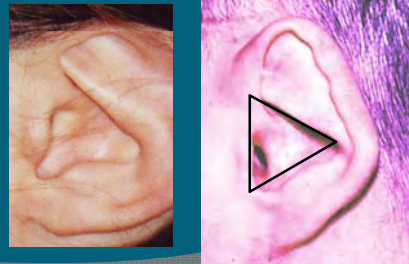
- Coloboma of the globe
- Choanal atresia/stenosis
- Cranial nerve anomalies
- **Characteristic CHARGE ears**

Characteristic CHARGE Ear: OUTER EAR

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

Characteristic CHARGE Ear

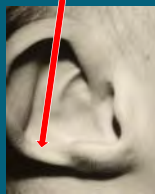
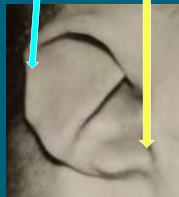
- Short, wide, **triangular concha**, absent lobe



Characteristic CHARGE Ear

Snipped off helix with small or absent earlobe

Discontinuity between the antihelix and antitragus

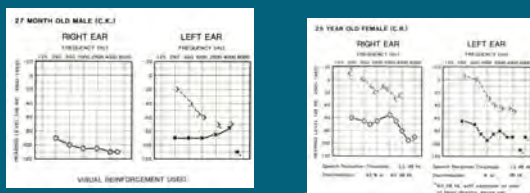


Characteristic CHARGE Ear: MIDDLE AND INNER EAR

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

Hearing in CHARGE

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



Inner Ear Balance Problems

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

CHARGE Syndrome Minor Diagnostic Features

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

CHARGE Syndrome Minor Features Unique to CHARGE

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

Truncal hypotonia

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



Characteristic CHARGE Face

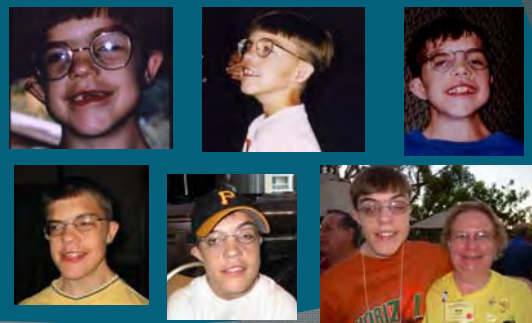
- Square face
- Broad forehead
- Round eyes
- Nose
 - Prominent bridge
 - Square root
 - Prominent columella
- Small chin, gets bigger with age

Characteristic CHARGE Face

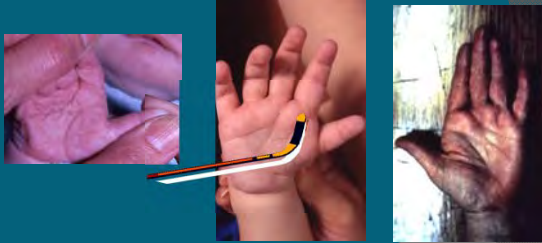


To see more faces: "About CHARGE" link at
www.chargesyndrome.org

CHARGE: 7 to 19 years



Minor feature: Hockey-stick palmar crease



CHARGE Syndrome
Minor Feature: Heart defects

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



Heart surgery at four years old

CHARGE Syndrome
Other Minor Feature: Clefts

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



Repaired cleft lip

CHARGE syndrome minor feature: Esophagus/Trachea

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

CHARGE syndrome minor feature: Renal (kidney) anomalies

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

40% have renal anomalies

CHARGE Syndrome
Minor Feature: short stature

Due to:

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



Treated with growth hormone

CHARGE Syndrome

Minor Feature: genital hypoplasia

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



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

Variability

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

Beyond diagnosis:

Other features (any organ system!)

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

More Other features

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

CHARGE *management* issues beyond the diagnostic features

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

Behavior in CHARGE syndrome

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

Intelligence in CHARGE

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

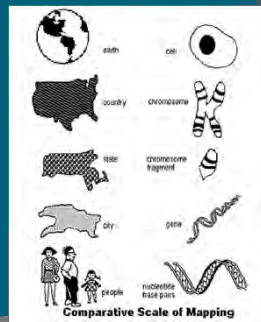
How well do they do?

The typical 2 year old with CHARGE:

- has spent 6 months in the hospital
 - has had at least 6 surgeries
 - is followed by 10 medical specialists
 - is fed by G-tube
 - is not walking or talking
 - has some unusual behaviors
- Looks pretty “retarded,” but probably isn’t

A major gene for CHARGE

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



Major CHARGE gene: CHD7 on chromosome 8q12

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

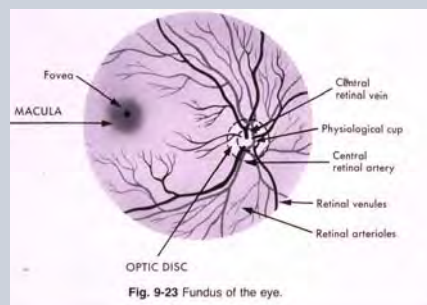
Now what - How is this gene helpful?

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

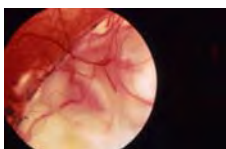
Sensory Loss: How does that Change Early Development?

Sandra L.H. Davenport, M.D.
Sensory Genetics/Neuro-development

952-831-5522
slhdaven@umn.edu



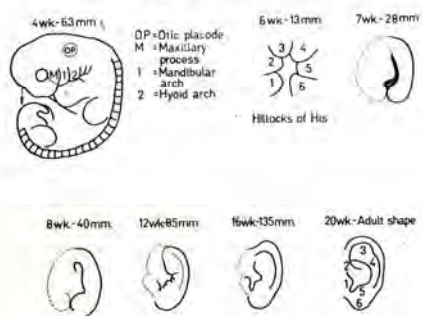
Coloboma – cleft of the eye



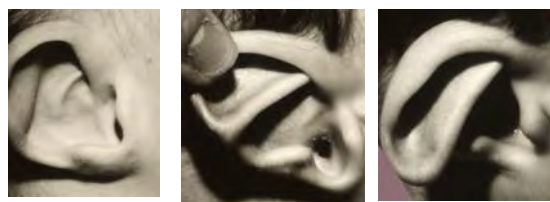
- Retinal Coloboma
– Visual field defects
- Retinal coloboma causes upper visual field defects
- Iris Coloboma
– Light sensitivity

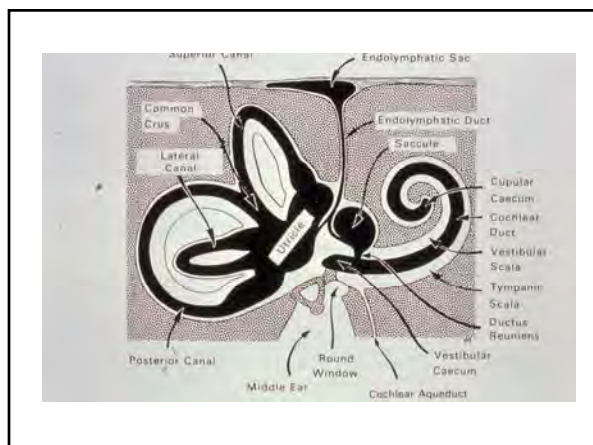
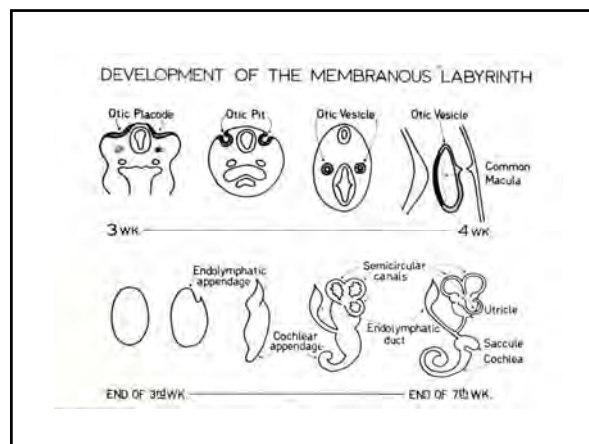


DEVELOPMENT OF THE AURICLE



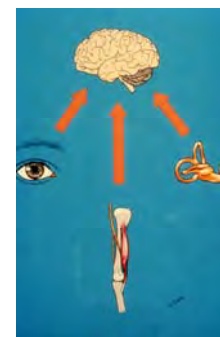
Characteristic CHARGE Ear





Components of Balance

- Muscles & joints – proprioception
- Vision
- Vestibular – inner ear balance
- Cerebellum
- Cerebrum – CPU (central processing unit)



Communication Bubble

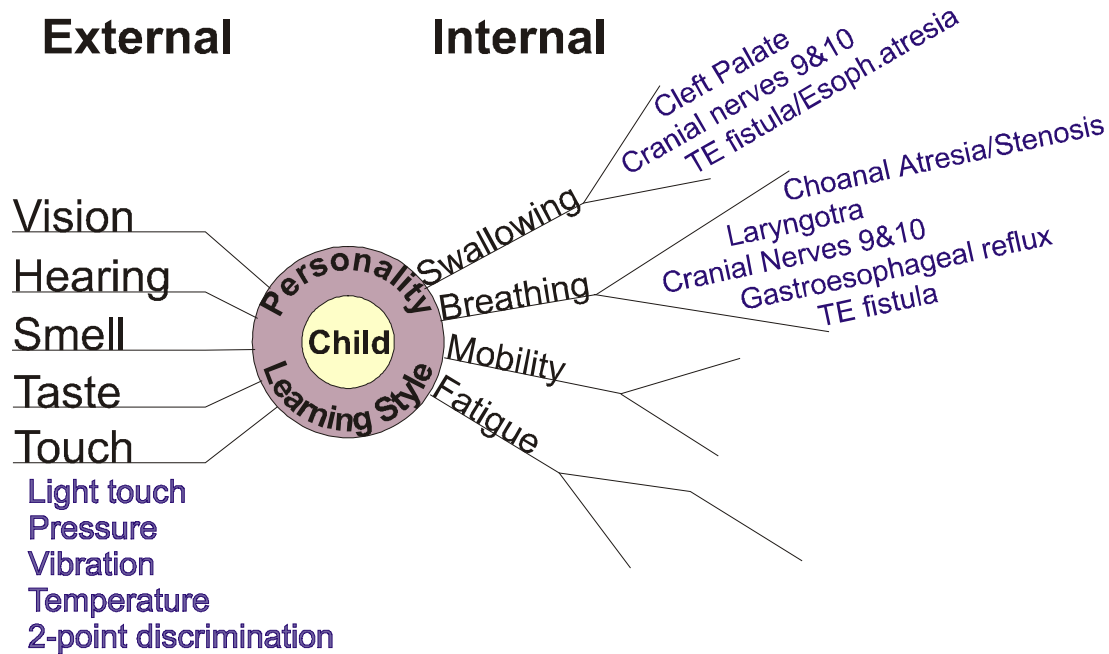
- The area within which the individual can
 - See
 - Hear
 - Smell
- Touch and taste do not give information at a distance
- Anything outside the bubble is “off the radar”

Kids with CHARGE may be “Hard of”

- | | |
|---------------|-------------------|
| • hearing | • swallowing |
| • seeing | • breathing |
| • smelling | • balancing |
| • feeling | • multitasking |
| • empathizing | • space awareness |

CHARGE 102: PHYSICAL INFLUENCES ON DEVELOPMENT IN CHARGE

Sandra L.H. Davenport, M.D.
SensoryGenetic/Neuro-development,
952-831-5522 slhdaven@umn.edu



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

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

A more complete discussion of this is in the Manual

INFLUENCE OF SENSORY LOSS ON DEVELOPMENT: The Communication Bubble

Are all five major senses working?

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

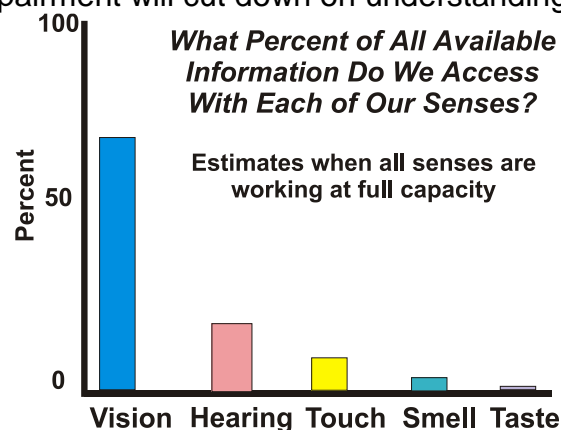
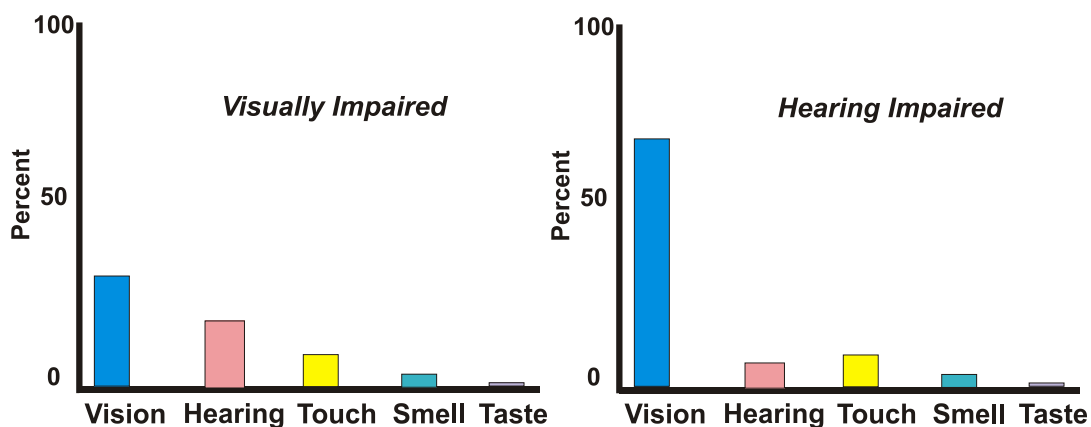


Figure 1: Full access to information from all senses.

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



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

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

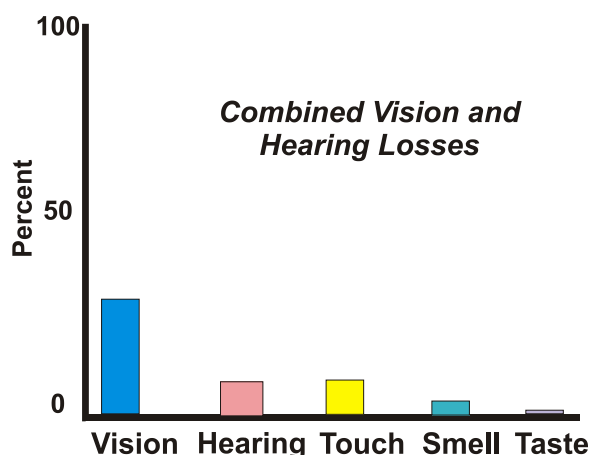


Figure 4: Combined vision/hearing loss with half of each gone.

What is Deaf-Blind?

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

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

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

What combination of vision & hearing is present?

When one or more senses are impaired, additional educational consultants are needed, even (or perhaps especially) during the Early Childhood phase of development. Research out of Denver shows that, if a communication system is established BEFORE six months of age, a deaf child can develop completely normal language. Even if they learn to read sign language, they do not fall behind their hearing peers in expressive language and reading. The implications are staggering. Even though medical issues in CHARGE occupy the thoughts and minds of parents and caretakers during the first 2 years of life, it would appear to be very important to start some form of communication as early as possible.

The Need to Establish A Communication Bubble

Establishing what Susan Smith, the parent of two children with CHARGE, calls the "Communication Bubble" is essential if you want to make sure the child knows you are there and are trying to communicate. The concept of a "bubble" is a good one because you can imagine the space within which you need to be. As Eric Kloos says, you need to be "on the child's radar." Get outside the bubble and you might as well not exist. Too many times, we think the child is tuned out, not paying attention, or is too "retarded" to answer when, in fact, he/she may not even know that you are trying to say something.

Each parent should know how far a child can see what kinds and colors of objects, with or without movement, in different lighting conditions and against different shaded backgrounds. The eye doctor will usually not be able to tell you this except by guessing though some do this kind of testing. A vision teacher, however, can do a Functional Vision Evaluation, to help establish these visual abilities. Figuring out what the child hears clearly, at what distance and on which side is also important. Neither of these is easy to determine so parents and team members will likely have to engage in a continuing evaluation process.

The other very important point is that a lot of communication precedes formal language. Every mother can "read" her child's communication regardless of whether that child uses any formal language. Sometimes it is guessing, but the child will let the mother know when she's got it right. Therefore, it is important to lead up to formal communication step by step. We use sight cues and verbal cues like outstretched arms with some encouraging words to let a hearing/sighted child know what we want to do next. For a child without good vision or hearing, concrete object cues and touch cues are used instead. DeafBlind project staff can help the regular and special education staff learn how to use these methods. As parents and educators we may get hung up on wanting our children to speak our own language using the method we use. That is as natural as immigrants wanting their children to continue speaking their language and remembering their culture. However, the issue is not the form but the substance. The child needs to learn that an object, touch, picture, word or sign is a symbol that stands for a thought. Many of them strung together convey complex ideas or stories. They include naming of objects, actions, remembering the past, anticipating the future and, later, getting into abstract thoughts and discussions.

So, above all, just get to it! COMMUNICATE, COMMUNICATE, COMMUNICATE.



Category: Family Support

Friday
Breakout Session #1:8: 10:45-11:45
Moor 2

Grandparents Gathering

Robert Last

Presenter Information:

Rob is trained as an Early Childhood Teacher with Vision Australia involving children with sensory disabilities in particular in hearing and vision. He has developed communications systems to meet individual needs of children with CHARGE syndrome since the early 1990s and has been an invited presenter at every International CHARGE Syndrome Conference. . He is primarily engaged in home based early intervention with families. He has done longitudinal studies of the progress made by people who have CHARGE syndrome, beginning in 1993. This is through video interviews in 1993, 2000 and 2007. Rob has been very active in the Australasian CHARGE association since its inception and help with the even-year Australasian CHARGE conferences.

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.



Category: Medical/Genetics,
and Behavioral

Friday
Breakout Session #2: 9: 1:00-2:00
Palomino 1 & 2

**“How to Identify Pain Non-Vocally and
the Relationship of Pain to
Challenging Behavior”**

Kasee Stratton, Ph.D.
**Kennedy Krieger Institute/Johns
Hopkins University School of Medicine**

Presenter Information:

Dr. Kasee Stratton has been researching and working with children and young adults who have CHARGE for the past 7 years. She is a previous student of Dr. Timothy Hartshorne. Dr. Stratton's primary research and clinical interests include: reducing challenging behaviors, teaching appropriate adaptive skills, identifying non-vocal pain behaviors, and reducing the pain experience for individuals with CHARGE. Kasee is currently finishing her Post-Doctoral Fellowship at the Johns Hopkins University School of Medicine and the Kennedy Krieger Institute. She plans to continue her work with CHARGE following completion of her fellowship and hopes to open a CHARGE clinic in the near future.

Presentation Abstract:

This presentation is designed to highlight the importance of pain for individuals with CHARGE and how to identify pain non-vocally. The presentation will emphasize results from two pain studies that are the first of their kind for individuals with CHARGE. Results are presented in a manner appropriate for parents, caregivers, educators, and physicians. Specific highlights include: the variety of pain experiences (both acute and chronic), how to identify and track pain for your child, how pain impacts behavior, and strategies for reducing the pain experience.

HOW TO IDENTIFY PAIN AND THE RELATIONSHIP OF PAIN TO CHALLENGING BEHAVIOR

11th International CHARGE Syndrome Conference
Kasee Stratton, Ph.D.

Thank you!

- CHARGE Syndrome Foundation
 - Research funding support
 - Supporting our participant lists
- Families of children with CHARGE
- Central Michigan University
 - Funding support

CHARGE and Pain Overview

- ◆ Pain in developmental disabilities
- ◆ CHARGE syndrome and pain
- ◆ Are we identifying pain?
- ◆ How pain is related to challenging behavior
- ◆ Areas of future research

A Parent's Story...

"Since my son was born almost 29 years ago, every aspect of his care has been a challenge. Being a single parent has been hard, but never harder than when my son is in pain and I can't help him. As a parent, it is my job to make sure his needs are met and that he is loved. I feel like I have let him down when he is having pain and I can't make it better. Unless it is something obvious, I have to play the guessing game of what hurts and why.

...continued

...My son is non-vocal and cannot tell me what is wrong. One of the biggest barriers to our children is others (e.g. doctors) understanding children with disabilities can have chronic pain too. And they don't understand that pain contributes to behavior issues, such as SIB, that can be life-threatening. My son has had two subdural hematomas from SIB. It took me 6 months to get a CT scan of his head. In that 6 months he was in such excruciating pain. There is a fight everyday to get him what he needs."

Pain and Developmental Disabilities

- "Higher" threshold for pain
 - Has been suggested in CHARGE (Davenport, 2002)
 - Limitations with Communication: Changes expression of pain
- No evidence
- Higher risk for experiencing more frequent pain
- High Pain Threshold vs. High Pain Tolerance

Sources of pain related to CHARGE

- Surgery
 - 1 to 63 procedures
 - Average 13
- Procedures
- Doctor visits
- CHARGE Characteristics



Does your child experience pain from?

Pain Experience	(N= 61)	Percentage of Participants
Ear Infections	41	67.2
Sinus Infections	27	44.3
Gastroesophageal Reflux	26	42.6
Constipation	26	42.6
Surgery	23	37.7
Tactile Defensiveness	21	34.4
Migraine	15	24.6
Stoma Pain	12	19.7
Abdominal Migraine	12	19.7
Muscle Pain	12	19.7
Back Pain	8	13.1
Hip Pain	6	9.8
Jaw Pain	5	8.2
Pain During Sleep	5	8.6

Migraines

- Trigeminal nerve (CN V)
 - Sensation and function to your jaws, face, tongue, sinus, palate, eyes, teeth, and lips.
 - Also has a role with chewing and swallowing
 - CN dysfunction in CHARGE

Blake, K.D., Hartshorne, T. S., Lawand, C., Dallor, A. N., & Thelin, J. W. (2008).
Cranial nerve manifestations in CHARGE syndrome. *American Journal of Medical Genetics*, 146A, 585-592.

Migraines

- Most intense pain for children with CHARGE
- Range of 2-40 days/year
 - $M = 14$
- Average Rating: 2.67 (Range: 2-4)



Abdominal Migraine

- Typically children ages 5 to 9
- Linked to adult migraines
- Lasts 1 to 72 hours
- Acute stomach pain with
 - Nausea
 - Vomiting
 - Light sensitivity
 - Diarrhea
 - Loss of appetite

Abdominal Migraine

- Range from 2-365 days/year ($M = 97$)
- Average Rating: 2.45 (Range 1-4)



Constipation

- Painful bowel movements
- Dry or hard stool
- Nausea
- Cramps, abdominal pain
- Average pain for 52 days/year (1-203)
- Hurts more rating (2.38) (1-4)
- Fecal impaction
 - Abdominal cramping
 - Rectum discomfort

Otitis Media

- Range from 1 to 160 days a year; $M=26$ days
- Average rating 2.24 (Range 0-4)



Gastroesophageal reflux disease

- Heartburn
 - Involves a burning pain in the chest (under the breastbone)
 - Increased by bending, stooping, lying down, or eating
 - More frequent or worse at night
 - Relieved by antacids
- Nausea and vomiting
- Regurgitation of food
- Sore throat
- 10-365 days/year ($M = 169$)
 - Average rating 2 (hurts more)

Tactile Defensiveness

- textured materials/items
- "messy" things
- vibrating toys
- a hug or kiss
- certain clothing textures
- rough or bumpy bed sheets
- seams on socks
- tags on shirts
- tags on shirts
- light touch
- hands or face being dirty
- shoes and/or sandals
- wind blowing on bare skin
- bare feet touching grass or sand

Most Intense Pain and Average Duration

Characteristic	Pain Intensity			Days per Year in Pain	
	<i>M</i>	<i>SD</i>	Range	<i>M</i>	<i>SD</i>
Migraine	2.67	.87	2-4	13.50	13.51
Abdominal Migraine	2.45	1.10	1-4	97.47	128.95
Constipation	2.38	.80	1-4	52.25	58.38
Surgery Pain	2.34	.97	1-4	9.52	9.40
Chronic Recurrent Otitis Media	2.24	.99	0-4	22.88	32.18
Sinusitis	2.17	.82	1-4	35.13	41.51
Gastroesophageal Reflux	2.06	1.14	0-4	169.29	133.70
Breathing	2.00	1.03	1-4	108.67	131.82
Hip/Back Pain	1.86	.95	1-4	98.09	144.14
Muscle Pain	1.82	.87	1-3	95.70	136.07
Coughing	1.61	.80	1-3	66.48	99.42
Jaw Discomfort	1.56	.88	1-3	13.22	11.17
Difficulty Swallowing	1.50	.83	1-4	129.00	154.04

Identifying Pain in CHARGE

- Are you able to determine when your child is experiencing pain?
 - 75% -Yes
 - Did not vary significantly by age of child
- What about educators, therapists, & doctors?
- Zero parents could identify chronic pain and no child could indicate chronic pain

What behaviors indicate pain?

- Vocal
 - Crying, screaming, moaning
- Social
 - Irritable, withdrawn, doesn't follow directions
- Facial
 - Grinds teeth, changes in eyes (glassy), furrowed brow
- Activity
 - Lethargic, inconsolable, decreased movement/activity
- Body and Limb Movement
 - Rubbing area of pain, holding body in unusual posture
- Physiological
 - Fever, splotchy appearance, bowel movements, congestion
- Eating/Sleeping
 - Tired, changes in sleep

What behaviors indicate pain?

- Behavioral Challenges
- Self-Injurious Behavior (SIB)
- Dangerous Behaviors
- Aggressive, bites, hits head, throws objects, punches, pulls out g-tube

Amount of Pain



Why is it difficult to measure pain in CHARGE?

- Limited or no communication strategies
 - Cannot use the gold-standard
- Possible social-communicative deficits
 - (Craig, 2006)
- Possible social referencing deficit
 - (Recchia, 1997)

Measuring Pain

- Facial Reactions to Pain
 - Limited research
 - Facial palsy in CHARGE
- Rating Pain
 - Numerical ratings with pictures
 - Multidimensional pain tools

SCALES

- Non-Communicating Children's Pain Checklist-Revised (NCCPC-R)
 - 30 items; 7 subscales
 - vocal, social, facial, activity, body and limbs, physiological, and eating/sleeping

II. Social

Not cooperating, cranky, irritable,
 unhappy..... 0 1 2 3 NA
 Less interaction with others,
 withdrawn..... 0 1 2 3 NA

PPP

- Pediatric Pain Profile (PPP)
 - 20 items
 - Rate: 0-3
 - Not at all, a little, quite a lot, and a great deal
- Examples:
 - Grinds teeth or makes mouthing movements
 - Is restless/agitated or distressed
 - Tenses/stiffens or spasms

Measuring Pain

- Baseline:
 - Complete NCCPC-R and PPP on a good day
- NCCPC-R and PPP day of pain
- A significant difference was found

Common Pain Behaviors

NCCPC-R

- not moving, less active, quiet
- tears
- not cooperating, cranky, irritable, unhappy
- crying
- moaning, whining, whimpering
- less interaction w/others, withdrawn
- not smiling
- being difficult to distract, unable to satisfy
- furrowed brow

PPP

- not cheerful
- crying, moaning
- not socially responsive
- grimaces, screws up eyes and face
- frowns/furrowed brow
- hard to console/comfort

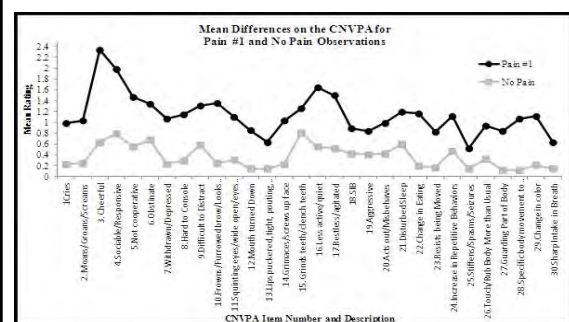
Not entirely useful for CHARGE

- Did not display a meaningful difference:
 - Flexing inward/drawing legs up (PPP #15)
 - Stereotypical movements/jumping/seizures (PPP #20)
 - Flopping (NCCPC-R #16)
 - Shivering (NCCPC-R #22)
 - Jumping around/agitation/fidgety (NCCPC-R #15)

CNVPA

- Items from parental input and previous study
- Significant difference between no pain and pain ratings; strong reliability
- For 36% of our sample, physicians were able to confirm a diagnosis that is known to produce pain (e.g. sinus infection)

CNVPA

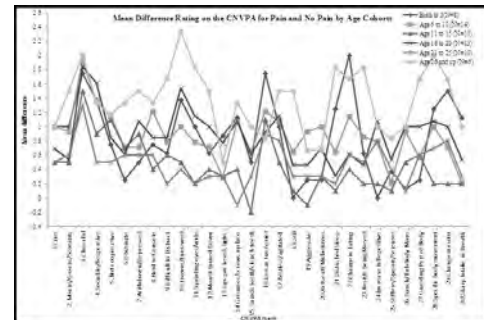


CNVPA

At least 1 point difference: **.9 difference**

- Not Cheerful
- Aggressive
- Not Sociable
- Frowns/furrowed brow/looks worried
- Less active/quiet
- Restless/Agitated
- Change in Eating
- Specific movement to indicate pain
- Not cooperative
- Change in color

Differ by age?



Differ by Age?

- 1 month to 5 years
 - Fewer challenging behaviors
 - Change in eating
 - Less active/quiet
 - Change in color
- 11 to 15 years
 - Lower mean difference
 - aggressive behaviors
 - Grinding teeth/clenching teeth
- Age 26 and +
 - Squinting eyes/eyes wide open/eyes frowning
 - Mouth turned down
 - SIB
 - Disturbed sleep
 - Resist being moved
 - Specific body part held

CNVPA: Is it helpful?

Do parents find the CNVPA to be relevant to identify their child's pain (non-vocally)?

- Approximately 85% endorsed the CNVPA to be a relevant assessment to identify pain

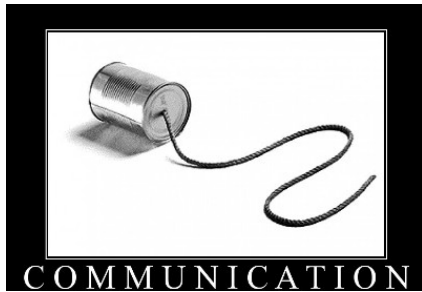
CNVPA helpful?

- Why might this instrument not be relevant?
 - Child can verbalize pain vocally (12)
 - "Never complains of pain and seems to tolerate it well."
 - "I've already developed ways to identify pain for my child" (3)
 - "After 24 years, I am in tune to my child's health"

Functions of Behaviors

- Attention
- To gain access to preferred items/activities
- To escape/avoid demands or less preferred items/activities
- Stimulatory

All Behavior is...



Challenging Behavior in CHARGE

- Common challenging behaviors
 - Preference for certain items
 - Restricted range of interests
 - Difficulty with social relationships
 - Repetitive behaviors; increase under stress
 - High levels of sensation seeking; may include self injurious behavior
 - Executive Dysfunction
 - Regulatory Disorder

Does pain affect behavior?

- Evidence that pain is associated with behavior problems in typical-developing children
 - De Lissovoy (1962) head banging and otitis media
 - Hart, Box, & Jenkins (1984) tantrums and upper respiratory infection
- Evidence that pain is associated with behavior problems in children with disabilities
 - O'Reilly (1997) self-injury and otitis media
 - Carr & Owen-DeSchryver (2007) sick days
 - Lekkas & Lentino (1978) constipation
 - Kennedy & Meyer (1996) allergies

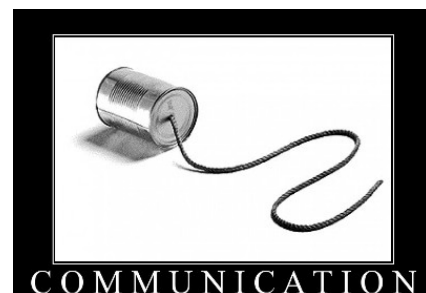
Does pain affect behavior?

- Aggressive behavior, destructive behavior, and self-injury (Kennedy and O'Reilly, 2006)
- Elevated pain → elevated self-injury (Symons and Danov, 2005)
 - We found similar results
- Attachment and Adaptive Functioning
 - Withdrawing and decreased communication
- Quality of life may be compromised (Oberlander & Symons, 2006)

Understanding Pain

- Unknown what children with CHARGE know about pain
 - How to predict when and how it will be resolved
 - Increase the intensity of the experience and also increase challenging behaviors
 - Individuals with CHARGE may need to be explicitly taught coping strategies to help identify pain and how to control these events in their lives

Challenging behavior as...



Reducing the pain experience

- Use CNVPA to track progress over time
- Mitigation
 - Analgesics
 - Dietary change
- Redesigning the environment
 - Reducing the demands
- Teaching coping skills
 - Self advocacy
 - Functional communication alternatives
- Parental Interaction with Physicians

Conclusion

- Children with CHARGE experience considerable amounts of pain and often exhibit problem behavior
- Problem behavior may have many causes, but one of them can be pain
- Pain can be managed when we know the child is experiencing pain, but not all children with CHARGE can easily communicate this
 - CNVPA may be a useful alternative

Future Pain Research

- Relationship between behavior and duration of pain
 - Impact adaptive, academic, and overall functioning
- Relationship between challenging behavior, pain, and communication (adaptive behaviors)
- Analgesics, neurological development, and the treatment of pain
- Controlled validity studies (e.g. surgery)
- Further investigation of age and sex differences

Contact information

Kasee Stratton, Ph.D.

Post-Doctoral Fellow
Kennedy Krieger Institute
Johns Hopkins School of
Medicine
Baltimore, MD
stratton@kennedykrieger.org
strat1kk@cmich.edu

Soon to be joining Mississippi State University



Category: Behavior,
Medical/Genetics, Education

Friday
Breakout Session #2:10: 1:00-2:00
Palomino 3

**Balancing the Scales: Impact of
Internal and External Factors on
Health, Behavior, and Learning**

Kimberly Lauger, RN
Certified HANDLE® Practitioner
Redtail Neurological Center

Kristina L. Manning, M.A.
Teacher for the Deaf and Hard of Hearing
Teacher for Students with Visual Impairments
Arizona State Schools for the Deaf and the
Blind, SER-Coop

Megan Mogan, MS, CCC-SLP
Communication Specialist
Arizona State Schools for the Deaf and the
Blind, Tucson Campus

Presenter Information:

Kimberly Lauger, RN, is a parent and professional in the field of deafblindness and neurodevelopment. Kim has one son who struggled with attention and learning difficulties, one son with CHARGE Syndrome and vision and hearing loss, and a daughter for whom learning and “being” is relatively easy. Driven to understand the reasons beneath their struggles and desperate for ideas on what to do, Kimberly began her studies into neurodevelopmental difficulties, eventually becoming a HANDLE® Practitioner. Kimberly works as a consultant and intervener trainer for the Arizona Deafblind Project and is owner of the Redtail Neurological Center.

Kristina Manning is a former Air Force officer with a lifetime fascination centering on multiculturalism and multilingualism who pursued advanced training in bilingual methodologies for deaf children and in deafblindness. She loves the puzzle of unlocking communicative access for pre-linguistic children and has family members on the Autism Spectrum. Making literacy functional and meaningful--joyful--for all children is the reason she teaches. She has never met a child she did not believe could learn to read via creative methods. She is the grandmother of three.

Megan Mogan is a Speech-Language Pathologist at the Arizona State Schools for the Deaf and the Blind-Tucson Campus. She studied Communication Disorders at Northern Illinois University in Dekalb, Illinois and received a Master's degree in Speech and Hearing Sciences from the University of Arizona. Megan worked in the Sunnyside Unified School District in Tucson before specializing in working with students who have multiple disabilities and sensory impairments, including deafblindness. Megan works directly with students on the development of early communication skills, and especially enjoys working with other educators on building optimal communicative environments for all types of learners.

Presentation Abstract:

Moving beyond labels—such as autism— into action, this session will briefly touch on how shifts in internal and external factors change the day-to-day function of individuals. Ideas will be illustrated using the case study of one boy with CHARGE syndrome. The remainder of the session will focus on educational, and communication strategies (external factors) that effectively tipped the scale for this boy leading to improvement in all areas of the Autism Treatment Evaluation Checklist; communication, sociability, sensory and cognitive awareness, behavior and health.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013

Balancing the Scales

Internal and External Factors that influence learning

Kimberly Lauger, BSN, RN, Certified HANDLE® Practitioner
Kristina Manning, MA, TVI, Teacher for the Deaf
Megan Mogan, MS, CCC-SLP, Speech Language Pathologist

Weight of multiple diagnoses

As a parent we are hit with one diagnosis after another. We are told what is "wrong" often leading to a sense of despair and hopelessness.

What we want to know is what is right and what we can do to help/support our child?

Weight of multiple diagnoses

As a teacher or provider we read the file. We are told what is "wrong" often leading to a sense of fear and a paralysis of not knowing where to start.

What we want to know is what is right and what we can do to help this student learn?

Even with the diagnoses,
we notice
our child/student learns
and what they do
changes moment by
moment.

Sometimes they "do" better than others.

What influences that change?

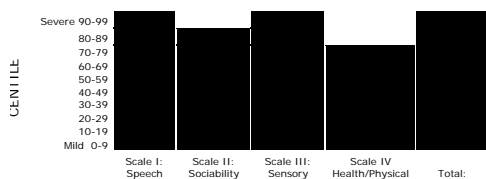
Internal factors: Everything going on inside the person.

External factors: Everything going on outside the person.

Understanding these factors tells us what to do, contributing to a sense of hope and possibility.

When Internal factors
and external supports
are balanced the
person's function is
optimal

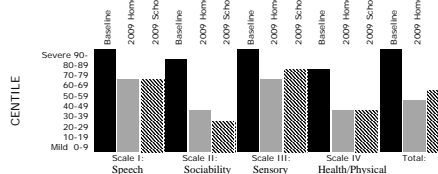
Figure 1: Autism Treatment Evaluation Checklist at Baseline May 1, 2002



Case study to illustrate change in function for one student

Year of greatest concern with total score in the 90-99th percentile for severity of symptoms of the autism spectrum

Figure 2: Autism Treatment Evaluation Baseline (May, 2002) and Follow-Up (April, 2009)



During Dylan's year of best function, total score dropped to the 45th to 55th percentile for severity of symptoms..

This corresponded with noticeable improvement in self-regulation and ability to interact and learn.

Internal Factors

Everything going on inside a person

- Physical feelings and sensations
- Emotional feelings and states
- How the body processes information
- How the body is made and how that influences access to information such as vision or hearing loss
- How the body functions such as ability to digest food well, integrate sensory information, and self-regulate
- Acute illness



As parents and educators/ providers we do not have direct control here.

External Factors

Everything going on around a person

- The physical environment such as lights, sounds, smells, movement and who is there
- How information is presented and use of educational strategies such as wait time, partial participation, rituals and routines, hand under hand
- Level of expectations, does the expectation match the student's internal ability to meet that demand or is it too much



External factors influence internal factors positively or negatively
This is where we have the ability to influence change



When the scale is out of balance the body is stressed

Increased repetitive, obsessive, and other challenging behaviors; increased illness; decreased attention and availability for exploration, interaction, and learning.



When the scale is balanced stress is managed and people function better

The next sections will focus on external factors (educational, communication, and sensory strategies) used with Dylan during the year of his optimal function.

Creating a Community

Establish common, accessible language and symbols

Create thematic, functional learning

Build routines, patterns, rituals

Control the schedule, environment

Recruit partners

Take ownership

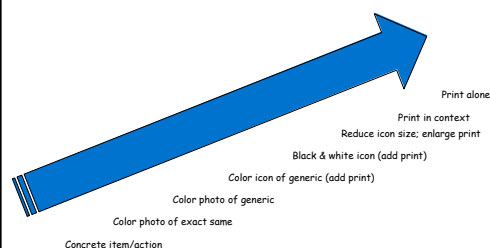
Establish Accessible Language, Symbols

Use Vygotsky to promote natural language acquisition through peer partnership

Teach symbols through use by peers in context

See Ladder of Abstraction

Ladder of Abstraction:



Adapted by K. Manning from Dr. Jane Erin (lecture), 2005

Create Thematic Functional Learning

- Literature (EVERYONE READS)
 - SMARTBoard technology
 - Differentiated Instruction
- Themes that apply to students' lives now
 - Ties to CBI, Science, Math
- Create class-made experience books
 - Reliving = retelling

Build Routines, Patterns, Rituals

- Predictable Sequencing
 - Use motor memory
- Control the Environment
 - Only the topic is new
 - Allow no external interruptions
- Build Anticipation for Application
 - CBI trips
 - Reveal newest experience book

Control Schedule, Environment

- Practice extreme environmental engineering
 - See Neurological Visual Triggers Slide
- Strictly limit pull-outs; collaborate on pull-ins
- Hang a "stop" sign outside the door; wear earplugs

How do we get the brain to look?

Neurological Visual Triggers include:

- Color contrast
- Light sources
- Movement
- Lack of visual "clutter"
- Lack of glare
- Favored topics/items



Controlling the environment for neurological visual triggers helps the brain look.

Adapted from Dr. Christine Roman-Lantzy, AFB Press, 2007
Kristine Manning, M.A.

Recruit Partners

Team Planning!

Intervener
(HI, VI teachers--if that's not you)
Speech-Language Pathologist
Orientation & Mobility Specialist
ASL, OT, PT
Admin (funding, CBI vehicles)
Parents
General Education Neighbors

Take Ownership

YOU must become THE teacher.
(You can do it.)

Fight for the right to follow student needs in developing
curriculae based on living participants.

Ask for what you need, but be willing to make decisions.

BELIEVE!!!

Non-Traditional Support Services for Non-Traditional Learners

Adapt to a push-in model (vs. pullout)

Engineer your caseload and schedule starting
at the beginning of the school year

Prioritize your push-in students based on
classroom teacher's limited options for
scheduling

Non-Traditional Support Services for Non-Traditional Learners

Accept overlapping areas of expertise in a team
model (we are all here for the same reason)

Model your specific knowledge and skill set
through instruction

Know when to rely on other team members'
unique backgrounds and skills

Non-Traditional Support Services for Non-Traditional Learners

Build *observation* into service delivery time and let this guide subsequent instruction

Observation of team members (collaborators)

Learn their routine and identify successful strategies

Learn their teaching and communication style

Observation of students

DO NOT intervene at first!

Learn how they access, understand, and express information

Pushing in(to) Thematic Learning

The concepts you are teaching do not change, only the topic does!

Play the same games

Utilize the same vocabulary templates

Practice the same conversational "scripts"

Model the same role-play scenarios

Work off the same sight word list

Use the same literacy formats and tools

The Future...

If you bond with your fellow educators/team members, your students WILL bond with their fellow peers.

If you take the time to intentionally observe your students, you WILL provide richer instruction as a result.

If you are committed to structure and routine, your students WILL regulate themselves for learning.

If you are having fun, your students WILL have fun!

Kimberly Lauger, klauger@centurylink.net www.handle.org

Kristina Manning, kristina.manning@asdb.az.gov

Megan Mogan, megan.mogan@asdb.az.gov



Category: Medical/Genetics
(Research Updates)

Friday
Breakout Session #2:11: 1:00-2:00
Palomino 4 & 5

Presentation Title
CHARGE and not so CHARGE-Genetic
conditions that overlap with CHARGE
syndrome

Seema R. Lalani, MD
Assistant Professor
Department of Molecular and Human Genetics
Baylor College of Medicine
Houston, TX 77030
Email: seemal@bcm.edu

Presenter Information:

Seema R. Lalani, MD

Assistant Professor

Department of Molecular and Human Genetics

Baylor College of Medicine

Houston, TX 77030

Email: seemal@bcm.edu

Presentation Abstract:

CHARGE syndrome is characterized by multiple congenital anomalies including characteristic external and inner ear abnormalities, hearing dysfunction, coloboma, choanal atresia, cranial nerve abnormalities, facial palsy, and congenital heart defects. The majority of patients with CHARGE syndrome have changes/mutations in the *CHD7* gene. However, the genetic basis in some children with atypical CHARGE syndrome is less clear. We completed whole exome sequencing in a subset of children who were negative for the *CHD7* mutation or deletion in our study. Our data show that some of the *CHD7* negative children suspected to have CHARGE syndrome have other known genetic disorders, with overlapping features of CHARGE. These syndromes will be discussed in the presentation. In addition, an overview of the genetic study results and clinical presentation of all patients will be given for the families.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013

CHARGE AND NOT SO CHARGE

Genetic condition that overlap with CHARGE syndrome

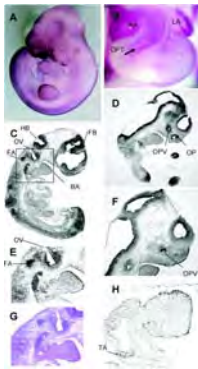
Seema R. Lalani, MD
Baylor College of Medicine
Houston, TX



Clinical characteristics of CHARGE syndrome



Expression of *chd7* in mouse



Expression of *Chd7* in heart, cranial nerves, brain, ears and nose

Lalani et al. AJHG 2006

Frequency of various anomalies in 134 individuals with CHARGE syndrome

Coloboma	83%
Choanal atresia	53%
Deafness	91%
Cleft lip/palate	32%
Facial palsy	54%
Heart defects	81%
Short stature	66%
Swallowing problems	75%
Urogenital problems	65%
Developmental delay	94%
Characteristic CHARGE syndrome ears	99%
Tracheoesophageal atresia	17%

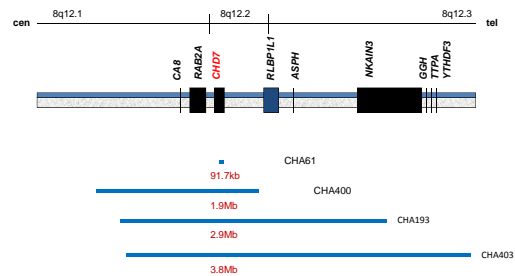
CHD7 mutations

- Most children with combined coloboma, choanal atresia and abnormal semicircular canals have mutations in the *CHD7* gene
- Majority of children with 4 major criteria of Blake (coloboma, choanal atresia, characteristic ears and cranial nerve dysfunction) have mutations in the *CHD7* gene
- There is significant intrafamilial variability associated with *CHD7* mutations

What does it mean when *CHD7* sequencing is normal in CHARGE diagnosis?

There can be **DELETION** of the **CHD7** gene rather than misspelling!

Large deletions of **CHD7**



In our study, **CHD7** deletion was observed in 4 out of 50 mutation negative individuals (8%)

Genetic conditions overlapping with **CHARGE- Branchiootorenal syndrome**



EYA1 deletion

- Absent left kidney and small right kidney
- Left iris and chorioretinal coloboma, small left eye
- Semicircular canal abnormalities on both sides
- Moderate hearing loss in the right ear and mild loss in the left
- Facial asymmetry
- Cup-shaped, simplified ears
- Right clubfoot

[Sanchez-Valle et al, 2010](#)

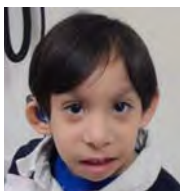
Mandibulofacial dysostosis with microcephaly



- **EFTUD2** mutations
- Choanal stenosis
- Hearing loss
- Ear malformations
- Inner ear abnormalities
- Esophageal atresia
- Cleft palate
- Small head size

[Lines et al, 2012](#)
[Luquetti et al, 2012](#)
[Gordon et al, 2012](#)

EFTUD2 mutation



- Failed the initial hearing screen in the newborn period
- Severe to profound hearing loss in the left ear and moderate loss in the right ear
- Small jaw
- Abnormal semicircular canals
- Small head size
- Short stature

CHD7 negative **CHARGE** syndrome

- Five families-EXOME sequencing



SIX1 mutation, related to Branchiootorenal syndrome

Child has choanal atresia, Mondini malformation, hearing loss, short stature, and swallowing problems

Summary

- Some children with a suspected diagnosis of CHARGE syndrome may have **Branchiootorenal syndrome** with changes in *EYA1* or *SIX1* genes
- ***EFTUD2*** mutations have also been identified in some suspected/atypical CHARGE syndrome
- **Chromosomal abnormalities** can be present in some children with atypical CHARGE diagnosis
- Some children may still have mutations in **parts of the *CHD7* gene that are not routinely tested in DNA analysis**

Acknowledgements

Baylor College of Medicine

- Xueqing Wang
- Susan Fernbach
- Mahshid Azamian
- Gladys Zapata
- John Belmont

Other collaborators

- Meg Hefner
- Sandra Davenport
- Donna Martin

CHARGE Syndrome Foundation
Doris Duke Charitable Foundation
CHARGE families





Category: Medical Genetics
(Research Updates)

Friday
Breakout Session #2:11: 1:00-2:00
Palomino 4 & 5

**Functions for CHD7,
the Gene Altered in CHARGE, in
Developing Cells and Tissues**

**Donna Martin, Joseph Micucci, and
Ethan Sperry**
The University of Michigan Medical School

Presenter Information:

Mutations in the *CHD7* gene are a common cause of CHARGE. Our laboratory has been studying mice with mutations in the *Chd7* gene. We will discuss recent data indicating roles for CHD7 in development of skeletal structures and in stem cells of the ear, nose, and brain.

Presentation Abstract:

CHD7, the gene for chromodomain DNA-binding protein 7, is a common genetic cause of CHARGE Syndrome. *CHD7* is highly expressed in developing human and mouse embryos, especially in stem cells and tissues that are affected in CHARGE. In order to identify the underlying mechanisms by which *CHD7* regulates organ growth and development, our laboratory has generated and analyzed mice with mutations in the mouse *Chd7* gene. Mice with reduced *Chd7* function have many of the same structural and functional deficits as those observed in CHARGE; thus, detailed understanding of *Chd7* function in mice can provide critical information for designing effective therapies. We will present recent data show *CHD7* functions in the development of stem cells of the ear, nose, and brain and in development of craniofacial structures. We will also discuss progress using induced pluripotent stem cells generated from skin cells of children with CHARGE and *CHD7* mutations. Together, these studies are helping to pave the way for novel, innovative strategies to develop regenerative therapies for individuals with CHARGE.

CHARGE Syndrome: Advances in Research

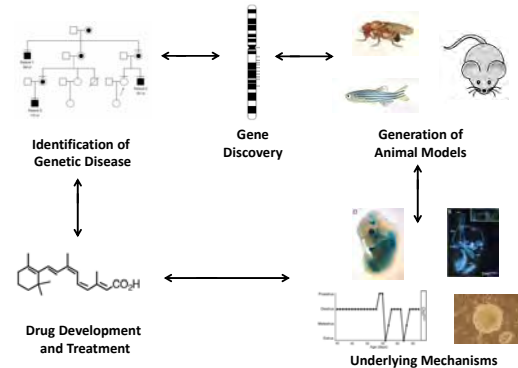


C.S. Mott Children's and von Voigtlander Women's Hospital, The University of Michigan

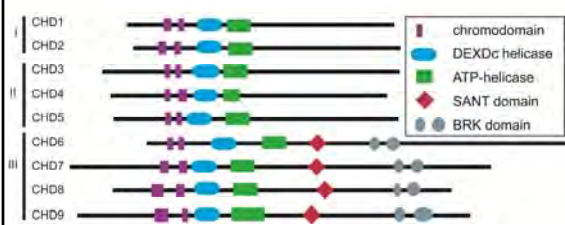
Donna M. Martin
MD, PhD
Joe Micucci, BS
Ethan Sperry, BS

The University of Michigan Departments of Pediatrics, Human Genetics, and the Medical Scientist Training Program

Exploring Genetic Mechanisms in CHARGE

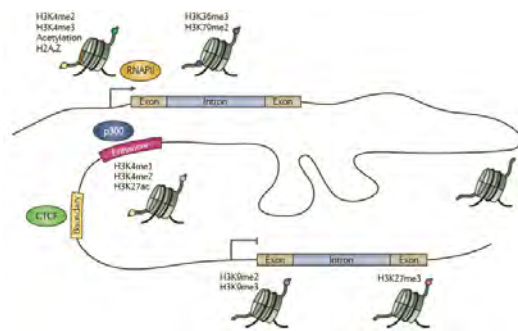


Chromodomain Helicase DNA-binding (CHD) Family



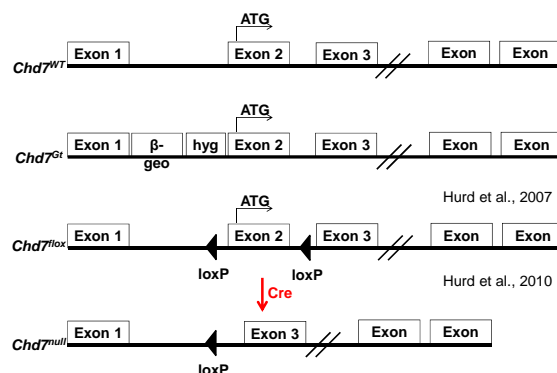
Layman et al. Clin Genetics 2010

Gene Structure and Chromatin Modifications



Zhou, VW, et al. Nature Reviews Genetics, 2011.

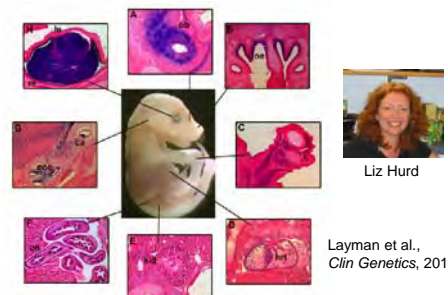
Chd7 alleles



Hurd et al., 2007

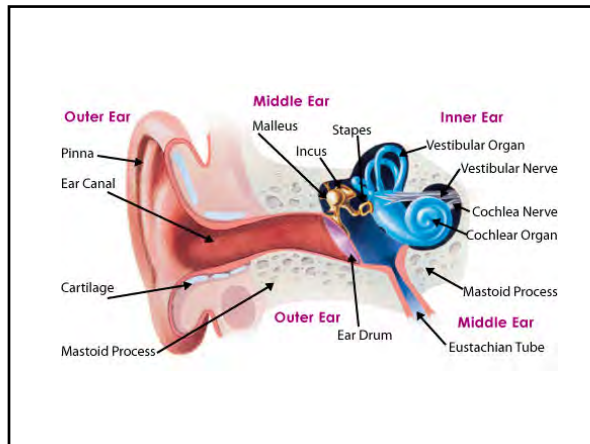
Hurd et al., 2010

Chd7 expression is tissue-restricted during development



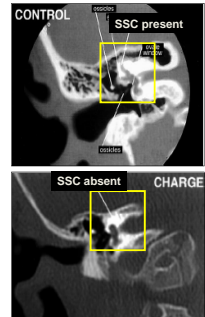
Liz Hurd

Layman et al., Clin Genetics, 2010



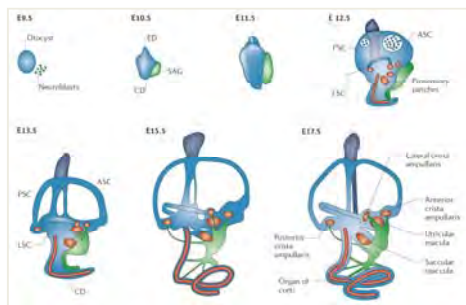
CHARGE is associated with semicircular canal hypoplasia

- Semicircular canal hypoplasia with vestibular dysfunction is common (temporal bone CT scan)
- Lateral SCC is always involved, while the superior or posterior may be normal
- Patients display abnormal canal vestibulo-ocular responses (>90%)



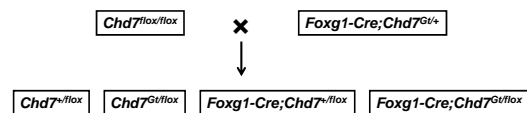
Morimoto et al., 2006 Am J Neuroradiol

Inner Ear Development



Kelley, 2006 Nat Rev. Neurosci.

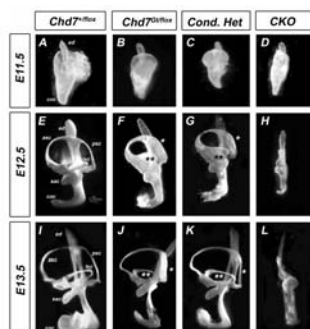
Conditional Mutant Mice



Hurd et al., 2010

Hebert and McConnell, 2000

Inner ear formation requires *Chd7*



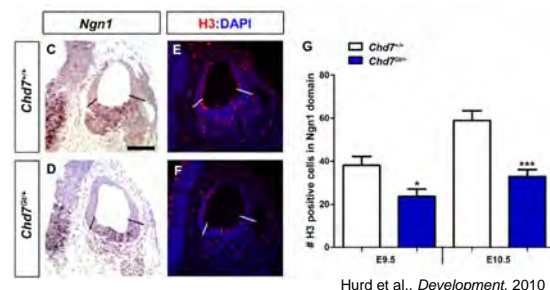
Liz Hurd



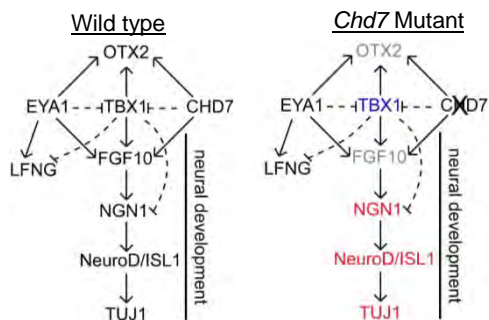
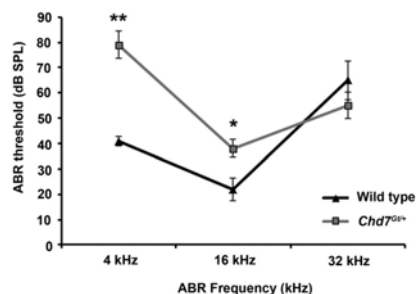
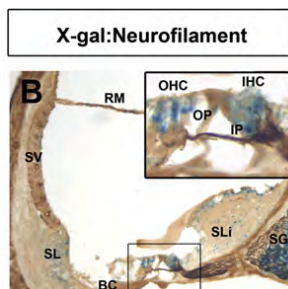
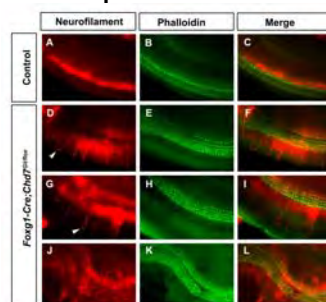
Elyse Reamer

Hurd et al., Mechanisms of Development 2012

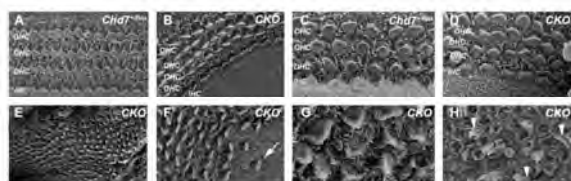
Chd7 mutants have reduced proliferation in the neurogenic domain



Hurd et al., Development, 2010

Model for *CHD7* Developmental Gene Regulation in Inner EarHurd et al., *Development*, 2010*Chd7*^{Gt/+} mice have mild hearing lossHurd et al., *Hearing Research* 2011*Chd7* is expressed in the mature cochleaHurd et al., *Hearing Research* 2011Cochlear innervation is abnormal with complete loss of *Chd7*

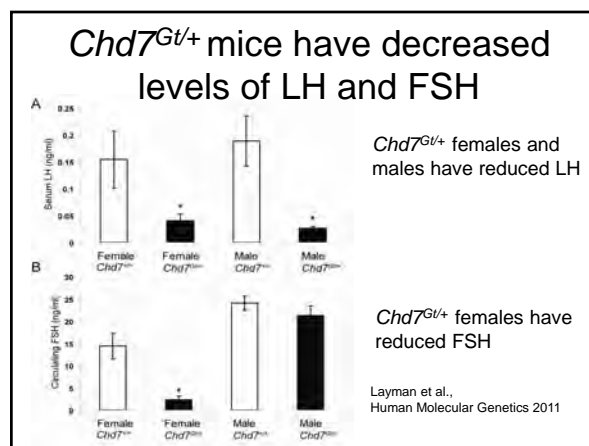
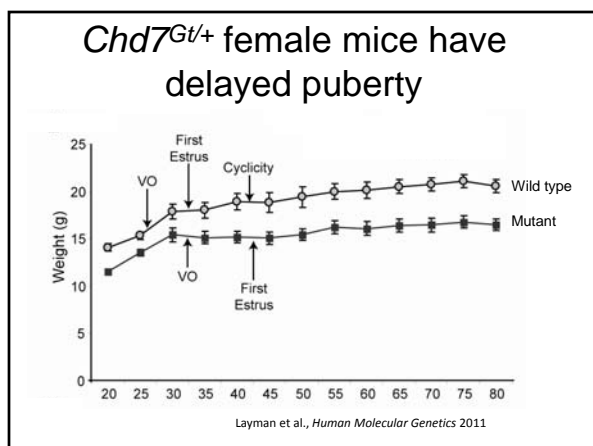
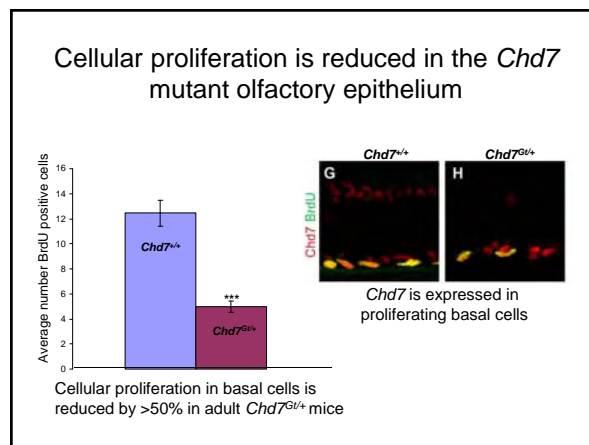
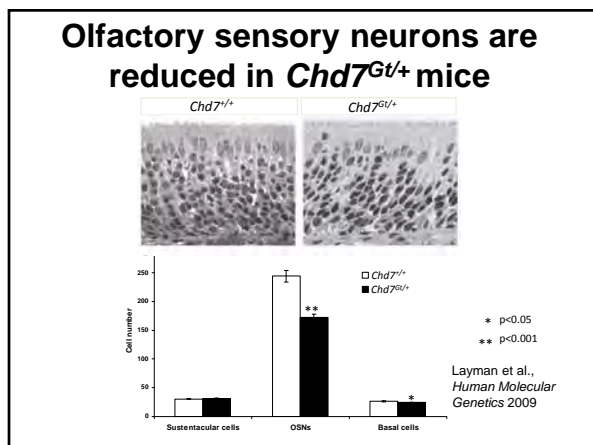
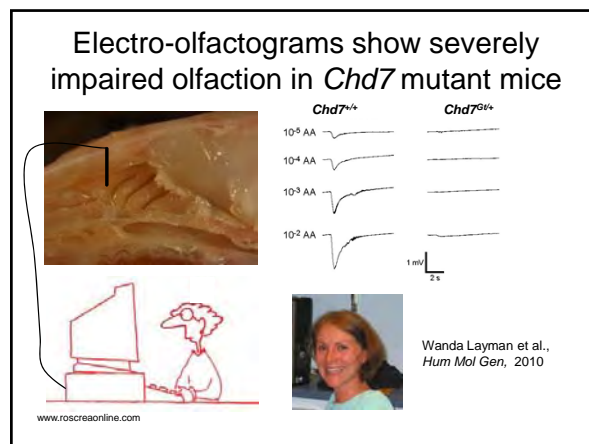
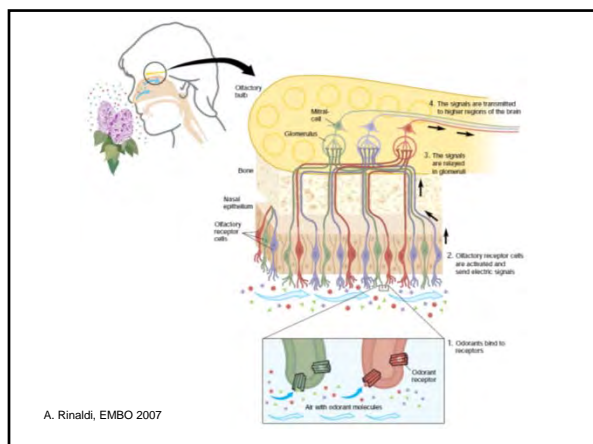
Hurd, unpublished

Cochlear hair cells are disorganized with loss of *Chd7*

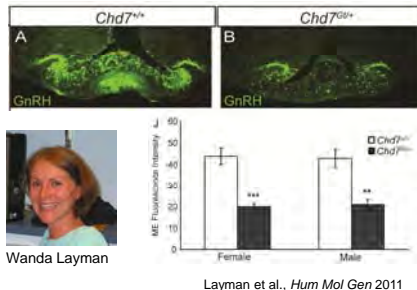
Hurd, unpublished

Lessons Learned

- CHARGE is a genetic disorder caused by *CHD7* mutations
- Mice are an excellent model of CHARGE
- The mouse *Chd7* gene is required for proper inner ear development and for hearing



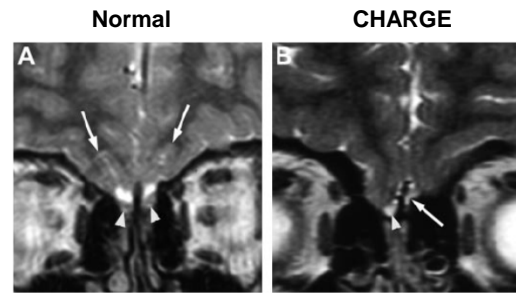
GnRH is reduced in the median eminence of *Chd7^{Gt/+}* mice



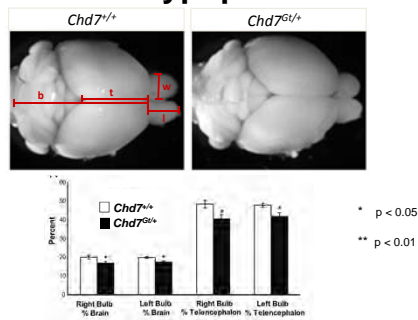
Wanda Layman

Layman et al., *Hum Mol Gen* 2011

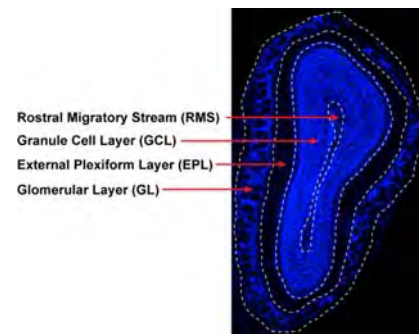
Olfactory bulb defects in CHARGE

Pinto et al., *J. Clin. Endocrinol. Metab.* 2005

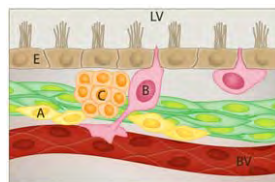
Chd7^{Gt/+} mice have olfactory bulb hypoplasia

Layman et al., *Human Molecular Genetics* 2009

Olfactory bulb anatomy



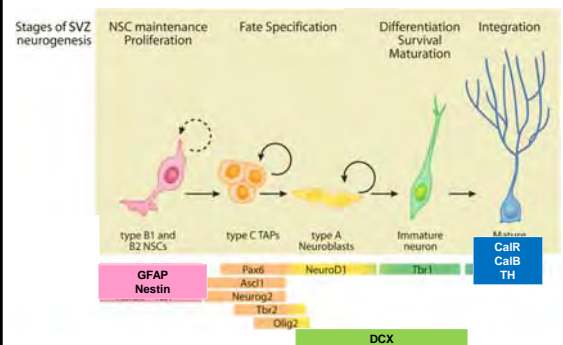
Stem Cells in the Subventricular Zone (SVZ) Give Rise to All Olfactory Bulb Interneurons



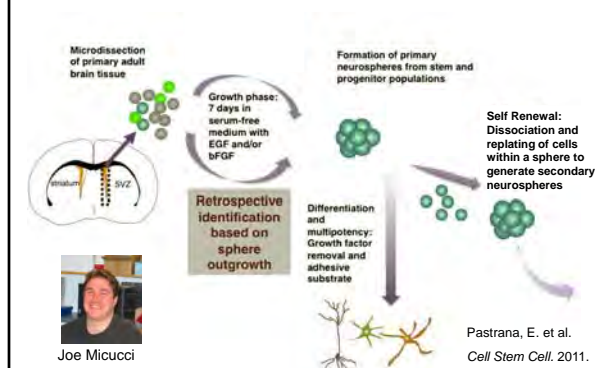
LV: Lateral Ventricle
 E: Ependymal Cell
 B: Type B stem cell
 C: Type C transit amplifying cell
 A: Type A neuroblast
 BV: Blood Vessel

Hsieh J. *Genes and Development* 2012.

Many Cell Types are Present in the SVZ

Adapted from Hsieh J. *Genes and Development* 2012.

Neurosphere Assay



Individuals with CHARGE syndrome have skeletal abnormalities

- 30%-50% of patients present with skeletal abnormalities (Brock et al., 2003; Tellier et al., 1998)
- Isolated cases
 - Neck and shoulder abnormalities (Issekutz et al., 2005)
 - Spine anomalies (Stromland et al., 2005)
 - Hypoplastic vertebrae (Jongmans et al., 2006)
 - Tracheoesophageal fistula (Lee et al., 2008)
 - Other hand and foot deformities (various authors)

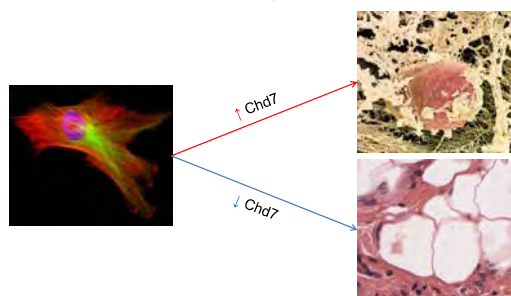
Hartshorne et al., 2011

Craniofacial features in CHARGE

- External ear abnormalities
- Micrognathia, glossoptosis (>95%)
- Cleft lip and/or palate (20%)
- Cranial nerve dysfunction (VII, VIII, IX, X)
- Dental abnormalities
- Tracheomalacia and/or laryngomalacia

Hartshorne et al., 2011; Hall and Hefner, 1999; CHARGE Syndrome Foundation

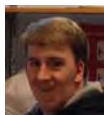
Chd7 is necessary for proper bone development



Adapted from Takada et al., 2007; Nature Publishing Group (Dennis Discher); Science Photo Library (Paul Gunning); University of Michigan Medical School (Michael Hortsch, Ph.D.)

Skeleton Preparations

- Isolation of bone and cartilage from post-natal mice
- Stain with **Alizarin red (for bone)** and **Alcian blue (for cartilage)**
- De-stain using gradient mixtures of glycerol and potassium hydroxide



Ethan Sperry

Lessons Learned

- Mice with heterozygous *Chd7* mutations have anosmia, fewer olfactory sensory neurons, and fewer GnRH neurons
- *Chd7* is required for normal proliferation and neurogenesis in the olfactory epithelium
- *Chd7* appears to be necessary for subventricular zone neural stem cell function and for skeletal development

Current Research Goals

- Identify critical target genes and interacting partners that mediate CHD7 function
- Characterize roles for CHD7 in adult cells and tissues using induced pluripotent stem cells
- Design regenerative strategies for treating CHARGE-related disorders
- Determine the cause of the remaining 15-25% of CHARGE cases not due to *CHD7* mutations

Acknowledgements

Current lab members

Jennifer Skidmore
Joe Micucci
Ethan Sperry
Mark Durham
Sophia Frank

UM Collaborators

Yehoash Raphael
Jeff Martens

Other Collaborators

Peter Scacheri (CWRU)
Bob Hevner (U Washington)

Funding

-NIH-NIDCD
-National Organization for
Hearing Research (NOHR)
-Hearing Health Foundation
-The CHARGE Syndrome
Foundation

Former lab members

Elizabeth Hurd
Wanda Layman
Elyse Reamer
Heather Poucher





Category: Medical/Genetics
(Research Updates)

Friday
Breakout Session #2:11: 1:00-2:00
Palomino 4 & 5

**Gene therapy induces nerve fiber
regeneration in the inner ear of deaf
mutant mice**

**Yehoash Raphael, Hideto Fukui, Yohei
Takada, Donna M. Martin**
Credentials & Organization
The University of Michigan, Ann Arbor

Presenter Information:

Yehoash Raphael is Professor of Otolaryngology, Head & Neck Surgery, working in Kresge Hearing Research Institute at The University of Michigan. He specializes in inner ear biology, with a special focus on ear trauma, repair and regeneration. The lab personnel study both hereditary and environmental ear disease and design therapies for prevention and for hearing restoration.

Drs. Hideto Fukui and Yohei Takada have performed the laboratory work presented at the meeting. They are Otolaryngology specialists originally from Kansai Medical University in the Osaka area, Japan.

Dr. Donna Martin is a colleague who works with Dr. Raphael on characterizing ears of a mouse model for CHARGE. Dr. Martin is also a spouse, and together, Donna and Yehoash are parents of a 19 year-old son, Noam Raphael, with CHARGE, and a 17-year old daughter, Maya Raphael. Both Noam and Maya are also attending the meeting.

Presentation Abstract:

The outcome of cochlear implant therapy depends on a healthy auditory nerve. We tested whether *BDNF* gene transfer into the cochleae of deaf mice can influence the fate of neurons. We determined that the diameter and number of nerve fibers in the auditory epithelium were increased compared to non-treated ears, and that spiral ganglion cell density in Rosenthal's canal was also increased. The data suggest that nerve fiber regeneration treatment may augment cochlear implant therapy.



Category: Medical/Genetics

Friday
Breakout Session #2:12: 1:00-2:00
Palomino 6 & 7

**Addressing Sensory/Oral
Placement/Feeding Difficulties
Associated with CHARGE Syndrome**

Whitney B. Pimentel, MA, CCC-SLP
TalkTools

Presenter Information:

Whitney B. Pimentel is a Speech Pathologist that specializes in Oral Placement, Feeding & Speech Therapy for clients of all ages and ability levels. She has a private practice in the East Valley of Phoenix, AZ. Whitney also lectures and provides evaluations and travel clinics for TalkTools.

Presentation Abstract:

Whitney will teach a unique approach to assessing the sensory system and, when it is appropriate, to begin feeding and Oral-Placement Therapy techniques. This presentation will include techniques to normalize the sensory system, improve nutritional intake, and maximize oral movements necessary for safe feeding as well as improving speech clarity. The information presented will assist therapists and other educators to create a therapeutic intervention program to address the needs of these individuals.

Addressing Sensory/Oral Placement/Feeding Difficulties Associated with CHARGE Syndrome

TALKTOOLS®
DISCOVER THE FEEL OF FEEDING AND SPEECH

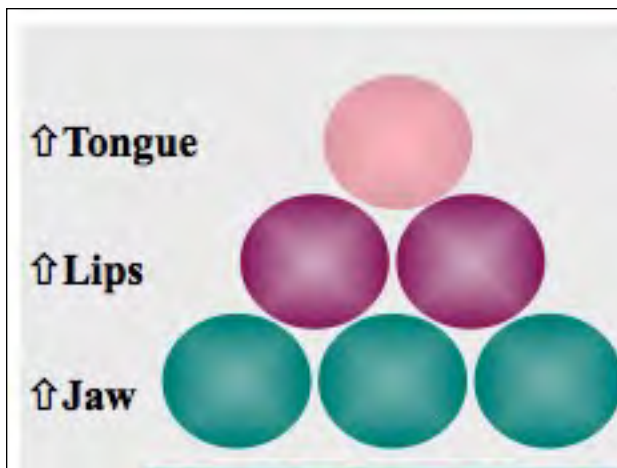
Presented by Whitney B. Pfeiffer
MA, CCC-SLP

The Feel of Speech

What is OPT vs Traditional Speech Therapy:

Traditional speech therapy presents information through visual and auditory stimulation. However, many individuals have difficulty learning through their eyes and their ears. OPT adds the "feel" of speech.





Common Deficits In CHARGE Syndrome

Deficits which effect sensory, feeding and speech clarity; each of these deficits may range from non-existent to severe.

1. Hypotonia
2. Visual Impairment
3. Hearing (ossicle infections)
4. Sensory deficits may range from minor to severe: smell, taste, touch
5. Oral tactile sensitivity: hyposensitivity, hypersensitivity, mixed sensitivity, flavoring sensitivity "The 'feel' of feeding and the 'feel' of speech"
6. Tactile defensiveness
7. Cognitive deficits

Addressing Sensory & Processing Using Sensi-Activities with CHARGE Syndrome

Common Deficits In CHARGE Syndrome

Deficits which effect sensory, feeding and speech clarity; each of these deficits may range from non-existent to severe.

8. Gap between expressive and receptive language skills
9. Weak jaw musculature: Symmetrical or Asymmetrical
10. Incomplete lip closure, decreased tongue mobility/grading results in limited retraction, lateralization, and tongue tip pointing
11. Weakness in the muscles of the velum
12. Blocked nasal passages: /m, n, ng/
13. Motor planning deficits
14. Difficulty coordinating oral airflow with vocalizations to initiate speech sounds production

Goals of Oral Placement/Feeding Therapy

- To improve feeding skills and nutritional intake
- To improve speech sound production and improve intelligibility/clarity

Goals of Oral Placement/Feeding Therapy

- To increase awareness of the oral mechanism
- To normalize oral tactile sensitivity
- To teach more normal movement patterns
- To increase differentiation of oral movements

a. *Dissociation*: The separation of movement, based on stability and strength, in one or more muscle groups.
b. *Grading*: The controlled segmentation of movement through spaces based on dissociation.
c. *Fixing*: An abnormal posture used to compensate for reduced stability which inhibits mobility.

Some Statements to Consider

- "We do not monitor our speech clarity by how it 'sounds' or how it 'looks.' Instead we base our assumption that we are speaking intelligibly on how it 'feels.'"
- "Why does my child bite and put everything in his/her mouth but avoids certain foods?"
- "Why does my child grind his teeth, suck his thumb, etc.? How can we eliminate these behaviors?"

The answer is SENSORY!

So how do we get started??

Muscle-Based Exercises(Oral-Motor)

Begin with a sensory program and a stable posture:

Work from whole body to mouth to achieve acceptance of touch and to develop trust.

Before you Begin to work on feeding or speech:

1. Establish a supported feeding position:

Stability in the body allows for mobility within the oral cavity



Before you Begin to work on feeding
or speech:

2. Evaluate the Sensory System:

- a. Toothette w/Vibrator
- b. Sensory Bean Bags
- c. Jigglers



Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:
An under-reaction to tactile input.

Addressing Sensory/Feeding Problems: Learning Goals, Assessment, and Intervention Strategies

Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:
An under-reaction to tactile input.

Tactile Hypersensitivity:
An over-reaction to tactile input

Addressing Sensory/Feeding Problems: Learning Goals, Assessment, and Intervention Strategies

Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:
An under-reaction to tactile input.

Tactile Hypersensitivity:
An over-reaction to tactile input

Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:
An under-reaction to tactile input.

Tactile Hypersensitivity:
An over-reaction to tactile input

Mixed Sensitivity:
Any combination of hyper, hypo or normal sensitivity.

Addressing Sensory/Praxis Issues Using Sensory Assessment with SMART Strategies

Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:
An under-reaction to tactile input.

Tactile Hypersensitivity:
An over-reaction to tactile input

Mixed Sensitivity:
Any combination of hyper, hypo or normal sensitivity.

Fluctuating Tactile Sensitivity:
Responses that change over time.

Addressing Sensory/Praxis Issues Using Sensory Assessment with SMART Strategies

Sensory Diagnosis/Tactile System

Tactile Hyposensitivity:

An under-reaction to tactile input.

Tactile Hypersensitivity:

An over-reaction to tactile input

Mixed Sensitivity:

Any combination of hyper, hypo or normal sensitivity.

Fluctuating Tactile Sensitivity:

Responses that change over time.

Tactile Defensiveness:

A learned tendency to respond negatively or emotionally to tactile input

Addressing Sensory/Oral Problems Using Sensi-Awareness with SENSAP Equipment

Before you begin to work on either feeding or speech:

1. Evaluate the Sensory System and make diagnosis
2. Eliminate tactile defensive behaviors by establishing trust.
3. Treating Sensory Deficits: Sensory Integration Deficits: Begin with a sensory warm-up as prescribed by an S.I. trained Occupational Therapist

Addressing Sensory/Oral Problems Using Sensi-Awareness with SENSAP Equipment

Before you begin to work on either feeding or speech

Oral Placement (Muscle-Based) Activities:

1. Speech: Use non-food items to increase strength and stability in the muscles of the abdomen, velum, jaw, lips and tongue as a prerequisite for the emergence of standard speech sound production
2. Feeding: Teach these movements prior to introduction of foods to improve bolus control, confidence in oral feedings and feeding safety.

Addressing Sensory/Oral Problems Using Sensi-Awareness with SENSAP Equipment

Before you begin to work on either feeding or speech:

Why is feeding so important:

*Nutritional Concerns

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

"How will Oral Placement Therapy help my child to speak more clearly?"

ADDRESSING SWALLOWING AND FEEDING DIFFICULTIES USING SECOND AND THIRD Molars

Before you begin to work on either feeding or speech:

Exercises for tube-fed children:

1. Associates movements in the mouth with feeding
2. Exercises in the mouth: The muscles that are used in feeding are the same muscles that are used in speech
3. When the child receives medical approval for oral feedings they will be ready to eat both from a sensory position and a strength position. Working on oral-phase feeding exercises improves swallowing proficiency

Before you begin to work on either feeding or speech:

Preventing tactile defensive behavior patterns associated with feeding:
These exercises will also improve jaw strength and teach the motor plan for chewing.

Exercises for infants without teeth:

1. Gloved finger
2. Infadent



Muscle-Based Exercises (Oral-Motor)

2. Jaw Exercises:

- Chewing on Back Molars
- ARK's Z-Vibe or Probe
- Chewy Tubes (Red – Yellow)
- ARK's Grabbers (Purple – Green)
- TalkTools® Jaw Grading Bite Blocks



Overview of Optimal Feeding Positions and Techniques

1. Purees: Spoon Feeding

- Placement of the spoon: Side, Front or pointed tip at lip midline?
- Wait for your child to close his/her lips before you remove the spoon



Addressing Sensory/Oral Processing Feeding Issues: Assessment with SLP/OT Strategies

Overview of Optimal Feeding Positions and Techniques

2. Liquids

- Cup Drinking: Sippy-cups, are they right for my child? What are the alternatives
- Straw Drinking: Why is the Straw Hierarchy so important?



Addressing Sensory/Oral Processing Feeding Issues: Assessment with SLP/OT Strategies

Straw #1



Addressing Feeding/Oral Posture/Feeding Issues Associated with SIB/ARF Disorders



Addressing Feeding/Oral Posture/Feeding Issues Associated with SIB/ARF Disorders

Overview of Optimal Feeding Positions and Techniques

3. Solids

- a) Cube or julienne stick shape?
- b) Why is it so important that my child learn to chew on his/her back molars?

Addressing Feeding/Oral Posture/Feeding Issues Associated with SIB/ARF Disorders

The Need for Practice

- *OPT activities must be practiced a minimum of 3 times per week
- *Once the skill is mastered it must be transitioned into function
- *OPT and traditional speech and language work together
- *Have fun and enjoy the successes!

Addressing Literacy in Preschool: Using Sound Assessment with DASH-2 Screening

Whitney B. Pimentel
M.A., CCC-SLP

TalkTools
1652 Wallace School Road, Suite 11
Chattahoochee, SC 29407
Tel: 888-529-2373
Email: info@talktools.com
www.talktools.com

Addressing Literacy in Preschool: Using Sound Assessment with DASH-2 Screening



Category: Behavior

Friday
Breakout Session #2:13: 1:00-2:00
Palomino 8 & 9

**“Why self-stimulation is a
good thing, and how and why we
should interpret it”**

David Brown
Educational Specialist
California Deaf-Blind Services
San Francisco State University

Presenter Information:

David Brown is a special education teacher who has been working with children with CHARGE syndrome for 30 years. He has written extensively about CHARGE, and travels the world giving presentations about various aspects of the syndrome, and helping to assess children alongside their families and local professional teams. In 2013 David will be spreading the word about CHARGE in person on visits to New Jersey, Sweden, Maryland, Minnesota, Arizona, Tennessee, and Germany.

Presentation Abstract:

As the most multi sensory impaired of syndromes, people with CHARGE are challenged to explore a range of apparently unusual behaviors in order to function effectively to satisfy their own needs. Many of these unusual behaviors seen in people with CHARGE are attempts to compensate for sensory losses and obtain the best and most reliable information possible, both from the environment and also from their own bodies. Other of these behaviors originate as attempts to modulate arousal levels – what we would call self-regulation. All these behaviors can be characterised as ‘self stimulation’, which is a normal part of human behavior but often occasions extreme and persistent opposition when seen in people with CHARGE. Indeed, in the field of special education self-stimulation, or ‘stimming’, is often regarded as a cardinal sin to be opposed and removed at all costs. This presentation will try to clarify the role of unusual postures and self stimulation behaviors, and will encourage more careful and informed observation as the essential prelude to any intervention.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013



Category: Medical/Genetics,
Family Support

Friday
Breakout Session #2:14: 1:00-2:00
Palomino 10

We Are In CHARGE

Catherine Rose, PhD/MBA

Presenter Information:

Catherine Rose is mom to Alexis (7) with CHARGE and Jessica (5). Catherine has a PhD and MBA and works for Philips as a Product Manager for Healthcare Lighting applications. Inspired by Alexis' vision impairments, Catherine is creating an interactive grid of color changing LED lights, called LightAide. In her free time, Catherine works to support other families in their challenging medical journeys.

Catherine has collaborated with Cynthia Antaya for this presentation. Cynthia is an individual with CHARGE Syndrome only diagnosed at age 27 when her son, Brady, was born and diagnosed with the same syndrome. Cynthia is a Nationally Certified School Psychologist and works with Methuen Public Schools in Massachusetts with children who have varying disabilities. Cynthia and her husband, Keith, were given a laundry list of things Brady would never be able to do given his CHARGE Syndrome diagnosis when he was born. Six years later, Brady has proven he is in CHARGE.

Presentation Abstract:

The presentation will be very interactive, fun and engaging – sharing stories from other families and their experiences along the CHARGE journey as well. The presentation will also encourage parents to be empowered to advocate for their children – to step into the challenging role we must fulfill. We will share personal experiences as teaching lessons in challenging authority and being successful in achieving outcomes in challenging environments where parents are not trusted as experts.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013



Category: Medical/Genetics,
Family Support

Friday
Breakout Session(s) #2:15, 1:00-2:00
Dorado

**CHARGE 102: Language and
Communication Options
& Questions, Questions,
Questions**

Nancy Hartshorne, PhD
School Psychology
Delta College
Bay County, Michigan

Presenter Information:

Nancy Hartshorne, Ph.D. is a Professor of Psychology, Delta College in Michigan. She is an educational psychologist and the mother of Jacob, a young adult with CHARGE. Nancy has been involved with language and education of children with CHARGE for 20 years.

Presentation Abstract:

Communication, Communication, Communication. All learning stems from communication. Individuals with CHARGE syndrome can have enormous communication challenges, stemming from hearing and/or visual impairments, mobility issues, and medical problems. CHARGE 102 will focus on an introduction to different modes of communication available, and the importance of the earliest stimulation to encourage these modes.

In this session, we will specifically leave time for your questions. Nancy, Meg, Sandy and Kim will all be available to answer questions. We, along with a few "experienced" parents (who have been to previous conferences) will address your questions and try to help guide you to other sessions or people to best address your needs.

Language/Communication: What's so Hard About That? Isn't it a Natural Process?

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

- Imagine you were just born into the world.
- Imagine you are hooked up to tubes, monitors, and pumps, some of which are painful.
- Imagine you have just been given a tracheostomy or gastrostomy.
- Imagine you have been in the hospital for weeks on end.
- Imagine you are recovering from several surgeries, tests, and procedures, with the pain and fatigue that goes with those.
- Imagine 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 eye.
- Imagine your hearing is limited: You can hear very little from one ear, and nothing from the other.
- Imagine you cannot move to what you want to explore: Your muscles are weak, you can't stand the feel of the carpet/floor on your hands or feet.
- Imagine 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.
- Imagine all of this doesn't allow for growthful parent/baby interaction.

Now: How “available” are you to learn?

Communication Modes Used by Individuals with CHARGE Syndrome

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

- 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.



Category: Behavior,
Medical/Genetics

Friday
Breakout Session #3:17: 2:15-3:15
Palomino 6 & 7

**The Challenge of Mealtimes for
Children with CHARGE**

Steve Rose
Head of Children's Specialist Services /
Specialist Speech and Language Therapist
Sense

Presenter Information:

Steve Rose is a Health Professions Council registered speech and language therapist specialising in working with people with deafblindness. He has previously worked as an intervenor and for Sense working with family support groups until returning to study to train as an SLT. Graduating from UCL in 2003 he subsequently worked in special schools in North London with children with physical disabilities, sensory impairments, autistic spectrum disorders and learning difficulties. Steve has particular interest in the development of eating and drinking skills and early interventions, including parent-child interaction therapy. He has recently completed his MEd in deafblindness at Birmingham University and currently heads Sense Children's Specialist Services.

Presentation Abstract:

It is well documented that individuals with CHARGE syndrome may face difficulties with eating and drinking. There is emerging literature that documents the prevalence of these issues and support intervention for such difficulties.

This presentation explores literature of the prevalence, nature and range of difficulties and the subsequent challenges presented. The features of CHARGE that relate to difficulties with mealtimes are also identified. It will outline some observations on the nature of the challenges facing children with CHARGE seen at our centre in North London. These challenges will be discussed in the context of case studies to illustrate individual challenges and possible management strategies.

It is recognised that the early oral experiences of children who experience challenges eating and drinking have an impact on later development and prognosis. The involvement of cranial nerve dysfunction in CHARGE leads to a high percentage of children having eating, drinking and swallowing disorders – up to an estimate of 80% of all cases (Sanlaville and Verloes 2007). In addition there are numerous other factors and co-occurring features that impact on these difficulties, combined these present significant challenges for the child that are often seen as ‘behavioural feeding issues’.

Case studies will identify some of the ‘mealtime behaviour’ of children seen at our centre and begin to develop some understanding of the challenges facing this group of children and the sort of support that could help to overcome them. This is an area where there is less specific literature and it is hoped that this paper will begin to contribute to a greater understanding of these issues for this group of children.

3rd Professional Day & 11th International CHARGE Syndrome Conference
Fairmont Scottsdale Princess Hotel, Scottsdale, AZ July 25-28, 2013

The challenge of mealtimes for children with CHARGE syndrome



Steve Rose

Speech and Language Therapist / Head of Children's Specialist Services
11th International CHARGE Syndrome Conference
July 25-28 2013



The plan for this session is to



- explore the nature of eating and drinking difficulties in it's broadest context.
- look at the neurological control required to eat and drink, including to trigger the swallow (a pattern elicited response).
- look at which aspects of CHARGE impact on eating and drinking, exploring themes which have emerged from the evidence base in this area.
- identify other issues based on observations made within our service paying particular attention to sensory-based feeding disorders illustrated with case studies.

© Sense 2013

2



"For most of us, food and drink are sources of pleasure; they satisfy hunger and thirst, help to structure our day and provide opportunities for social interaction"

(Winstock 2007 p.1)

© Sense 2013

4

"The primary goal in managing eating and drinking difficulties should address the child's safety [of swallow], nutrition and hydration needs along with emotional well-being of the whole family."

(Winstock 2007 p 128)

© Sense 2013



Challenges



- 1) children who present with some sort of oral / motor problem (dysphagia)
- 2) children who present with a sensory based feeding difficulty
- 3) children who have difficulty organising the transfer of food to their mouth.

© Sense 2013

6

Risks

Dysphagia:

- Dehydration and malnutrition (leading to organic failure to thrive)
- Aspiration
- Choking
- Altered development of eating and drinking skills
- Infection (secondary to previous risks)

Sensory based feeding difficulties:

- Dehydration and malnutrition (leading to non-organic failure to thrive)
- Altered development of eating and drinking skills

© Sense 2013

7

Neurology and swallowing

The normal swallow is a four stage process

- Oral preparatory stage – food is manipulated in the mouth.
- Oral stage – tongue pushes food back.
- Pharyngeal stage – swallow reflex triggered, food moves down through pharynx.
- Oesophageal stage – food moves down to the stomach.

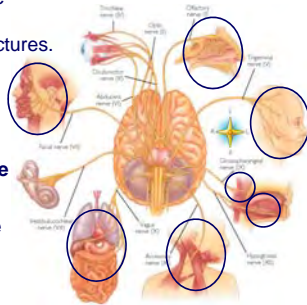
© Sense 2013

8

Neurology and swallowing

Cranial Nerves involved in the Innervation of the oral and pharyngeal muscles and structures.

- I Olfactory Nerve
- V Trigeminal Nerve
- VII Facial Nerve
- IX Glossopharyngeal Nerve
- X Vagus Nerve
- XI Spinal Accessory Nerve
- XII Hypoglossal Nerve



© Sense 2013

CHARGE

79 - 90%
of children with
CHARGE have
feeding
difficulties

(Blake et al 1998, Harvey et al 1991, White et al 2005, Hartsthorne et al 2005, Dobbelssteyn et al 2008)

© Sense 2013

10

Characteristics of CHARGE associated with eating and drinking difficulty.

Motor related

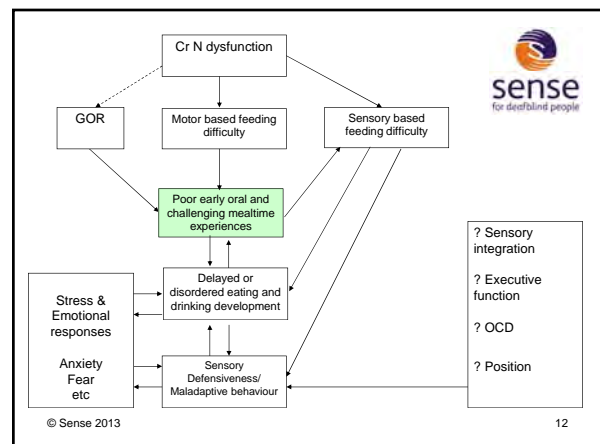
- Cranial Nerve dysfunction
 - Smell (Cr N I)
 - Weak suck / poor chew (Cr N V VII XII)
 - Swallowing difficulty (Cr N V IX X XI)
 - GOR (Cr N X)
- Respiratory problems
- Cleft (lip, palate or both)
- Choanal Atresia

Sensory related

- Cranial Nerve dysfunction
- GOR
- Long term NG tube
- Discomfort (related to GOR?)
- Numerous medical interventions

© Sense 2013

11



© Sense 2013

12

Our Assessment Centre

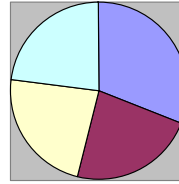


- A:** Children who are being non-orally fed
- B:** Children who are managing to eat orally
- C:** Children who are coping with oral eating (but it's not all plain sailing)
- D:** Children who are coping with oral eating and experiencing delay with the transition to a normal diet

© Sense 2013

13

Our Assessment Centre



- ☐ **A** Children who are being non-orally fed (31%)
- ☐ **B** Children who are managing to eat orally (23%)
- ☐ **C** Children who are coping with oral eating (but it's not all plain sailing) (23%)
- ☐ **D** Children who are coping with oral eating but experiencing delay with transition to a normal diet (23%)

© Sense 2013

14

Case studies



© Sense 2013

15

Preparing for mealtimes



- Take time
- Involve the child in preparation
- Set up the environment
- Sit in a good seating position
- Prepare the face / hands (massage?)
- Use the right cues (more, enough, stop!)
- Establish sensory equilibrium – Sensory diet activities before the mealtime
- Think about starting, continuing and finishing
- Prepare the right texture foods (and separate tastes)

© Sense 2013

16

During mealtimes



- Use the right utensils
- Relax!
- Don't have a battle
- Take a break – consider little and often?
- Think about the social elements
- Consider use of distractors and motivators

© Sense 2013

17

Food Play Texture Hierarchy



Easy			Challenging
Firm and dry: <ul style="list-style-type: none"> • Dry textures fall away and the child can control contact with the texture. • No brushing or wiping is required 	Soft and Dry: <ul style="list-style-type: none"> • Dry textures that mostly fall away, some particles or bits may stick to the hand, • Brushing residue required to break contact 	Wet: <ul style="list-style-type: none"> • Wet textures that maintain contact with the hand but the child can easily break contact with the texture • Requires wiping to break contact with texture. 	Wet and sticky: <ul style="list-style-type: none"> • Wet textures that stick to the hand and the child has least control when breaking contact with the texture. • Requires repeated wiping to break residue away.
<ul style="list-style-type: none"> • Marzipan • Ready roll icing • Broken pancakes • Cereals – cornflakes, rice krispies, cocoa puffs etc. • Uncooked noodles (hard) • Raw vegetables (either whole or in chunks – but dry) • Liquorice • Snack foods – crisps, wotsits, 	<ul style="list-style-type: none"> • Icing sugar • Cocoa powder • Dry angel delight mix • Dry porridge oats • Granulated sugar • Sherbert • Cake decorations – hundreds and thousands, strands etc. 	<ul style="list-style-type: none"> • Water • Cooked noodles • Cooked lentils • Cooked pasta / spaghetti • Cooked rice • Jelly • Mashed potato • Ice cubes • Piping icing • Custard • Yoghurt • Baked beans • Puréed fruit and vegetables 	<ul style="list-style-type: none"> • Malted chocolate • Mashed banana • Angel delight • Porridge • Ice cream • Tinned Spaghetti • Chocolate spread • Jam, lemon curd etc. • Ketchup • Sweet and sour sauce

© Sense 2013

© Steve Rose, Specialist Speech and Language Therapist, Sense, 2009

Oral motor / awareness programmes



Conclusions for management

- Proactive management promoting positive mealtime experiences
- Whilst anatomical and medical aspects are being managed the introduction of a positive oral stimulation program is considered and where possible implemented
- Mealtime assessment should be undertaken between SLT and /OT colleagues and within the context of a sensory integration approach.

© Sense 2013

20

"need for sensory integrative approach to management of eating and drinking. Feeding is a highly integrated multisystem skill and many contemporary clinicians support the approach that it is not helpful to focus solely on the child's mouth but to consider the whole child, including the environment, the child's sensori-motor profile and the context of mealtimes when developing a treatment plan" (Maune 2007 p.4)

© Sense 2013

21

Conclusions for management

- Multi-professional, trans-disciplinary management .
- Transitions are planned and reviewed frequently, with a duty to promote transition to oral feeding.
- Investigations should be undertaken for GOR and aspiration for all individuals with CHARGE syndrome
- Intervention is ongoing, multidisciplinary and multi-sensory system

© Sense 2013

22

"All children with CHARGE syndrome to be followed proactively by a multidisciplinary team consisting of gastroenterologist, ENT surgeon, nutritionist and other feeding specialists (occupational therapist and/or speech and language pathologist) to ensure appropriate management of feeding issues"

Dobbelsteyn et al 2008 p 132


© Sense 2013

23

"The presence of gastro oesophageal reflux and aspiration early in life will increase the probability of ongoing feeding problems.....
Dobbelsteyn et al 2008 p 132

© Sense 2013

24



"follow up should involve multi disciplinary feeding team and parents should be made aware of the potential long term feeding issues"
Dobbelsteyn et al 2005 p. 99

Contact Details

Steve Rose
Head of Children's Specialist Services
Specialist Speech and Language Therapist - Deafblindness

Children's Specialist Services
Sense, Novellus Court
61 South Street
Epsom
Surrey
KT18 7PX
tel: 0845 127 0076
email: steve.rose@sense.org.uk

Thank you to all the children and families who supported this presentation.





Category: Behavior

Friday
Breakout Session #3:18: 2:15-3:15
Palomino 8 & 9

**Self-regulatory strategies for children
with CHARGE syndrome**

Maria A Ramirez
**Doctoral Student –Central Michigan
University**

Presenter Information:

Maria Ramirez is a Doctoral student in the School Psychology program at Central Michigan University. For the past three and a half years she has been working with Dr. Tim Hartshorne exploring self-regulation in children with CHARGE. Her current research focuses on the assessment and validation of a Fun Chi video to be used in future research to assess the effects of Fun Chi on sleep, balance, and self-regulation in children with CHARGE.

Presentation Abstract:

The four windows of self-regulation (physiological, behavioral, cognitive, and emotional) provide the perfect areas for intervention in children with CHARGE. Although individuals with CHARGE may often use compensatory behaviors to aid in regulating their behavior, because of the presence of multisensory impairments and maladaptive patterns of behavior, positive self-regulatory strategies may at times be compromised. Using the four windows of self-regulation may prove to be an invaluable tool in understanding the function of the child's behavior, identifying the child's behavioral strengths that we can build up on, and in identifying specific self-regulatory areas to target for intervention. Strategies to enhance self-regulation in both the school and home will be presented. This presentation will be the third, preceded by Tim Hartshorne in the first hour and David Brown in the second hour.

FOSTERING SELF-REGULATORY STRATEGIES IN CHILDREN WITH CHARGE SYNDROME

Maria Alejandra Ramirez
School Psychology Doctoral Student
Central Michigan University

Agenda

1. Overview of 4 areas/windows of Self-Regulation
2. Example of Self-Regulation
3. What we know about Self-Regulation in children with CHARGE syndrome
4. Using the Windows of Self-Regulation to *identify strengths and identify difficulties*
5. Using the window of self-regulation to *prioritize areas to target*
6. In Detail: Specific strategies to foster each window of self-regulation



Discussion Goal 1:
Quick Overview Self-Regulation and the 4 Windows of Self-Regulation



What is Self-Regulation?

- Adaptive and flexible management of four domains.
 - Physiological, behavioral, cognitive, and emotion
- In order to describe regulation as “self-regulation” two components are necessary:
 - 1) self-awareness of the process
 - 2) a subsequent goal-directed action.
- Self-regulation must begin with a goal, and that goal describes what you want to have happen and what you must do to make that goal happen.
- Self-regulation involves gauging internal and external stimuli and responding appropriately under environmental expectations.

Self-Regulation:



→ **Goal** →



What are the 4 Windows of Self-Regulation?



Physiological



Behavior



Cognitive



Emotion

What is Physiological Self-Regulation?



Physiological

- Self's ability to react and alter its own states and responses to meet the needs of the body.
- In great part under the control of the somatic, endocrine and autonomic nervous systems.

What is Behavioral Self-Regulation?



Behavior

- Awareness of a behavior
- And choosing those behaviors most adaptive toward achieving a goal.
- Goal directed and purposeful behavioral patterns consisting of:
 - one's ability to inhibit,
 - regulate, pace, and delay gratification

■ Jahromi and Stifter (2008)

What is Cognitive Self-Regulation?



Cognitive

- Voluntarily regulating thoughts and mental processes by balancing inhibition and initiation of behavior in order to achieve a goal
- Involves
 - Attention
 - Memory
 - Learning (as well as using prior learning)
 - Reasoning
 - Problem-solving
 - Decision-making
 - Metacognition



What is Emotion Self-Regulation?



Emotion

- Using emotion regulation, the individual analyzes, controls, alters, or prevents behaviors related to the adaptive expression of emotions
 - (Lam & Lewis, 2010).
- May occur at different points in time during the emotional response



Discussion Goal 2: Example of Self-Regulation



Example: Dentist Appointment

- **Physiological:**
 - You make sure to brush and floss your teeth really well a couple days before the appointment
- **Behavioral:**
 - You make sure to put the appointment time/day in your planner
 - You get a babysitter
- **Cognitive:**
 - You mentally prepare yourself for the procedure
- **Emotion:**
 - You tell yourself the pain will be over soon
 - You ask for a lot of pain medicine to make sure you feel no pain



Discussion Goal 3:
What we know about Self-Regulation in children with CHARGE syndrome



What do we know about Self-Regulation in CHARGE?

- Multi-sensory difficulties may limit exposure to environmental stimuli, their exposure to interactions and reactions to the environment.
- As DeGangi (2000) states, early deficiencies in self-regulation may lead to challenging behavior, and deficits in attention and inhibition.

What do we know about Self-Regulation in CHARGE?

□ Physiological:

- Brown (2005) notes that individuals with CHARGE syndrome are truly multi-sensory impaired, often having challenges with vision, hearing, balance, touch, temperature, pain, pressure, smell, breathing, swallowing, eating, drinking, digestion, and temperature control

What do we know about Self-Regulation in CHARGE?

□ Behavior:

- May display behaviors typical of individuals with: Autism Spectrum Disorder, ADHD, OCD, Tourette's syndrome, and Deaf Blindness (Hartshorne & Cypher ,2004).
- These may include: restricted range of interest, stereotyped movements, fidgeting with objects, preference for certain objects or people, tactile defensiveness, staring at lights, vocal stimulation.

What do we know about Self-Regulation in CHARGE?

□ Cognitive:

- Children with CHARGE may present with executive dysfunction.
- Specifically in the areas of *shifting, monitoring, and inhibiting*.
 - Hartshorne, Nicholas, Grialou, and Russ (2007)

What do we know about Self-Regulation in CHARGE?

□ Emotion:

- This area is much less explored than the other areas of self-regulation
- Given that children with CHARGE have difficulty in the other areas of self-regulation and all the areas are related....it is possible that this may also be an area of difficulty.

Discussion Goal 4:
Using the Windows of Self-Regulation to:

1. *identify strengths*
2. *identify difficulties*



Identifying Regulatory Strengths/Difficulties in YOURSELF



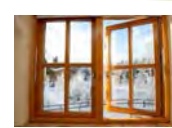
Physiological



Behavior



Cognitive



Emotion

Identifying Regulatory Strengths/Difficulties in YOUR CHILD



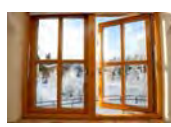
Physiological



Behavior

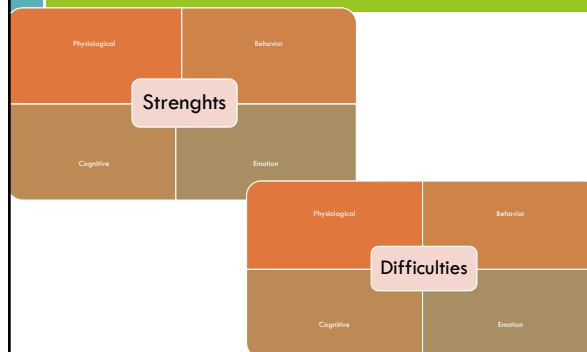


Cognitive



Emotion

Complete handouts for yourself and child



Discussion Goal 5:
Using the Windows of Self-Regulation to
Prioritize areas to target



Prioritizing Areas to Target

Things to consider:

- ☐ What would increase the most function?
- ☐ What would increase quality of life?
- ☐ What would save time?
- ☐ What would save resources?
- ☐ Do I have time to target this area?
- ☐ Do I have the resources to target this area?
- ☐ Are the final outcomes worth it?

Example:



Discussion Goal 6:

Specific Regulatory Strategies to Foster Each Window



Strategies: Behavior

Factors to consider:

- ☐ ID problematic Behavior
- ☐ What preceded the behavior?
- ☐ What follows the behavior?
- ☐ Who is present when the behavior occurs?
- ☐ Where does the behavior take place?

Strategies: Behavior

- ☐ Modeling, Role-playing, and Reinforcement
- ☐ Predictable routines
- ☐ Visual Schedule
- ☐ Increasing choices
- ☐ Pre-correction
- ☐ Red Dot Timer
- ☐ First-Then Statements
- ☐ Scaffolding
- ☐ Requesting breaks with visuals
- ☐ Pressure vests and deep tissue massages

Strategies: Physiological

- ☐ Sensory stimulation
- ☐ Environmental modifications
- ☐ Relaxation Therapies
- ☐ Diet
- ☐ Feeding
- ☐ Toileting
- ☐ Sleep issues

Strategies: Cognitive

- ☐ Modeling thinking, planning, and inhibitory strategies
- ☐ Modification of stressful environments
- ☐ Preparation to enter stressful environments
- ☐ Use of mind-body and technology practices to foster concentration and inhibition
- ☐ Take advantage of the child's favorite activities to imbed teaching of waiting and engaging.

Strategies: Emotion

- To develop awareness of emotion: teaching feeling vocabulary (modeling, scaffolding, and reinforcement)
- Mirroring feeling and modeling appropriate emotional responses
 - "I am happy, this is what happy looks like"
 - "You look upset, like this"
- Repetition and rehearsal of skills
- Use of visuals: colors, faces, traffic lights – to represent feeling or state
- Recognizing triggers that produce emotional response
- Teaching strategies to deal with emotions (deep breathing, location to calm down, attachment objects, etc.)
- Using all daily events to teach feelings, reactions, and modeling appropriate ways of coping

Strategies: Emotion

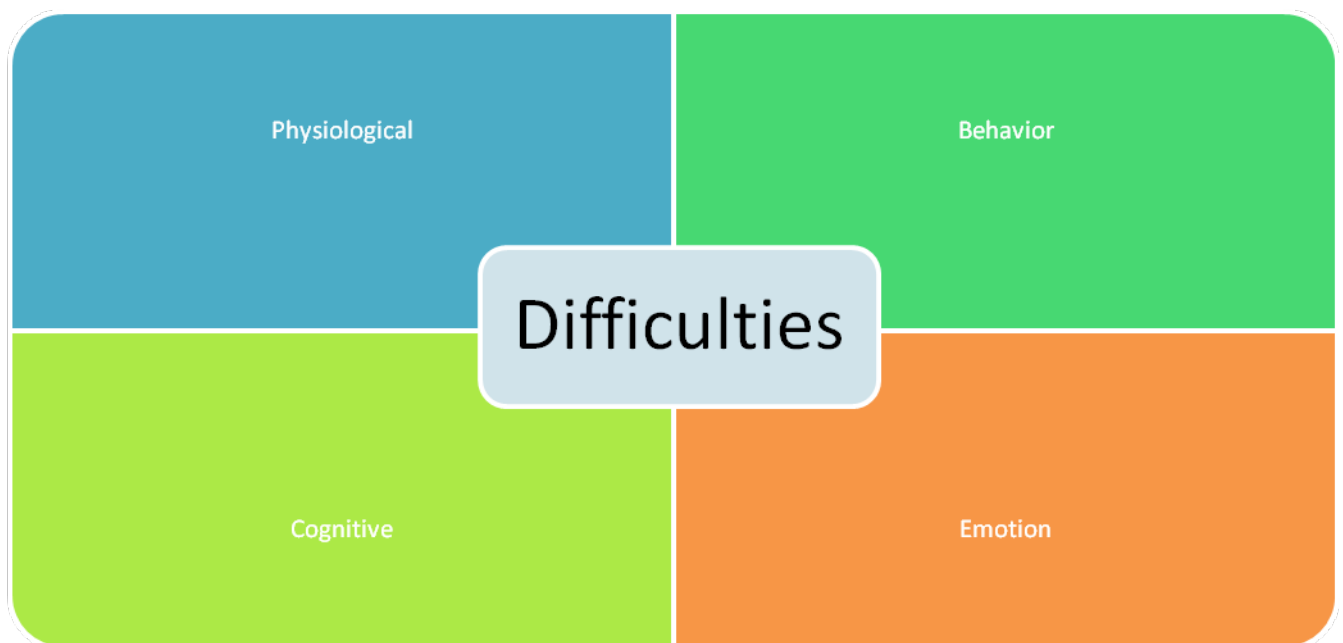
Remember to:

- Teach in different environments (home, school, grocery store)
- Teach with different people (peers, parents, siblings, etc.)
- Reinforce all appropriate behaviors in new environments

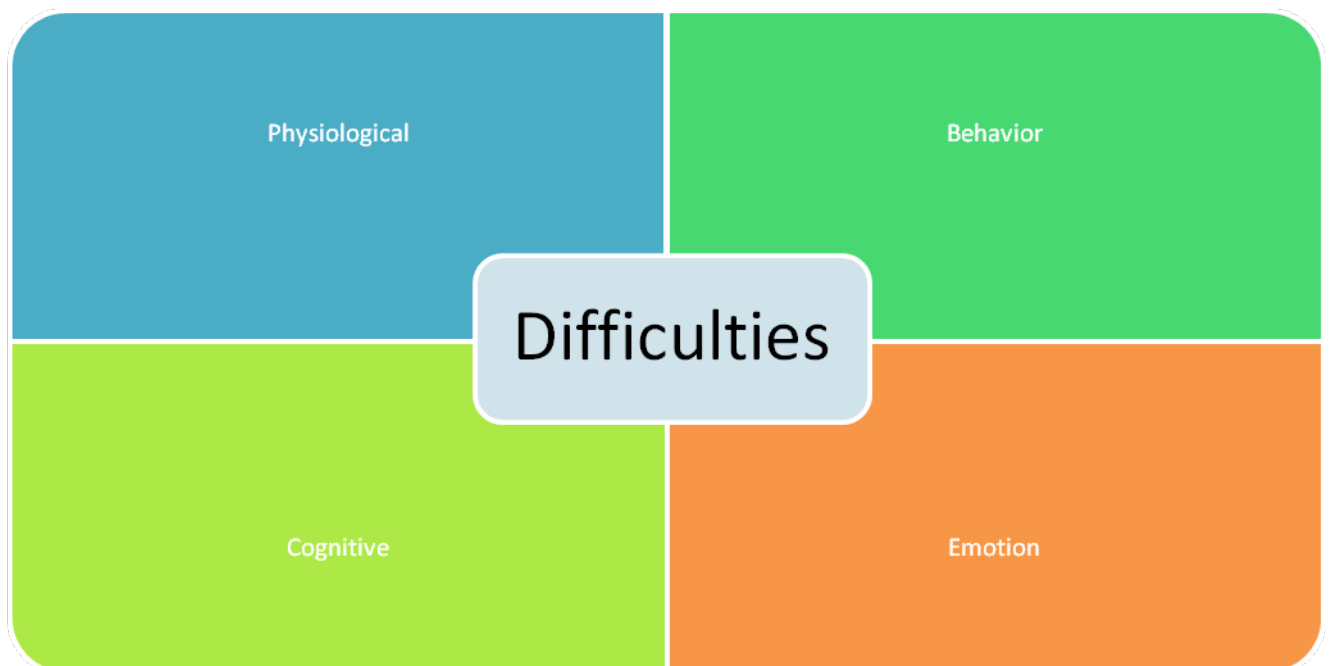
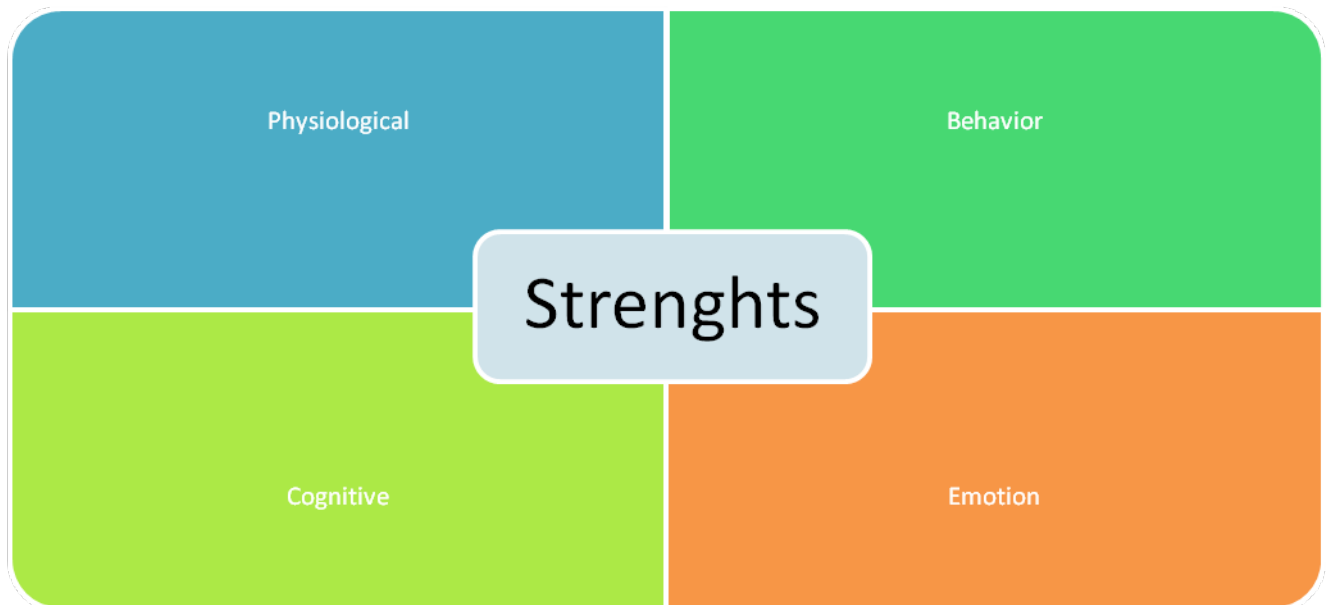
Thank you for your time!!!

If you have questions, please contact me at:
ramir1ma@cmich.edu

Identifying Regulatory Strengths and Difficulties in ME



Identifying Regulatory Strengths and Difficulties in MY CHILD





Category: Family Support

Friday
Breakout Session #3:19: 2:15-3:15
Palomino 10

FATHERS' FORUM

John Gaudin

Presenter Information:

John Gaudin is an auditor for the Windham School District in Huntsville, Texas. He serves on the Board of Directors for Texas Chargers Inc., a non-profit group dedicated to serving those with CHARGE syndrome along with their families and professionals who support them. He has facilitated the Fathers' Forum at the annual Texas Chargers Retreat for the past 4 years. He is happily married to Crissy, and they have 3 beautiful children – Jacob (12), Rachel (10), and Joey (8).

Presentation Abstract:

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



Category: Medical/Genetics,
Family Support

Friday
Breakout Session(s) #3:20, 2:15-3:15
Dorado

**CHARGE 103: New Families:
Breathing and Anesthesia as
critical issues
&
Questions, Questions, Questions**

Kim Blake, MD, Developmental Pediatrics
Dalhousie University, Halifax NS Canada

Presenter Information:

Kim Blake is a developmental pediatrician who first became involved with CHARGE in England more than 20 years ago. She helped start the UK support group before moving to Eastern Canada. She has been particularly interested in issues affecting adolescents with CHARGE and the breathing issues and the complications that go along with breathing. Kim has been instrumental in teaching medical students and residents about CHARGE - making sure the next generation benefits from her accumulated expertise. She has presented at every CHARGE Syndrome Foundation conference.

Presentation Abstract:

Anesthesia, botox, sleep apnea – what's going on with breathing in CHARGE? The typical two year old with CHARGE has had more than a half dozen surgeries. Many babies and children with CHARGE have unusual reactions to anesthesia and other issues which affect breathing. Kim will present information on some of these key medical issues in infancy – things you need to know when considering various surgeries and other procedures.

Again in this session, we will specifically leave time for your questions. Nancy, Meg, Sandy and Kim will all be available to answer questions. We will also be joined by several “experienced” parents (who have been to previous conferences). We will all answer questions and help you find the sessions and people to best address your needs.

CHARGE 101 Anaesthesia, Botox, Sleep Apnea and FAQ's

Dr. Kim Blake, MB, MRCP, FRCPC



11th International CHARGE Conference
Arizona, July 2013

Objectives

- To help you understand the risks of anaesthesia in CHARGE syndrome.
- To explain the use of Botox injections into the salivary glands.
- Research findings about sleep apnea and benefits of tonsillectomy and adenoidectomy.

There are Always Risks of Complications with Anaesthesia

- "...you sign a consent"
- Are you informed?
- Are Individuals with CHARGE Syndrome More at Risk?

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



APSEA 2006

Growing up With CHARGE Syndrome



Age 0-2 years: 7 surgeries



Age 2-4 years: 3 surgeries



Age 4-6 years: 6 surgeries

Kennedy

Kennedy's Four ICU Admissions

- 2 weeks – open heart surgery
- 6 months – G-tube/fundoplication extubation attempted (x 3)
- 18 months – aspiration pneumonia
- 6 yrs – heart surgery – pneumonia after heart surgery

Postoperative Airway Events of Individuals with CHARGE Syndrome

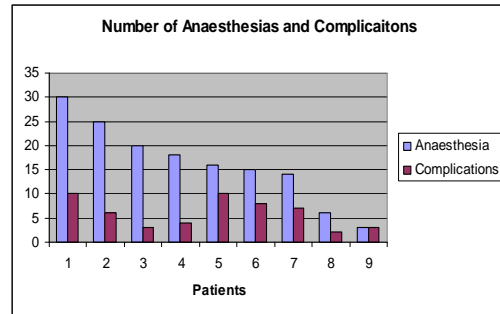
Population n=9
Mean age 11.8 years (± 8.0)
215 surgeries (mean 22 per child)
147 anaesthesias (mean 16 per child)



Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Blake K, International Journal of Pediatric Otorhinolaryngology, 2009

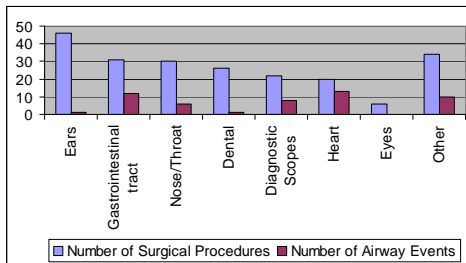
Results



35% (51/147) of anaesthesias resulted in post-operative events (>60% were major)

Results

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



Results

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

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

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



MacKenzie & Kennedy sharing lunch

Results

Feeding procedures and rates of postoperative airway events.

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

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

Summary

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

Blake K. International Journal of Pediatric Otorhinolaryngology, 2009

Summary cont'd

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

Frederick's Story

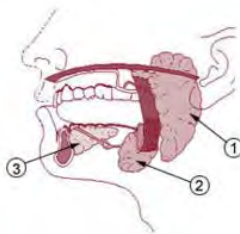


Freddy at 2 Months

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

Site of Botox Injections

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



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

Freddy at 7 Months

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

Botox Injection

Submandibular Gland Via Ultrasound and Parotid Gland
by Palpation



10 Botox Units/gland

Botox Injection

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



Freddy's growing up!



Summary - Botox

- Botox injections into the salivary glands may help to reduce the oral secretions (needs repeating every 4-5 months).
- Reduction in oral secretions may help prevent aspiration and pneumonia
- May help prevent and/or removal tracheostomy

MacCuspie. AJMG 2011

MacKenzie's Story



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

Understanding Sleep Apnea in Children with CHARGE Syndrome In Children 0-14 years old

Authors:

Carrie-Lee Trider
Dr. Gerard Corsten
Dr. Debra Morrison
Meg Hefner
Dr. Sandra Davenport
Dr. Kim Blake



Carrie-Lee & Freddy

Trider CL. International Journal of Pediatric Otorhinolaryngology 76 (2012) 947-953

Objectives

- To determine the prevalence of Obstructive Sleep Apnea (OSA)
- Apply two validated questionnaires to the CHARGE Syndrome population
 - Brouillette Questionnaire
 - Pediatric Sleep Questionnaire
- Assess the quality of life after treatment for OSA

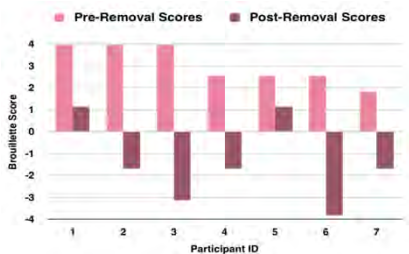
Thank you to the 34 Participants Results

- Over 80% of the study population had a diagnosis of sleep apnea
- Brouillette scores identified obstructive sleep apnea in CHARGE Syndrome
- Pediatric Sleep Questionnaire – Sub Scale was useful in diagnosing sleep apnea
- The OSA -18 Questionnaire (Quality of Life) demonstrated all treatments for obstructive sleep apnea improved quality of life

Tonsils and Adenoids

- How many of your children have had a tonsillectomy and/or adenoidectomy?
- How many of your children had anaesthetics after that?
- How many of your children had improved recovery after subsequent surgeries

Removal of Tonsils & Adenoids (Preliminary Results)



Scores >3.5 highly predictive of OSA, between -1 to 3.5 suggestive for OSA, <-1 absence of OSA

Take Home Messages

- Your children are at high risk of post-operative anaesthesia complications. Combining procedures during one anesthesia does not increase the risk of post-operative airway events.
- The anaesthesiologist needs to be aware that, even with simple procedures, the individual with CHARGE syndrome is at high risk of post-operative events.

Take Home Messages

- Botox into the salivary glands may help oral secretions
- Obstructive sleep apnea is highly prevalent in the CHARGE Syndrome population
- Remove all tonsils and adenoids may be beneficial

Thank You From Four Friends

