

Thursday, July 30, 2015 General Session: Utopia Ballroom 9:00-10:45am

Atypical features associate with CHD7 mutations and CHARGE syndrome: a proposal for revised clinical diagnostic criteria

Caitlin L. Hale, B.S., Jane Schuette, M.S. C.G.C., Stephanie Bielas, Ph.D., and Donna M. Martin, M.D., Ph.D., Departments of Human Genetics and Pediatrics, The University of Michigan, Ann Arbor

Presenter Information:

Ms. Hale is an M.S. student in Genetic Counseling at the University of Michigan. She is interested in the wide phenotypic spectrum associated with many complex genetic conditions, and how genotype-phenotype correlations can improve medical management. She has dedicated her Master's thesis to the study of CHARGE syndrome. Ms. Hale's long-term career goal is to work with children and families affected by genetic disorders as a genetic counselor in the pediatric setting.

Ms. Schuette is a board-certified genetic counselor in the Department of Pediatrics and Communicable Diseases and Clinical Instructor of Human Genetics at the University of Michigan. Ms. Schuette works with children and families diagnosed with various genetic disorders, and serves on the executive faculty for the University of Michigan Genetic Counseling Training Program. She is co-editor of the first and second editions of A Guide to Genetic Counseling, the first textbook on the principles and practice of genetic counseling

Dr. Bielas is an Assistant Professor of Human Genetics at the University of Michigan. She is dedicated to the study of neurodevelopment and the pathogenic mechanisms of human neurodevelopmental disease genes. Her research is focused on understanding the cellular and molecular mechanisms of mammalian neurogenesis. Her laboratory aims to identify novel disease genes, and to use mammalian models of brain development to understand the pathophysiology of these genes.

Dr. Martin is an Associate Professor of Pediatrics and Human Genetics at the University of Michigan. In addition to working with children and families affected by numerous genetic disorders, including CHARGE syndrome, Dr. Martin devotes a significant amount of time to the study of CHD7 and related genes implicated in CHARGE pathogenesis. Her laboratory has developed several mouse models of Chd7 deficiency used to study CHARGE syndrome by investigators worldwide. Dr. Martin has published extensively on the role of CHD7 in inner ear and neural development. She is the Chair of the Scientific Advisory Board for the CHARGE Syndrome Foundation.

Presentation Abstract:

Since the discovery of CHD7 as the causative gene for CHARGE syndrome in 2004, the phenotypic spectrum associated with CHD7 mutations has greatly expanded to include individuals with CHD7 and mild or atypical clinical features that do not meet clinical diagnostic criteria for CHARGE. We will discuss ongoing genetic studies, CHD7 genotype-phenotype correlations, and results of a case review that prompted us to propose new CHARGE diagnostic criteria.

Atypical phenotypes associated with CHD7 mutations and a proposal for broadening CHARGE syndrome clinical diagnostic criteria

> Caitlin Hale CHARGE Syndrome Foundation Biennial Meeting 2015

Outline

- Review of CHARGE syndrome
- History of CHARGE syndrome as a clinical and molecular diagnosis
- Emerging spectrum of CHD7 associated phenotypes
- UM patient cohort data
- Literature review of atypical phenotypes
- Proposed diagnostic criteria
- Future directions

CHARGE syndrome

- Prevalence of 1 in 10,000
- <u>C</u>oloboma
- <u>H</u>eart defects
- Atresia of the choanae
- <u>R</u>etardation of growth
- <u>**G**</u>enital and/or urinary anomalies
- <u>Ear malformations, including</u> deafness and vestibular disorders





chargesyndrome.org

CHARGE as a clinical diagnosis

- 1981: described as a non-random association of features by Pagon et al. "CHARGE" acronym coined
- 1998: Blake et al. clinical criteria updated
- 2001: CHARGE recognized as a syndrome
- 2005: Verloes et al. clinical criteria updated
- Syndrome vs. an association
- Why are clinical diagnostic criteria important?

CHARGE as a clinical diagnosis

Blake et al. (1998)

Verloes (2005)

- Major · Coloboma
 - criteria · Atresia of choanae
 - Hypoplastic semicircular canals

- Coloboma
- Atresia of choanae
- Characteristic external ear anomaly
- Cranial nerve dysfunction

- Congenital cardiovascular malformation
- Tracheoesophageal defect
- Growth deficiency
- Genital hypoplasia or delayed pubertal development
- · Developmental delay
- Characteristic facial features
- Orofacial cleft

- Typical CHARGE: 4 majors
 - OR 3 majors+3 minors

- - criteria Rhomboencephalic dysfunction including
 - sensorineural deafness
 - Heart or esophagus malformation
 - Mental retardation
 - Hypothalamo-hypophyseal dysfunction

- Inclusion Typical CHARGE: 3 majors or 2 rule majors + 2 minors;
 - Partial CHARGE: 2 majors + 1 minor;
 - Atypical CHARGE: 2 majors but no minors, OR 1 major+2 minors

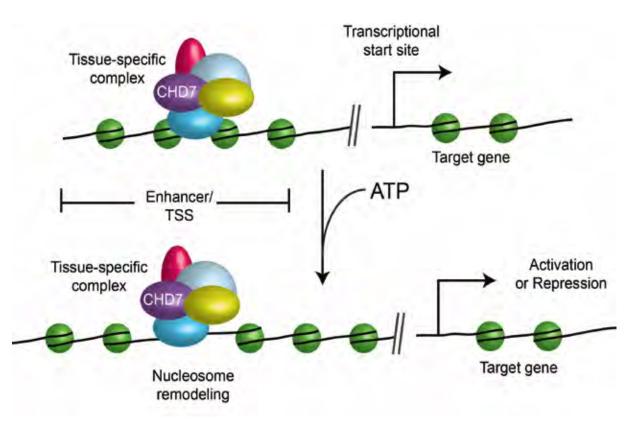
Palumbo et al. 2012. Gene.

CHARGE as a molecular diagnosis

- 2004: Vissers et al. identify 2.3 Mb overlapping microdeletion on 8q12 in 2 individuals with CHARGE
 - Follow-up sequencing of the region identifies mutations in CHD7 in 10/17
- Today: CHD7 mutations identified in 70-90% suspected cases of CHARGE
 - 2nd gene vs. non-coding variants vs. phenotypic overlap?

Function of the CHD7 Protein

- Member of the <u>Chromodomain</u> <u>Helicase</u> <u>D</u>NA binding family
- Forms protein complexes involved in chromatin remodeling
 - Tissue-specific regulation of gene expression



Expanding phenotype associated with CHD7 mutations

Clin Genet. 2009 Jan;75(1):65-71. doi: 10.1111/j.1399-0004.2008.01107.x. Epub 2008 Nov 17.

CHD7 mutations in patients initially diagnosed with Kallmann syndrome--the clinical overlap with CHARGE syndrome.

Jongmans MC¹, van Ravenswaaij-Arts CM, Pitteloud N, Ogata T, Sato N, Claahsen-van der Grinten HL, van der Donk K, Seminara S, Bergman JE, Brunner HG, Crowley WF Jr, Hoefsloot LH.

Proc Natl Acad Sci U S A. 2014 Dec 16;111(50):17953-8. doi: 10.1073/pnas.1417438111. Epub 2014 Dec 3.

Functionally compromised CHD7 alleles in patients with isolated GnRH deficiency.

Balasubramanian R¹, Choi JH², Francescatto L³, Willer J³, Horton ER⁴, Asimacopoulos EP⁵, Stankovic KM⁵, Plummer L⁴, Buck CL⁴, Quinton R⁶, Nebesio TD⁷, Merico V⁸, Merino PM⁸, Meyer BF⁹, Monies D⁹, Gusella JF¹⁰, Al Tassan N⁹, Katsanis N³, Crowley WF Jr¹¹.

Nature. 2012 Apr 4;485(7397):246-50. doi: 10.1038/nature10989.

Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations.

O'Roak BJ¹, Vives L, Girirajan S, Karakoc E, Krumm N, Coe BP, Levy R, Ko A, Lee C, Smith JD, Turner EH, Stanaway IB, Vernot B, Malig M, Baker C, Reilly B, Akey JM, Borenstein E, Rieder MJ, Nickerson DA, Bernier R, Shendure J, Eichler EE.

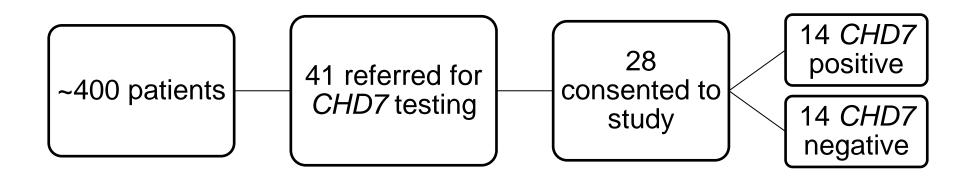
Am J Hum Genet. 2013 Aug 8;93(2):249-63. doi: 10.1016/j.ajhg.2013.06.012. Epub 2013 Jul 11.

Detection of clinically relevant genetic variants in autism spectrum disorder by whole-genome sequencing.

Jiang YH¹, Yuen RK, Jin X, Wang M, Chen N, Wu X, Ju J, Mei J, Shi Y, He M, Wang G, Liang J, Wang Z, Cao D, Carter MT, Chrysler C, Drmic IE, Howe JL, Lau L, Marshall CR, Merico D, Nalpathamkalam T, Thiruvahindrapuram B, Thompson A, Uddin M, Walker S, Luo J, Anagnostou E, Zwaigenbaum L, Ring RH, Wang J, Lajonchere C, Wang J, Shih A, Szatmari P, Yang H, Dawson G, Li Y, Scherer SW.

University of Michigan patient cohort

- Search of the University of Michigan Pediatric Genetics clinic scheduling database (8/2003-8/2014)
 - Terms: CHARGE, coloboma, choanal atresia, sensorineural hearing loss, hearing loss



Construction of a clinical database

- Added 28 patients who have been considered for a diagnosis of CHARGE and underwent CHD7 sequencing and del/dup
 - 14 CHD7 positive, 14 CHD7 negative

Construction of a clinical database

 Reviewed medical records and assessed for presence of 19 features associated with CHARGE syndrome

Typical vs. atypical CHARGE

- In our cohort, features most commonly associated with CHD7 mutation were inner ear anomaly (vestibular phenotype), external ear anomaly, hearing loss, cranial nerve dysfunction, and developmental delay
- 3/13 CHD7 mutation positive patients have atypical CHARGE due to lack of coloboma and choanal atresia
 - Is this distinction useful?
 - What is sufficient for a diagnosis of CHARGE?

Literature review

Literature search using terms: CHARGE, CHD7, CHARGE phenotype, atypical CHARGE, and CHD7 phenotype

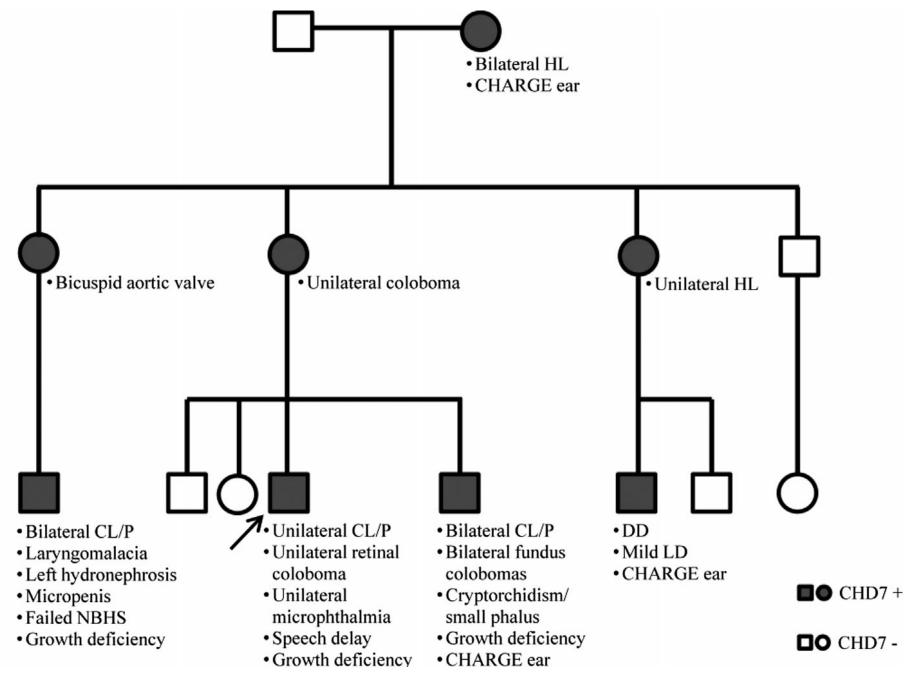
Identified 12 publications reporting on atypical phenotypes

Total of 31 *CHD7* mutation positive patients reported as having atypical CHARGE

Assessed if each patient met current diagnostic criteria and if they would meet our proposed criteria

Literature Review

- Identified 31 previously described cases of CHD7 mutation positive atypical presentations in large patient cohorts and unique case studies
 - Some were considered atypical due to lack of major features including coloboma and choanal atresia
 - Several reports of inherited CHD7 mutations
 with very mildly affected parents and relatives



Starling Hughes et al. 2014. Am J Med Genet Part A.

Motivation for changing diagnostic criteria

- CHARGE syndrome is a highly variable condition with a wide phenotypic spectrum
- Typical vs. atypical distinction not useful
- Better able to assess recurrence risk
- Inform medical management

Proposed diagnostic criteria

- We propose an update to the CHARGE diagnostic criteria to include CHD7 mutation status as a major feature
 - Allows individuals with milder phenotypes (including instances of inherited CHD7 mutations) to be considered as having CHARGE syndrome
 - Under new criteria, pathogenic CHD7 mutation + one major feature is sufficient for a diagnosis of CHARGE
- We propose broadening the description of supportive features associated with CHARGE to include skeletal/limb anomalies, brain anomalies, and renal anomalies

Future Directions

- Expansion of clinical database
 Search of EMR to identify more patients
- Whole exome/whole genome sequencing in *CHD7* negative patients

ACKNOWLEDGEMENTS

<u>Thesis Committee</u> Donna Martin, MD, PhD Stephanie Bielas, PhD Jane Schuette, MS, CGC

<u>Martin Lab</u> Ethan Sperry Jennifer Skidmore Diana Syam Hui Yao

<u>UM Pediatric Genetics Clinic</u> Bruce Skinner

<u>CHARGE Syndrome Foundation</u> Meg Hefner, MS, CGC <u>Funding</u> NIH R01 DC009410 Rackham Graduate School University of Michigan Genetic Counseling Program



Thursday, July 30, 2015 General Session: Utopia Ballroom 9:00-10:45am

Synaptic abnormalities in a Drosophila model of CHARGE syndrome

Daniel R. Marenda, PhD, Drexel University, Department of Biology

Presenter Information:

Dr. Marenda is an Associate Professor of Biology at Drexel University's Department of Biology and Drexel University College of Medicine's Department of Neurobiology and Anatomy. Dr. Marenda's laboratory focuses on understanding the developmental neurobiology of neural circuits using the fruit fly, *Drosophila melanogaster* as a model system. He has been working on CHARGE syndrome since 2009, and published the first *Drosophila* model of CHARGE syndrome in 2010. He sits on the Scientific Advisory Board for the CHARGE Syndrome Foundation.

Presentation Abstract:

The Drosophila neuromuscular junction (NMJ) is a glutamatergic synapse that is highly similar to mammalian glutamatergic synapses. Here we show that Kismet, the Drosophila homolog of CHD7, is important for synaptic morphology and transmission at this synapse, suggesting that Kis is part of the machinery that modulates the function of the NMJ. Our data also suggests novel avenues of investigation for potential synaptic defects associated with CHARGE syndrome.

Synaptic abnormalities in a Drosophila model of CHARGE syndrome

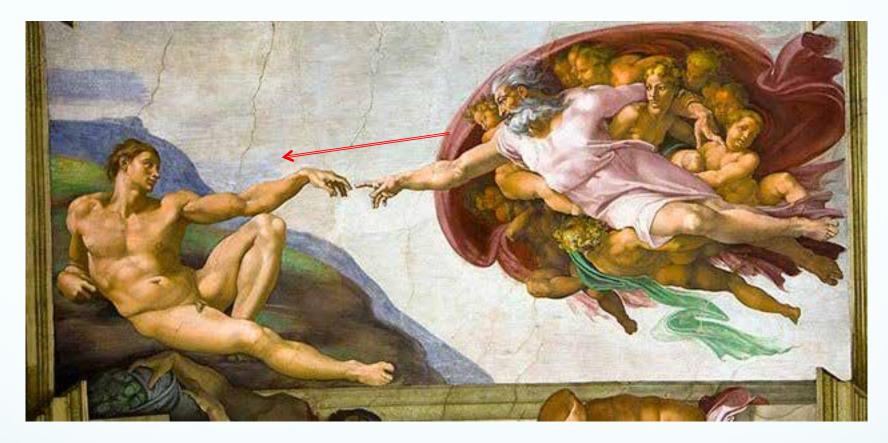
> Daniel R. Marenda, Ph.D. Associate Professor



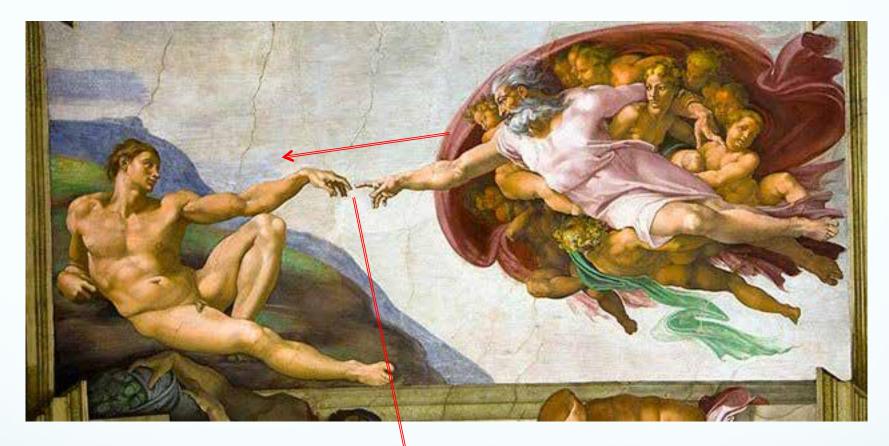


Dr. Faith Liebl, Southern Illinois University Edwardsville

Directional Information Flow



Directional Information Flow

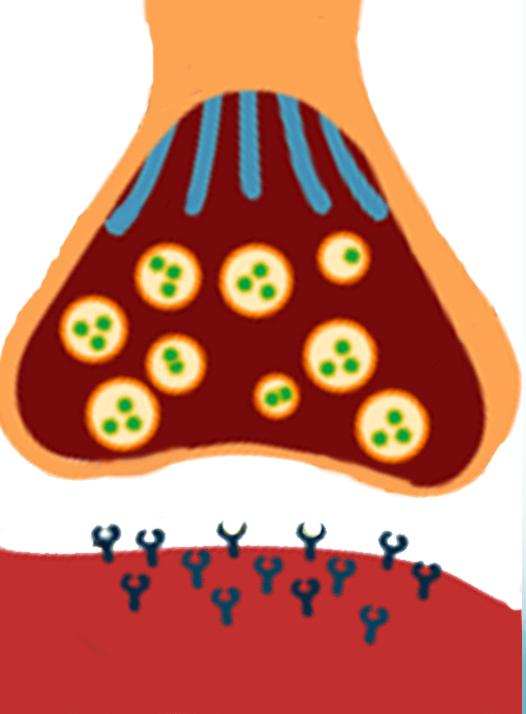


Synaptic cleft: Small space



Pre-synaptic Cell

Post-syanptic Cell



CHD7 is known to have important roles in the Central Nervous System in humans...

But little is known about the how loss of CHD7 function in mature neurons affects the ability of neurons to communicate...

For disorders affecting behavior, mature neurons are strong candidates for therapeutic intervention...

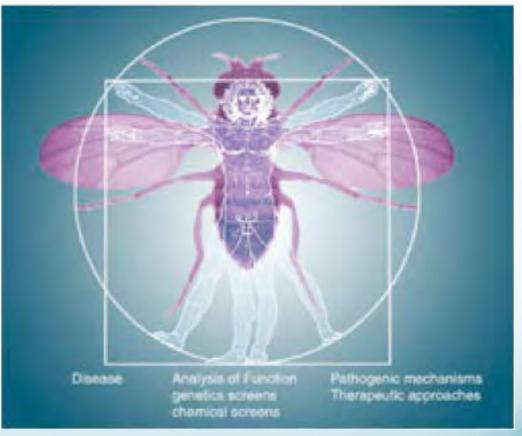
Drosophila: People, but smaller... and with wings

Little Fly Thy summers play, My thoughtless hand Has brush'd away.

Am not I A fly like thee? Or art not thou A man like me?

William Blake, 1794 (from *Songs of Experience*)

The expectation that discoveries made in the organism model will provide insight into the workings of other organisms.



Drosophila: People, but smaller... and with wings

Little Fly Thy summers play, My thoughtless hand Has brush'd away.

Am not I A fly like thee? Or art not thou A man like me?

William Blake, 1794 (from *Songs of Experience)*

Fully sequenced genome
Excellent molecular, cellular, genetic, behavioral assays readily available



Systems and development is well studied

CHD7 and Kismet: Chromatin Readers

SNF2 ATPase/Helicase SLIDE

Chromodomains

Kismet

CHD7

Chromodomains SNF2 ATPase/Helicase

BRK domain

BRK domain

SANT

Overall 51.3% identity and 64% similarity

- Chromodomains:
 - --Mediate protein-protein and protein-RNA interactions.
 - --Selective recognition of methylated histone tails (H3K4).

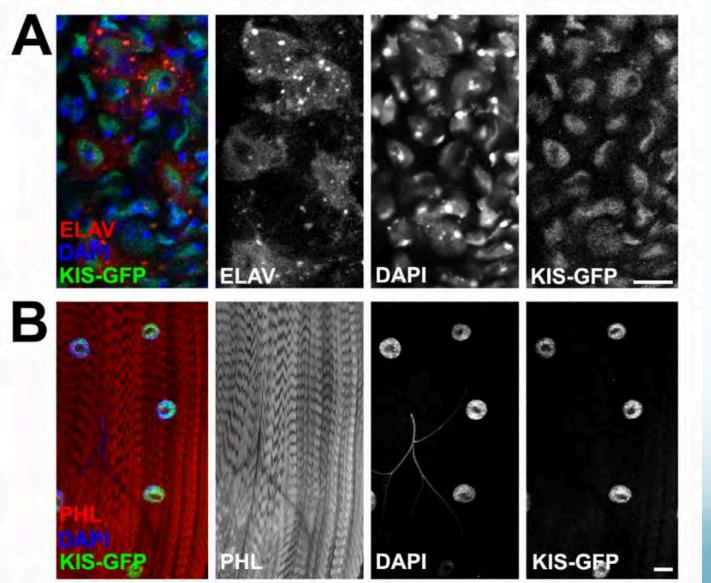
•SNF2 ATPase domain:

- --Associated with ATP-dependent chromatin remodeling activity.
- --50% identity between the ATPase domain of CHD7 and kismet.

•SANT-SLIDE domain:

--Interact with Histones and "slide" extranucleosomal DNA towards the histone octamer

Question: Is Kismet present in mature neurons at the fly NMJ? Ghosh et al., 2014



PRE

POST

Question: What happens if you knock down Kismet specifically in mature neurons?

Drosophila Larval Neuromuscular Junction (NMJ)

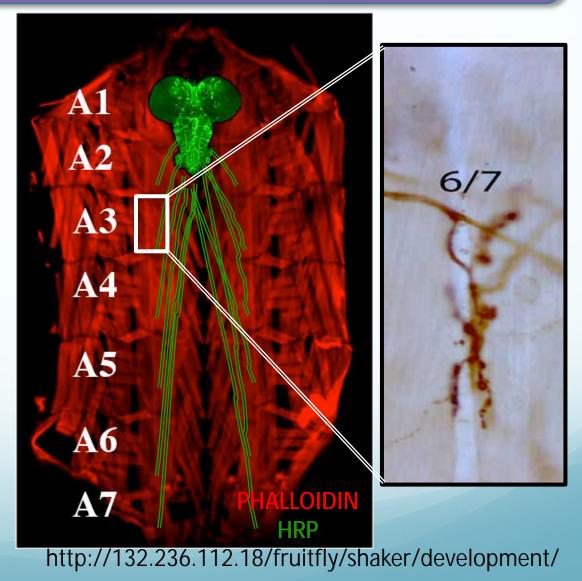
üPower of Drosophila Genetics

üWell studied system for synaptic structure and function

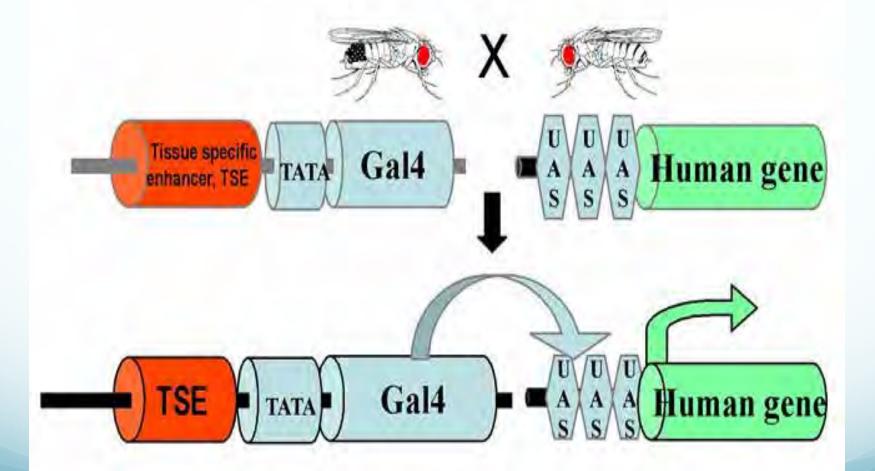
üGlutamatergic : Like CNS exitatory synapses in mammals with AMPA like GluR

ü Motor neuron innervation patterns: Stereotyped and repetitive

üExperimentally Accessible üBehavioral Studies üElectrophysiology üLive imaging

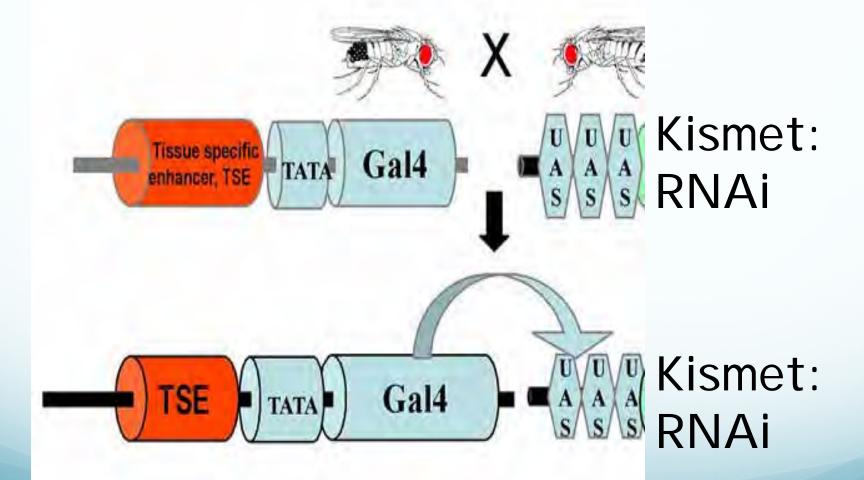


Gal4 / UAS System for temporal and tissue specific gene knockdown

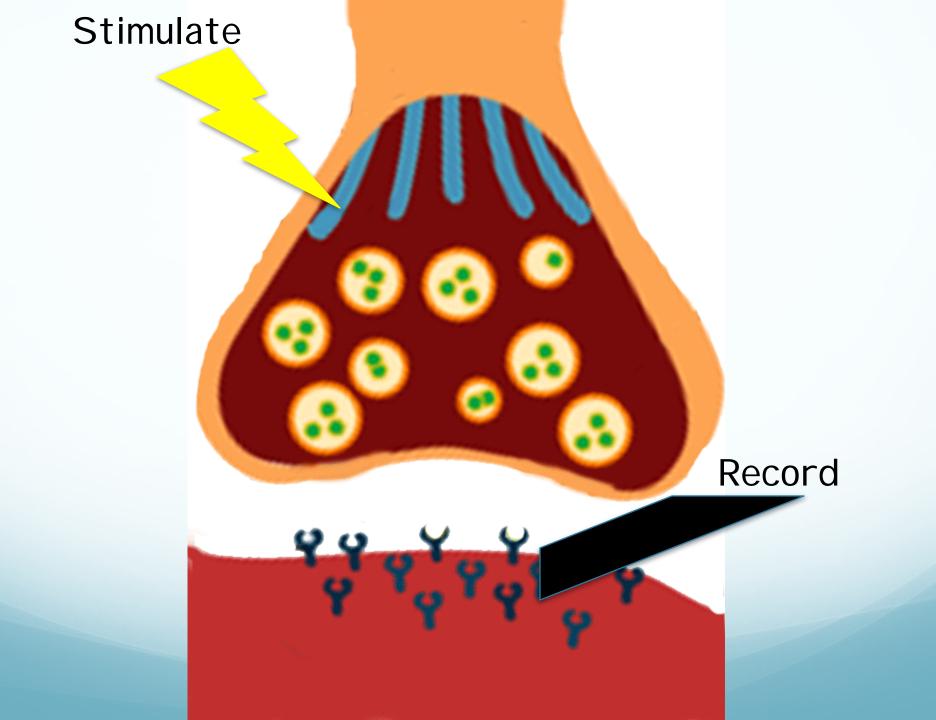


Taken from: Marsh, J. L. and L. M. Thompson (2006). "Drosophila in the study of neurodegenerative disease." *Neuron* 52(1): 169-78.

Gal4 / UAS System for temporal and tissue specific gene knockdown

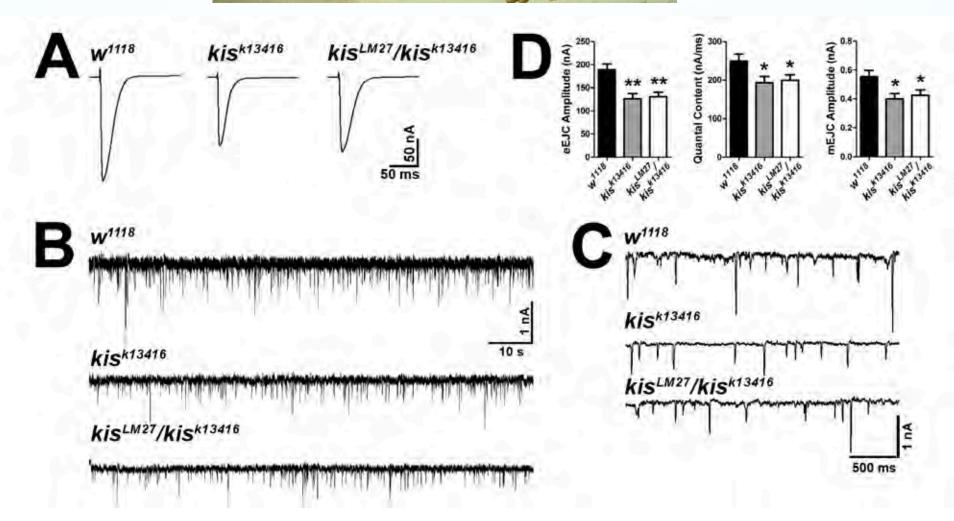


Taken from: Marsh, J. L. and L. M. Thompson (2006). "Drosophila in the study of neurodegenerative disease." *Neuron* 52(1): 169-78.



Directional Information Flow



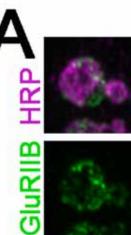


Decreased Kismet leads to decreased neurotransmission

Directional Information Flow: Post-synaptic Cell



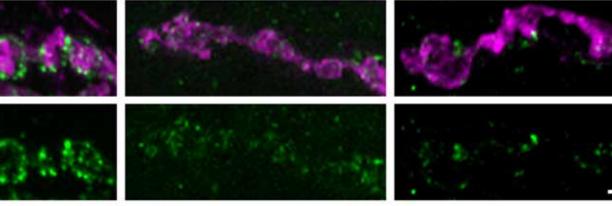


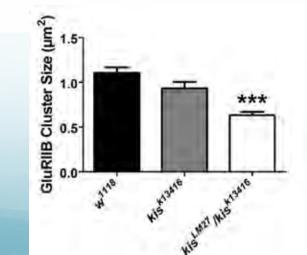


W¹¹¹⁸

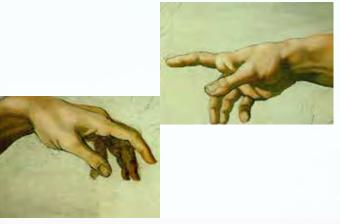
kis^{k13416}

kis^{LM27}/kis^{k13416}



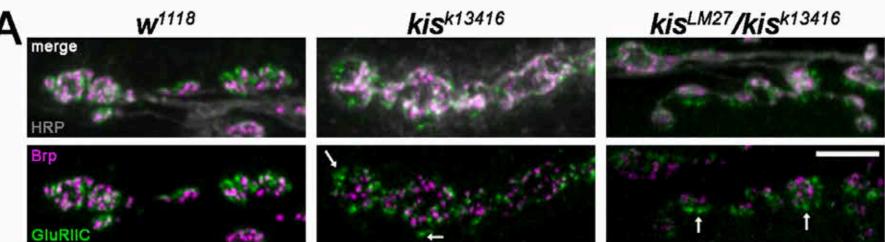


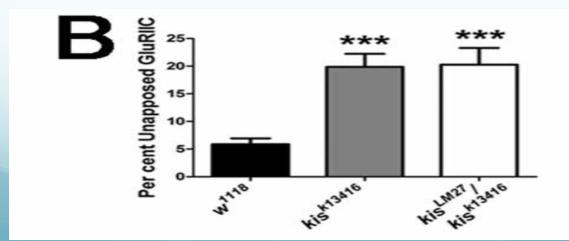
Directional Information Flow: Altered Alignment



kis^{k13416}





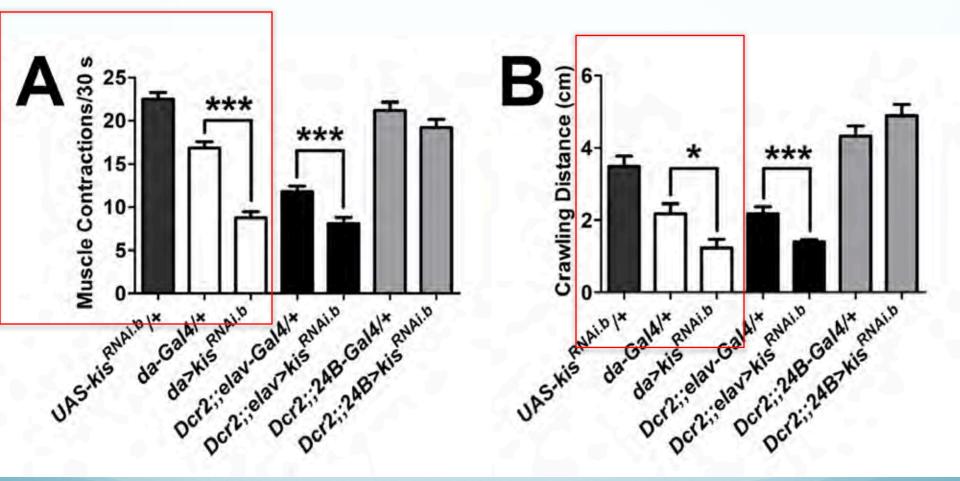


 Decreased Kismet leads to decreased neurotransmission

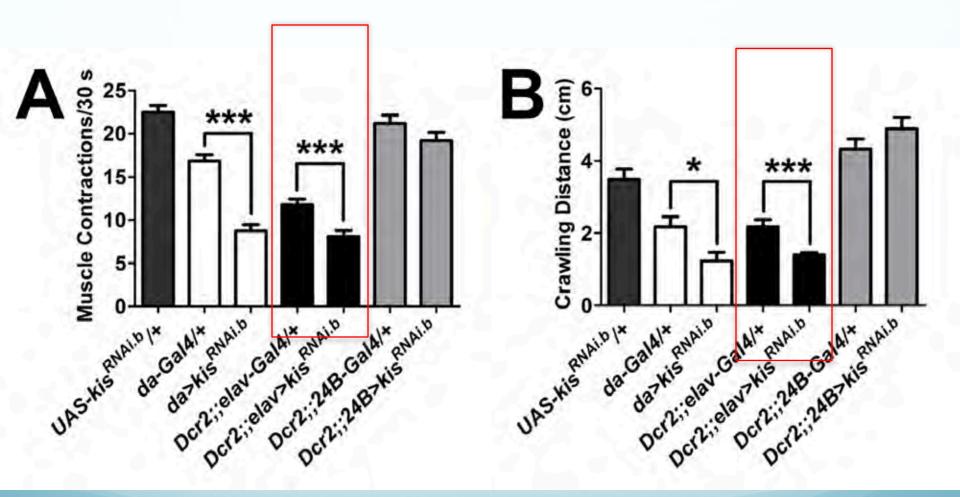
 Due to decreased glutamate receptor abundance in the postsynaptic cell, and altered apposition

 Question: Is the defect primarily pre-synaptic, post-synaptic, or both?

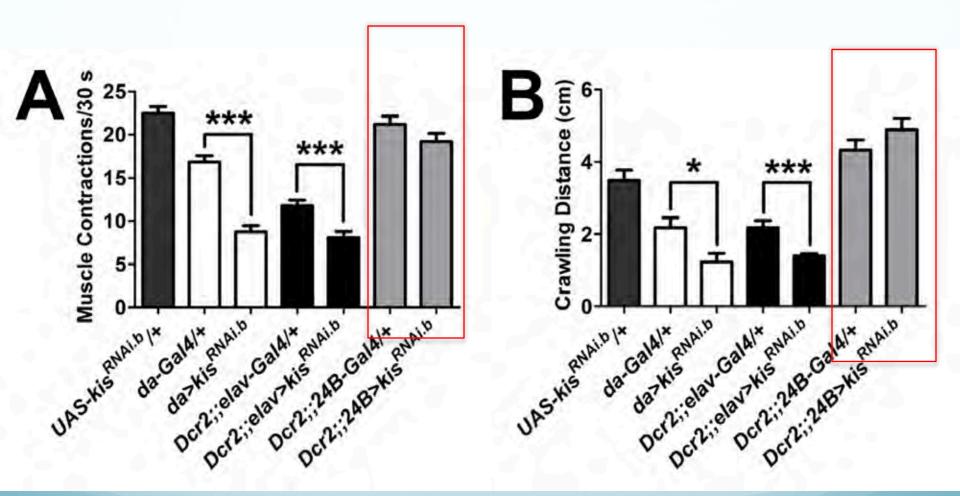




Pre- and Post- Knockdown



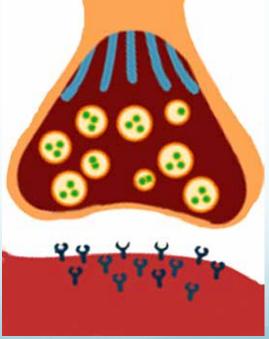
Pre- Knockdown

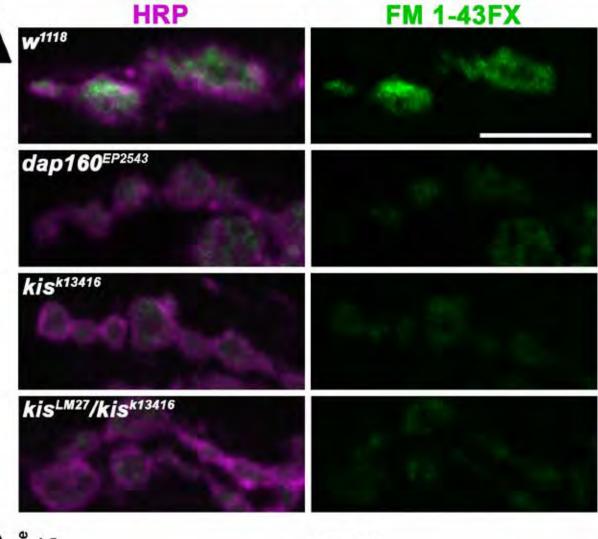


Post- Knockdown

Defect is mainly pre-synaptic.

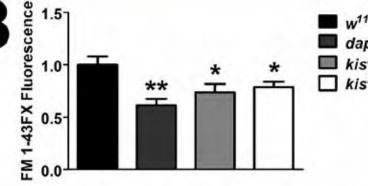
- Mechanisms of pre-synaptic machinery?

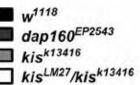




FM 1-43: Method of staining for exocytosis of membranes.

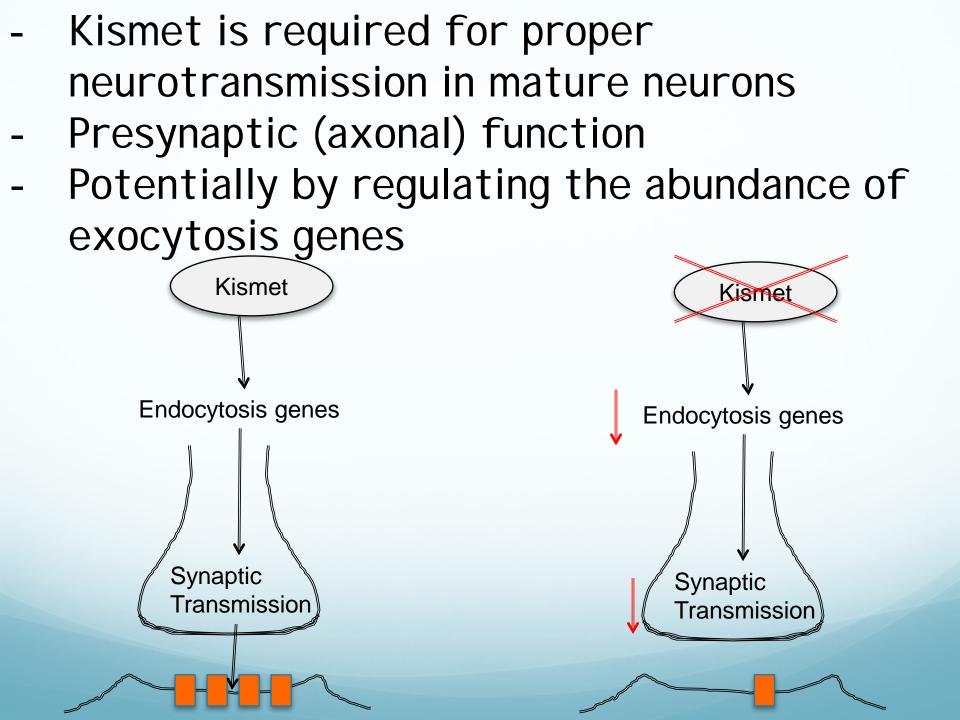


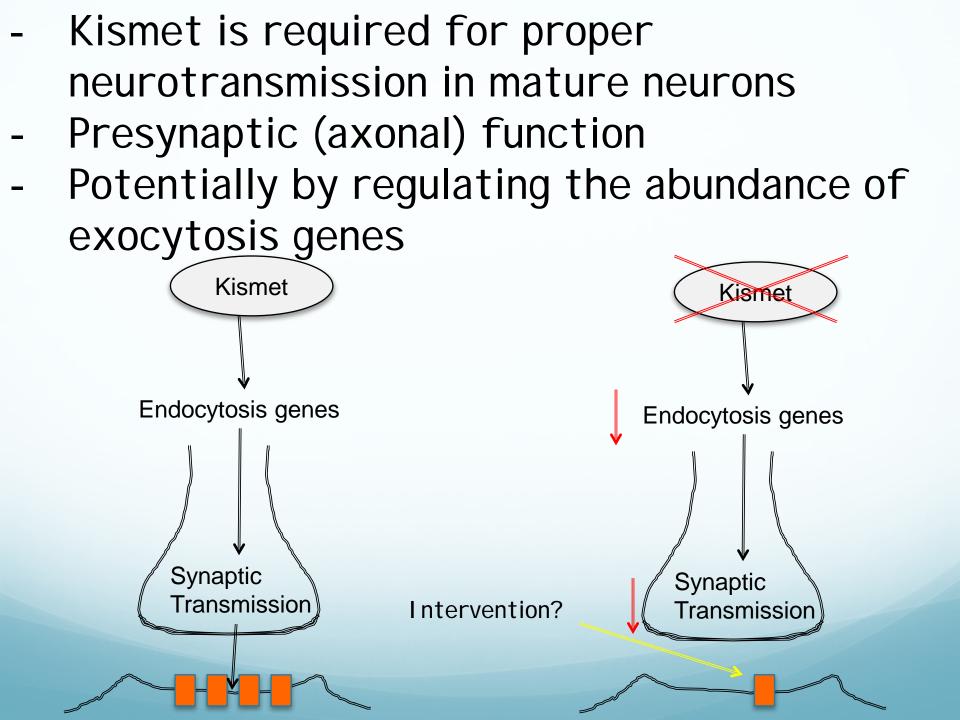




- Defect is mainly pre-synaptic.
- Defect is consistent with decreased exocytosis
- Hypothesis: Kismet regulated gene expression of exocytosis genes in mature neurons

- Microarray: Kismet in mature neurons
 - Decreased genes associated with exocytosis:
 - Synaptotagmin 7
 - SNAP 25
 - Rim
 - Neurexin 5





Nina Latcheva

- Edward Waddell
- Mitch D'Rozario
- Kaytie Innamorati
- Phuong Nguyen
- Allison Law
- Victoria Baccini
- Erin Robinson
- Sarah Ritter
- Rupa Ghosh
- David Melicharek







- Donna Martin
 - U. Michigan
 - Bing ZhangU. Oklahoma
- Faith Liebl
 - ISU-Edwardsville
- John Tamkun
 - UCSC
- Liqun Luo
 - Stanford
- Tzumin Lee
 - HHMI
- Kathy Siwicki
 - Swarthmore



National Center for Advancing Translational Sciences



Thursday, July 30, 2015 General Session: Utopia Ballroom 9:00-10:45am

Open-Hands, Open Access, Deaf-Blind Intervener Learning Modules: Using A National Resource to Meet State Specific Training Needs

Nancy Steele, Project Specialist for the National Center on Deaf-Blindness Michelle Clyne, M.Ed., Project Reach, Illinois Deaf-Blind Project, Jody Wolfe, Parent Leader, Chicago, IL, Amy T. Parker, Ed.D. & COMS

Presenter Information:

Nancy Steele is a Project Specialist for the National Center on Deaf-Blindness. She has worked for NCDB for 12 years. In this capacity, Nancy serves as the team lead on literacy, and supports the creation and adoption of the Open Hands, Open Access (OHOA) Deafblind Intervener Learning modules. Nancy has also hosted several field tests with parents, professionals and administrators who are reviewing the OHOA modules. Her background includes 17 years of classroom experience teaching Pre-K through grade 12 hearing impaired students as well as students with multiple disabilities in an array of settings. Her areas of expertise include accessing the general education, curriculum, and literacy.

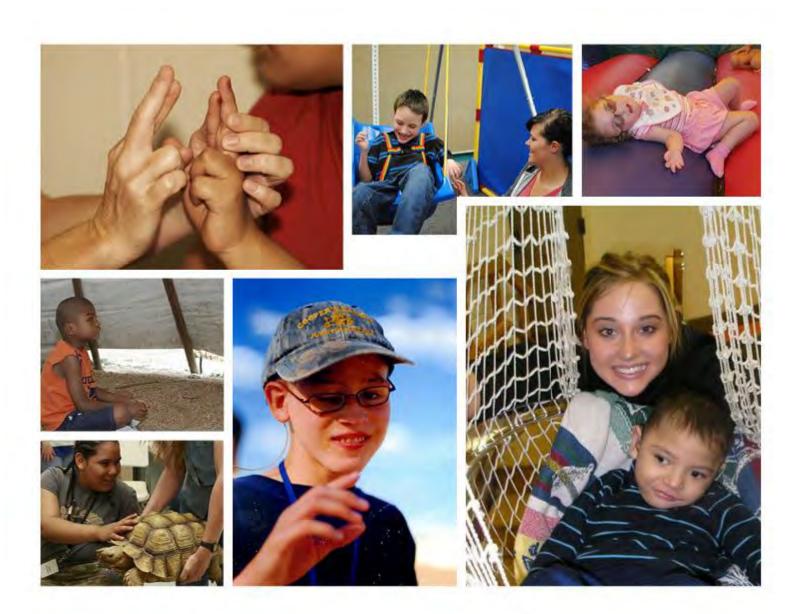
Presentation Abstract:

The Open Hands, Open Access Deaf-Blind Intervener Learning Modules are a national resource that has been created and refined by members of the national deafblind community. Parents have played a central role in their creation, field-testing and adoption. This presentation will describe the ways in which OHOA is being used within states to meet unique training needs. The team will also describe the dialogue with states that is leading to further improvement of the resource.



Open Access Modules as a Means of Reaching Diverse Adult Learners

Michelle Clyne, M. S. .Ed. Project Reach: Illinois Deaf-Blind Services Amy Parker, Ed.D. & COMS, Nancy Steele, M. A. Ed. National Center on Deaf-Blindness Jody Wolfe, Parent Leader, Director of Administration for the CHARGE Foundation 12th International CHARGE Syndrome Conference: July 31, 2015



Rationale from National Consortium on Deaf-Blindness Recommendations for Improving Intervener Services:

- Goal 2- Training & Support
- Establish a strong national foundation for intervener training and workplace supports.
- **Recommendation 3-** Develop a national open-access training resource that aligns with the CEC's Knowledge and Skills Competencies

Recommendations were created and published at the request of the Office of Special Education Programs.

A full copy of the data-based recommendations may be found at:

http://interveners.nationaldb.org/



What is an Intervener?

Interveners are specially trained personnel that provide vital support and access to individuals with deaf-blindness.

The National Center on Deaf-Blindness (NCDB) defines the educational role of Interveners thus:

"Interveners, through the provision of intervener services, provide access to information and communication and facilitate the development of social and emotional well-being for children who are deaf-blind. In educational environments, intervener services are provided by an individual, typically a paraeducator, who has received specialized training in deaf-blindness and the process of intervention. An intervener provides consistent one-to-one support to a student who is deaf-blind (age 3 through 21) throughout the instructional day" (NCDB, 2013).

OHOA Participatory Method of Creation:



- Advisory
 Committee
- Module Leads
- Module Contributors
- Field Participants
- Field Reviewers
- Expert External Advisor/Reviewers





Individuals who are deaf-blind were included as teachers in the modules.

In participatory methodologies, the purposeful inclusion of members of a community is, by design, a part of creating an authentic product and process.



Parent Leaders



- Partnered in module creation
- Participated in taking modules themselves
- Shared personal stories of intervener experiences







Accessibility Considerations

Use of a Moodle 2.23 Management System

Consultation with the Carroll Center for the Blind

Partnerships resulted in:

A more accessible JW Media player

Captioned and described video clips

Modules beginning to be translated into Spanish

Accessible text for all module materials

Consultation with DiCapta

Consultation with Described Captioned Media Program

Consultation with JKP Interpreting

Elements in Each Module:

Introduction- an opener to the main module themes

Inquiry Challenge- a practical problem



Learning Activities- Content and Assignments- sequenced learning path

Self-Assessments- could serve as a rubric for grading

Resources and References

September 2013, NCDB made OHOA Modules 1-4 available to state partners. IL was one of our first adopters.

OHOA modules offered by state partners in hosted conditions (re: specific purposes within the state).

OHOA modules also available (due to federal funding) to any learner that wants to review the 8 modules.

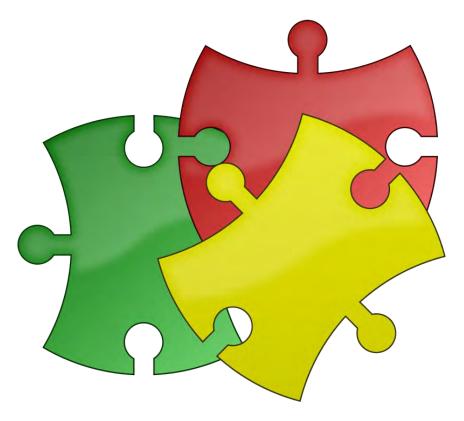




OHOA Online Modules in Illinois 2013 - Spring/Summer 2015

Prior to 2013

- Little awareness of intervener services in Illinois, especially at the administrative level.
- Deaf-Blind 101 was resource intensive.
- Distance activities were not standardized among teams.



Getting Intervener Services Before the Modules

- Educate School District about Interveners
- Discuss importance of intervener and get commitment from school
- Create Job Description
- Hire intervener
- Train intervener with assistance from family and deafblind project



Fall 2013, we started our first IL Cohort of Open Hands, Open Access Online Learning Modules.

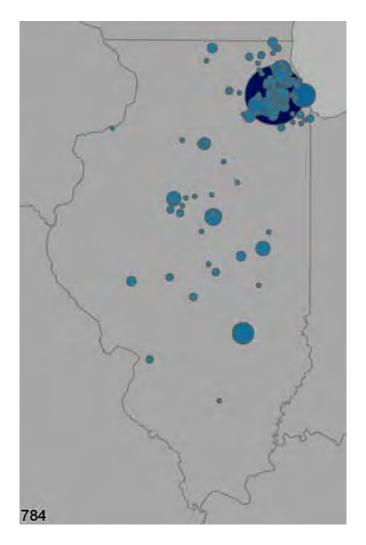


FREE ONLINE TRAINING IN DEAF-BLINDNESS And DEAF-BLIND INTERVENTION

Who registered?

- Special Ed Teachers
- Parents
- Paraeducators
- Related Service Providers: O&M, SLP
- Administrators
- Teachers of the Deaf/Hard-of-Hearing (TDHH)
- Teachers of the Visually Impaired (TVI)

People taking the modules have come from around Illinois, not always in the life of a student with deafblindness when taking the module. This is a map of access to the modules from January 1, 2014 until May 12, 2015.



PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES What did Project Reach provide?

- Dedicated hosts (Project staff, often person assigned to geographic region).
- Opportunity for follow through in classroom when applicable.
- Tech support (by Skype, phone, or in person if close).





What did participants think?

(5 point Likert scale- 5 = Strongly Agree)

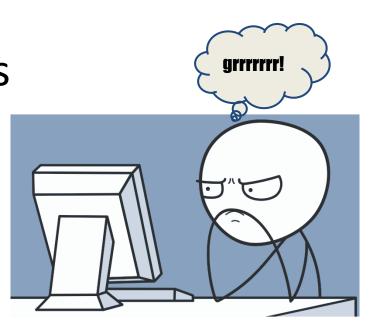
The presentation and materials were of high quality and professional: 4.83

The ideas and activities were relevant to the field of deaf-blindness: 4.87

I found the information useful: 4.85

Online learning challenges

- High-speed internet not available to all.
- Some schools block access to parts of the modules, especially YouTube.
- Online learning is not for everyone!



Advantages of Online Module Learning

- Potential for whole teams to access the same information and have a baseline of knowledge to build from.
- Conversations across and within disciplines: Teacher / Parent / Intervener/TVI; Parent to Parent.
- Online hosts who can provide encouragement and foster reflection, answer deeper content questions, suggest resources.

Advantages of OHOA

- Family perspective! Parents tell their child's story.
- Intervener/Para stories are also told, and professionals see how critical their full participation on the team is.
- Accessible media! Videos captioned, text accessible via screen reader. Access for Deaf, Blind/VI, Deaf-Blind participants is huge.

PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES

Outcomes

- Informal increased awareness (scheduled to take data Fall of 2015).
- Programs now discussing intervener services (sometimes for the first time, sometimes in new ways).
- Modules embedded in ongoing child specific technical assistance plans
- First Illinois Intervener Job Description

Back to the Big Picture! September, 2014- Present

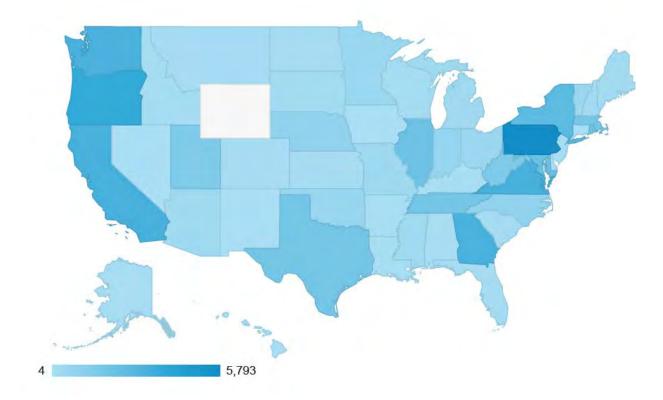
Users Active Daily: 32

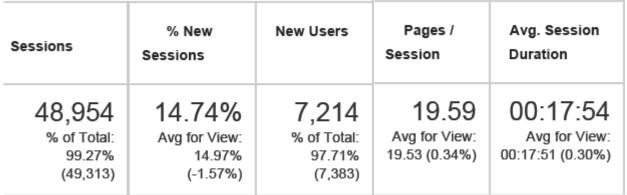
27 States + DC
32 State Collaborative Cohorts
5 Universities
24/37 completing all 8 modules.
All completing Welcome and Orientation
29 Host Training Sessions Completed

A Sample of Users- Demographics N= 593

Roles	% of Folks in the Modules	
Teachers	31.58%	
Intervener	12.06%	
Paraeducators	7.46%	
Other service providers	14.44%	
Parents	8.25%	
Interpreters	3.3 %	
Administrators	3.49 %	
Others	19.42% (TA providers, faculty, others)	

Jan 1, 2014 - May 11, 2015







Where have they logged on?

What do they think? (Sample size = 745 responses)

5 point Likert survey prompt-	% Strongly Agree or Agree (combined %)		
The learning outcomes for these modules were clear.	95.5%		
The content (readings, videos, etc.) focused on the learning outcomes.	97.9%		
The readings (articles, documents slides, etc.) were useful and interesting.	95.5%		
I learned new information in this module.	95.7%		
It was important for me to have this content.	94.8%		
Quizzes covered important information and concepts that was covered in the module.	93.1%		
I thought the content helped me achieve the learning outcomes.	97.4%		
I was able to navigate without problems.	81.3% 26		

Next Steps: Completing & Launching 26 Modules

8 OHOA modules are currently available

10 OHOA modules are in field testing and revision this year (2014-2015)

8 OHOA modules are being created by multidisciplinary teams this year and will be field tested next year (2015-2016)

- 1. An Overview of Deaf-Blindness and Instructional Strategies
- 2. The Sensory System, The Brain, and Learning
- 3. The Role of the Intervener in Educational Settings
- 4. Building Trusted Relationships and Positive Self-Image
- 5. Availability for Learning
- 6. Understanding Communication Principles
- 7. Emergent Communication
- 8. Progressing from Non-Symbolic to Symbolic Communication and Complex Language
- 9. Routines
- 10. Concept Development and Active Learning
- 11. Intervener Strategies
- 12. Maximizing Vision and Hearing
- 13. Calendars
- 14. An Introduction to Orientation and Mobility for Interveners
- 15. Orientation and Mobility in Everyday Routines
- 16. Self-Determination
- 17. Social Skills and Peer Relationships
- 18. Collaborative Teaming and Family Partnerships
- 19. Sexuality
- 20. AT & Access to Curriculum
- 21. Values, Ethics & Professionalism
- 22. Touch for Connecting and Learning
- 23. Behavioral and Environmental Supports
- 24. Transition to Adulthood and Community Living
- 25. Introduction to Sign Language and Braille
- 26. Putting it All Together

26 Modules cover the CEC's

Knowledge and Skills Competencies for Interveners

http://community.cec.sped. org/dvi/professionalstandard s

Resources

- Project Reach website: <u>http://www.</u> philiprockcenter.org/project-reach
- Module information: <u>http://www.philiprockcenter.</u> org/what-s-new
- NCDB website: <u>https://nationaldb.org</u>
- OHOA: https://nationaldb.org/ohoa
- OHOA contributors: <u>https://nationaldb.</u>
 <u>org/ohoamoodle/contributors.html</u>
- More about Interveners: <u>http://interveners.</u> <u>nationaldb.org/welcome.php</u>

OPEN HANDS, OPEN ACCESS: DEAF-BLIND INTERVENER LEARNING MODULES



WELCOME



Questions?

Michelle Clyne Project Reach: IL Deaf-Blind Services <u>mclyne@philiprockcenter.org</u> 630-790-2474

Jody Wolfe CHARGE Syndrome Foundation jody@chargesyndrome.org Nancy Steele National Center on Deaf-Blindness <u>nancy.steele@hknc.org</u>

Amy T. Parker National Center on Deaf-Blindness parkera@wou.edu



Thursday, July 30, 2015 General Session: Utopia Ballroom 9:00-10:45am

German CHARGE youth-week-ends "We have CHARGE – so what?!"

Claudia Junghans, 1st President of CHARGE Syndrom e.V. (Germany), Dr. Julia Benstz, Vice president of CHARGE Syndrom e.V. (Germany)

Presenter Information:

Claudia Junghans is the1st president of the German CHARGE Family Support Group and mom of a 10year-old son with CHARGE syndrome. She regularly lectures on CHARGE syndrome focusing on the feelings of a mother with an affected child. She is the Speaker of the German CHARGE Family support group.

Dr. Julia Benstz is the vice president of the German CHARGE Family Support Group and mom of a 15year-old daughter with CHARGE syndrome. She currently works as a hospital physician in neurology and is often involved in lecturing on medical issues.

Presentation Abstract:

The participants are young people (14+ years) with CHARGE Syndrome. The aim is to develop selfawareness and self-confidence of the participants within their peer-group. Special guided outdooractivities are routinely offered. Everything is embedded in this weekend: realizing their dream – making a film about their living with CHARGE syndrome and the way to setup relationships. The film is entitled "**We have CHARGE, so what?!**"



Thursday, July 30, 2015 Breakout Session #1: Euphoria Ballroom 1:00-2:30pm

Congenital arch vessel anomalies in CHARGE syndrome: a frequent feature with risk for co-morbidity

Nicole Corsten-Janssen, MD, Gideon J. du Marchie Sarvaas, MD²; Conny M.A. Van Ravenswaaij-Arts, MD PhD¹; Livia Kapusta, MD PhD ^{3,4}

¹ Department of Genetics ² Center for Congenital Heart Diseases, University Medical Center Groningen Groningen, the Netherlands. ³ Pediatric Cardiology, Dana-Dwek Children's Hospital Sourasky Medical Center, Tel Aviv University, Tel Aviv, Israel.⁴ Children's Heart Centre, Radboud University Nijmegen Medical Center, Nijmegen, the Netherlands.

Presenter Information:

Nicole Corsten-Janssen is a medical doctor who is in training to become a clinical geneticist. She is involved in CHARGE syndrome since 2009 when her PhD project on CHARGE syndrome, the CHD7 gene and heart defects started. Her supervisor is Conny van Ravenswaaij, whose group discovered the CHD7 gene in 2004.

Nicole has focused in her research on questions like which type of heart defects occur in CHARGE syndrome? Which patients can benefit from CHD7 analysis? What is the cause in someone with CHARGE (-like) syndrome if no CHD7 mutation is found? What syndromes overlap with CHARGE syndrome? What do we know about the CHD7 gene? She was involved in making the open access online database for CHD7 mutations (www.CHD7.org).

Presentation Abstract:

Arch vessel anomalies occur in a significant proportion of the patients with a CHD7 mutation. They may cause problems due to compression of the esophagus or airway. Since symptoms of vascular compression may mimic other problems in patients with CHARGE syndrome, it is important to be aware of arch vessel anomalies in this complex patient category.

Handout professionals day 2015

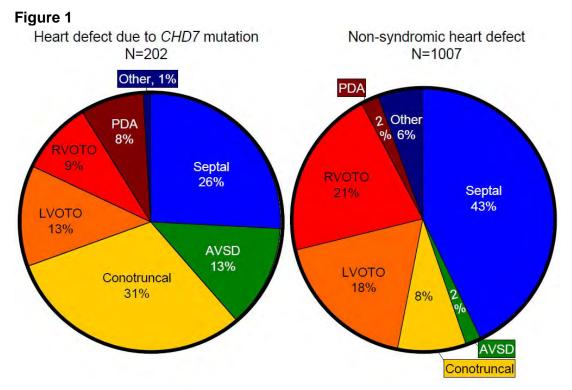
Congenital arch vessel anomalies in CHARGE syndrome: a frequent feature with risk for co-morbidity

Nicole Corsten-Janssen, MD

Clinical geneticist in training, University Medical Center Groningen, The Netherlands Contact: n.corsten@umcg.nl

Background

CHARGE syndrome is mainly caused by mutation in the CHD7 gene. We previously studied heart defects in 299 patients with CHARGE syndrome and a proven CHD7 mutation¹. We showed that heart defects occur in 220 (74%) of the patients with a CHD7 mutation, with overrepresentation of atrioventricular septal defects and conotruncal defects, including congenital arch vessel anomalies (figure 1). The story of a boy with CHARGE syndrome who suffered from periods of choking due to a symptomatic arch vessel anomaly for years, made us focus on arch vessel anomalies in our previous study cohort.



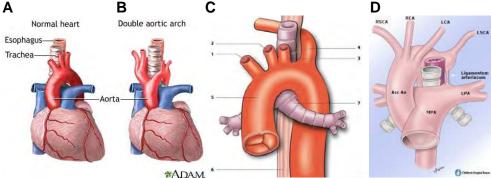
In these pie charts heart defects due to CHD7 mutations are compared to non syndromic heart defects. Conotruncal heart defects (yellow) and atrioventricular septal defects (AVSD, green) are overrepresented.¹

Conotruncal heart defects include: tetralogy of Fallot, double outflow right ventricle and arch vessels anomalies.

Methods:

We included patients from our previously studied cohort of 299 patients who had a vascular ring of any type, a right descending aortic arch (RAA), an aberrant subclavian artery, or an aberrant origin of an aortic arch vessel (figure 2). We studied cardiac phenotype and other symptoms in these patients.

Figure 2 arch vessel anomalies



A. A normal heart with a normal aortic arch and vessels in red

- B. An example of a vascular ring caused by a double aortic arch
- C. An aberrant subclavian artery
- D. Right- sided aortic arch

Results:

In our cohort 42 patients (14%) had an aortic arch anomaly, mostly aberrant subclavian artery or right aortic arch. The aortic arch anomalies usually occurred in combination with other congenital heart defects (81%). The other symptoms did not differ between patients with and without arch vessel anomalies. Most patients had feeding difficulty, but it is unclear if this was related to their arch vessel anomaly.

Conclusions:

Arch vessel anomalies are present in a significant portion of patients with a CHD7 mutation. In addition, due to the retrospective nature of our study we have probably underestimated the prevalence of arch vessel anomalies, because arch vessel anomalies are easily missed on cardiac ultrasound and our data are incomplete. This is an important observation because arch vessel anomalies can cause problems by compressing the esophagus and/or trachea. Also, identifying asymptomatic arch vessel anomalies is important before operations, because they are associated with a different anatomy of related structures.

Doctors caring for patients with CHARGE syndrome should be aware of arch vessel anomalies as possible cause for feeding or respiratory problems in this complex syndrome. However, future studies are needed to exactly identify the frequency of symptomatic arch vessel anomalies in CHARGE syndrome.

Reference

1. Corsten-Janssen N, Kerstjens-Frederikse WS, du Marchie Sarvaas GJ, Baardman ME, Bakker MK, et al. (2013) The Cardiac Phenotype in Patients with a CHD7 Mutation. Circ Cardiovasc Genet. 2013 Jun;6(3):248-54



Thursday, July 30, 2015 Breakout Session #1: Euphoria Ballroom 1:00-2:30pm

Clivus abnormalities, the 6th C in CHARGE syndrome?

Christa (C.M.) de Geus, MD, Jorieke E.H. Bergman MD PhD¹, professor Conny M.A. van Ravenswaaij-Arts MD PhD¹, Linda C. Meiners MD PhD²

Depts of ¹Genetics and ²Neuroradiology, University of Groningen, University Medical Center Groningen, Groningen, Netherlands

Presenter Information:

Christa de Geus is a medical doctor in training to become a clinical geneticist. Within clinical genetics she has a particular interest in neurology and neuroradiology. In 2014 she joined the research group of prof. Conny van Ravenswaaij as a PhD student. Her PhD focuses on neurological symptoms in CHARGE syndrome.

Presentation Abstract:

During a structured re-evaluation of cerebral MRI scans of patients with CHARGE syndrome, we found highly typical abnormalities of the clivus, a part of the skull base. We hypothesized that these abnormalities could aid diagnosis and wondered whether they might be clinically relevant. We found that more than 90% of CHARGE patients have an abnormal clivus, but no patients exhibited clinical symptoms related to this.



"The clivus, the sixth 'C' in CHARGE syndrome?"

About the presenter



My name is Christa de Geus. I am a clinical geneticist in training and a PhD student in the group of Prof. Conny van Ravenswaaij-Arts at the department of Genetics of the University Medical Center Groningen, in the Netherlands. My main research project is on neurological abnormalities in CHARGE syndrome.

Introduction

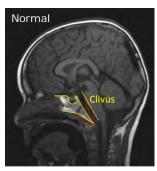
CHARGE syndrome has a number of specific neuroradiological features that aid diagnosis: hypo- or aplasia of the olfactory bulbs and specific dysplasia of the semicircular canals. In this presentation, we will show that clivus abnormalities can be added to this list of features.

How many C's?

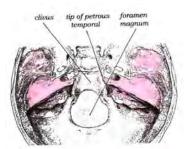
The 'four C's' of CHARGE syndrome are a well known CHARGE mnemonic. They stand for <u>c</u>hoanal atresia, <u>c</u>oloboma of the eyes, <u>c</u>haracteristic ear shape and <u>c</u>ranial nerve deficits. Of course, these are not the only symptoms typical for CHARGE syndrome. In fact, we have reason to add two more 'C's': the cerebellum and clivus. In this presentation I will focus on the clivus in CHARGE syndrome. The cerebellum in CHARGE will be discussed by prof. Van Ravenswaaij in another session.

The clivus

The clivus is a part of the basiocciput. The upper portion of the clivus is formed by (part of) the body of the sphenoid and is joined to the basilar occipital bone to form the complete clivus (figures below).



Midline sagittal view



Superior view of the skull base, Gray's anatomy

What is the relevance of clivus abnormalities?

Fujita et al. described CHARGE patients with an abnormally small clivus or platybasia. A small or malformed clivus results in an altered anatomy of the skull

base called basiocciput hypoplasia. It may also result in platybasia, a flattened skull base. Both basiocciput hypoplasia and platybasia are known to be associated with basilar invagination, which may lead to compression of the medulla. One of Fujita's patients had a Chiari 1 malformation with syringomelia, leading to neurological complaints.

In fact, in our clinic, a neonate with a congenital heart defect and dysmorphic features was found to have an abnormal clivus at a postmortem MRI. Recognition of this finding as typical for CHARGE syndrome directly led to his diagnosis, illustrating the value of this finding.

To study the nature, prevalence and clinical relevance of clivus abnormalities in CHARGE syndrome, we conducted a retrospective analysis of our CHARGE cohort.

Methods

<u>Patients and controls:</u> We included 23 *CHD7*-positive patients for whom MR imaging of sufficient quality was available. We included 72 age-matched controls. Radiological analysis of the clivus:

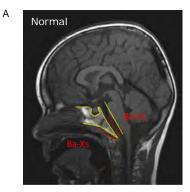
For every patient and control, we collected the following data:

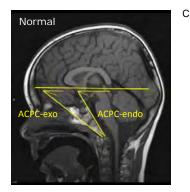
В

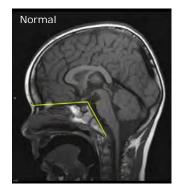
- Clivus:
 - size (Ba-Xs and Ba-Es, see figure A, below)
 - 'slope' of the clivus, as measured by the clivus-ACPC angle (ACPC-exo and -endo, see B) and the Welcker angle (C)
 - o morphology
- Presence of basiocciput hypoplasia, basilar invagination and Chiari I malformation

For every CHARGE patient:

 Clinical characteristics: symptoms, satisfaction of clinical criteria of Blake and Verloes





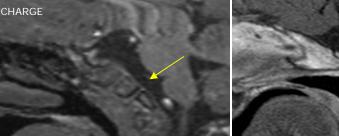


Results

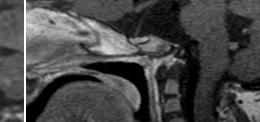
- Size/morphology: 91% had an abnormal clivus
 - o 59% of patients had a small (<2 SD below age-matched controls) clivus

- In 87% the clivus had abnormal morphology, such as extra synchondroses, loss of triangular shape or grossly abnormal anatomy of the skull base.
- Platybasia in 39% .
- Basilar invagination in 26%, with possible compression of the brainstem in two patients. These patients do not have a history of neurologic complaints
- We found no correlation between size of clivus and clinical parameters •





Extra synchondrosis



Nearly unrecognisable anatomy

Small, malformed clivus

Conclusions

This study affirms and expands on the data provided by Fujita et al., showing that a malformed and small clivus is commonly part of CHARGE syndrome. Although a guarter of our patients had basilar invagination, only two (less than 10%) had possible involvement of the brainstem. In these two patients, there was no history of neurological complaints. In addition, none of our patients had a Chiari I malformation. The clinical relevance of clivus abnormalities in CHARGE syndrome is therefore unclear. As we have seen in our centre, however, clivus malformations in CHARGE syndromes are of diagnostic value.

In conclusion, clival abnormalities can be added to semicircular canal dysplasia and olfactory bulb aplasia to form a triad of neuroradiological 'CHARGE findings'. We hope that familiarity with this triad will enable timely diagnosis in CHARGE patients.

Acknowledgements

The following people were involved in conducting the study:

- Prof. Conny van Ravenswaaij-Arts, clinical geneticist, UMCG
- Dr. Linda Meiners, pediatric neuroradiologist UMCG
- Dr. Jorieke van Kammen-Bergman, clinical geneticist, UMCG

Contact information

For further information or questions, please do not hesitate to contact me. You can send an e-mail to: charge@umcg.nl

References

1. Fujita K, Aida N, Asakura Y, et al. Abnormal basiocciput development in CHARGE syndrome. *AJNR Am J Neuroradiol*. 2009;30(3):629-634.



Thursday, July 30, 2015 Breakout Session #1: Euphoria Ballroom 1:00-2:30pm

Cerebellum abnormalities, the 5th C in CHARGE syndrome

Conny van Ravenswaaij-Arts, MD, PhD, Tian Yu¹, Linda C Meiners², Katrin Danielsen¹, Monica TY Wong², Timothy Bowler³, Danny Reinberg⁴, Peter J Scambler⁵, M Albert Basson¹

¹King's College London, United Kingdom, ²University Medical Center Groningen, Netherlands, ³Montefiore Medical Center, New York, United States, ⁴New York University School of Medicine, United States, ⁵University College London, United Kingdom

Presenter Information:

Conny van Ravenswaaij is a clinical geneticist with a long-lasting experience in genetic and clinical studies on CHARGE syndrome. Since her group discovered CHD7, the gene responsible for CHARGE syndrome, in 2004, she coordinates a multidisciplinary clinic and supervised several PhD research projects dedicated to the syndrome. Her research is mostly based on questions raised by the parents and often results in clinical guidelines. Thus far she has published over 25 papers and four book chapters on CHARGE syndrome.

Presentation Abstract:

CHARGE syndrome is a highly variable condition affecting multiple organ systems including the central nervous system. The exact effects on brain development are not known yet and the reasons for the huge clinical variability are not well understood. The responsible gene, CHD7, is known to control the expression of other genes during embryonic development, but the molecular mechanisms which lead to the neural defects found in CHARGE syndrome are unclear. In this study we focused on the role of Chd7 in mouse cerebellar development and we critically evaluated MRI scans of children with CHARGE syndrome for cerebellar defects.



Cerebellum abnormalities, the 5th C in CHARGE syndrome

Conny van Ravenswaaij-Arts

About the presenter

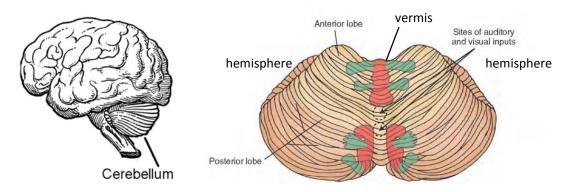


I am a clinical geneticist with a long-lasting interest in CHARGE syndrome. Since we discovered CHD7, the gene responsible for CHARGE syndrome, in 2004, I coordinate a multidisciplinary expert clinic and supervise PhD research projects dedicated to CHARGE. My research is mostly based on questions raised by parents. Thus far we published over 25 papers and four book chapters on CHARGE syndrome.

Introduction

CHARGE syndrome is a highly variable condition affecting multiple organ systems including the central nervous system. The exact effects on brain development are not known yet and the reasons for the huge clinical variability are not well understood. The responsible gene, CHD7, is known to control the expression of other genes during embryonic development, but the molecular mechanisms which lead to the neural defects found in CHARGE syndrome are unclear. It is also not known what the clinical effects of these neural defects are. In this study we critically evaluated MRI scans of children with CHARGE syndrome for cerebellar defects after such defects were noted in a mouse model of CHARGE syndrome by the group of Albert Basson [1].

Subsequently we initiated a study to evaluate the clinical effects of the anatomical cerebellar abnormalities that we detected. The cerebellum plays an important role in coordination of movements and balance. Balance involves the complex task of integrating postural responses and multisensory (visual, labyrinthine from the semi-circular canals, and proprioceptive) feedback. The human cerebellum,



consists of two hemispheres and a central vermis. By several interconnecting loops with the cortex, thalamus, basal ganglia and limbic system, the cerebellum integrates motor- and multisensory signals for fine-tuning of the motor input. Coordination of the limbs is regulated by the cerebellar hemispheres, whereas the vermis is involved in

axial stability during stance and gait. In this perspective, it is conceivable that developmental abnormalities of the vermis may have an impact on balance in CHARGE patients.

This study is important, because cerebellar dysfunction may be easily overlooked in children with CHARGE syndrome. These children often have motor coordination problems due to abnormalities of their balance organ, the semicircular canals. The cerebellum plays an important role in coordination of movements, gait and posture. Recognition of cerebellar dysfunction has implications for motor development and thus for the therapeutic support, e.g. by physiotherapy, of the children.

Methods

Studies in the mouse model

Chd7 and Fgf8 loss-of-function mouse models were used. Fgf8 is also known to be involved in cerebellar development and very likely to act in the same pathway as Chd7. The brains of live-born mice were studied for anatomical abnormalities and for the expression of genes.

Retrospective studies in the children with CHARGE syndrome

The MRI scans of 20 patients with CHARGE syndrome were evaluated by an experienced neuroradiologist using a standard protocol and MRIs of age-matched controls. FGF8 was analyzed for variants in the same group of children.

Prospective studies in the children with CHARGE syndrome

Balance and coordination is quantified by the Berg Balance Scale [2] and the Scale for the Assessment and Rating of Ataxia (SARA)[3]. These tests can be assessed reliably in children of 4 years and older.

Results

Cerebellum in the mouse model

Loss of Chd7 in mice results in decreased Fgf8 expression in the embryonic signaling center that directs cerebellar development. Combined loss of Chd7 and Fgf8 results in abnormal cerebellum development in mice.

Cerebellum in patients with CHARGE syndrome

In patients with CHARGE syndrome due to a proven CHD7 mutation, we found cerebellum abnormalities in 55%. The presence or absence of cerebellum abnormalities could not be explained by the type of CHD7 mutation or by additional variants in FGF8.

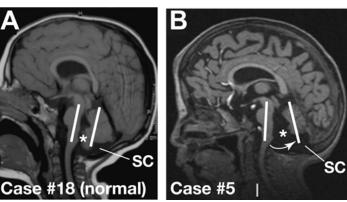


Figure (A) Sagittal T1 scan of patient #18 showing a normal vermis with a normal position, foramen of Magendi (asterisk) and subcerebellar cistern (SC). The orientation of the cerebellum relative to the brainstem is indicated by two parallel white lines. (**B**) Sagittal T1 scan of patient #5 showing pronounced vermis hypoplasia with an anticlockwise rotated axis relative to the axis of the

brainstem (arrow), and ensuing large foramen of Magendi (asterisk) and subcerebellar cistern (SC). [1]

Cerebellar dysfunction in CHARGE syndrome

These studies are still ongoing and I will demonstrate with short movies how the clinical tests are performed and summarize what we have seen thus far.

Conclusion

CHD7 plays a role in cerebellum development. Cerebellum defects are a clinical feature in CHARGE syndrome, being the 5th C after coloboma, choanal atresia, cranial nerve defects and cardiac abnormalities. The involvement of the cerebellum may have important implications for amongst others posture and gait.

References

- 1. Yu T, Meiners LC, Danielsen K, et al. Deregulated FGF and homeotic gene expression underlies cerebellar vermis hypoplasia in CHARGE syndrome. Elife 2013;2.
- Franjoine MR, Gunther JS, Taylor MJ. Pediatric balance scale: A modified version of the berg balance scale for the school-age child with mild to moderate motor impairment. Pediatric Physical Therapy 2003;15:114-28.
- 3. Brandsma R, Spits AH, Kuiper MJ, et al. Ataxia rating scales are age-dependent in healthy children. LID 10.1111/dmcn.12369 [doi]. Dev Med Child Neurol 2014.

Acknowledgements

The following people were/are involved in conducting the study:

- Christa de Geus, MD PhD student, UMCG
- Dr. Deborah Sival, pediatric neurologist, UMCG
- Dr. Linda Meiners, pediatric neuroradiologist, UMCG
- Dr. Albert Basson, King's College London
- Tian Yu, King's College London
- Prof. Conny van Ravenswaaij-Arts, clinical geneticist, UMCG

The study is supported by grants provided by the International CHARGE Foundation and the University Medical Centre Groningen (personal grant CdG).

Contact information

For further information or questions, please do not hesitate to contact us. You can send an e-mail to: <u>charge@umcq.nl</u>

12th International CHARGE Conference, Chicago 2015



Thursday, July 30, 2015 Breakout Session #1: Euphoria Ballroom 1:00-2:00pm

The Development of a Comprehensive Checklist Guiding Health Screening and Management Considerations of Individuals with CHARGE Syndrome: a Delphi Technique

Carrie-Lee Trider, MD (Pediatric Resident, PGY 2), Queen's University, Angela Arra-Robar, RN MSN, Clinical Nurse Specialist, IWK Health Center, Kim Blake, MD, MSc, FRCPC, Professor of Pediatrics, Dalhousie University, IWK Health Center

Presenter Information:

Carrie-Lee Trider is a Pediatric Resident at Kingston General Hospital, Queen's University, Ontario, Canada. She is a previous student of Dr. Kim Blake's. Meeting many wonderful children with CHARGE Syndrome at a prior conference has inspired her to continue with a research interest in CHARGE Syndrome.

Carrie-Lee is collaborating with Angela Arra-Robar, Clinical Nurse Specialist and Dr. Kim Blake for this presentation. Angela is a Registered Nurse who works with children and youth who have complex medical issues and who may be described as medically fragile. Angela has worked in pediatrics for over 18 years in both the United States and Canada and currently works with Dr. Blake at the IWK Health Centre in Halifax, Nova Scotia and coordinates a special multidisciplinary clinic for CHARGE families. 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.

Presentation Abstract:

There is a wide spectrum of medical, physical and psychological diagnoses in individuals with CHARGE Syndrome. No simple guidelines for an approach to screening and management of clinical problems have previously been published for CHARGE Syndrome. Therefore, we aimed to develop a comprehensive guiding checklist addressing these issues for CHARGE Syndrome across the lifespan: from head to toe. This checklist was evaluated qualitatively using the Delphi method to develop a final consensus. We will present and encourage discussion of these results.



Thursday, July 30, 2015 Breakout Session #2: Euphoria Ballroom 2:45-4:15pm

Pain and Coping: Identifying and Easing Pain

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

Presenter Information:

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She currently runs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome*, the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Stratton specializes in challenging behavior and improving adaptive skills.

Presentation Abstract:

Understanding when and where a child with CHARGE experiences pain can be perplexing due to limited communication challenges. Is it pain or behavior? This presentation is designed to assist with the difficult job of identifying pain non-vocally. Administration of a non-vocal pain measure will be reviewed, but identifying pain is only the first step. Next, participants will be introduced to various pain coping techniques designed to reduce pain and prepare individuals with CHARGE for future pain (i.e. surgery, immunizations, etc.)

PAIN AND COPING: IDENTIFYING & EASING PAIN

Presentation for the: 12th International CHARGE Syndrome Conference Chicago 2015 Kasee Stratton, Ph.D., NCSP

Thank you!

"The Fifth Vital Sign"

- Body Temperature
- Blood Pressure
- Pulse
- Breathing



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

Why is it difficult to measure pain in CHARGE?

Limited or no communication strategies

"Behavioral expressions of pain are merely a nonverbal form of self report."

(Bodfish, Harper, Deacon, and Symons, 2001)

- Facial Reactions and Palsy
- Possible social-communicative deficits (Craig, 2006)
- Possible social referencing deficit (Recchia, 1997)

Impact of Pain

Negative impact on neurological development

Untreated/poorly treated pain in infancy and early childhood reported lower pain thresholds later

Ruda et al. 2000; Blount et al. 2006, Brewer et al., 2006; Howard, 2003

Heightened levels of distress and anxiety

Ruda et al. 2000; Blount et al. 2006, Brewer et al., 2006; Howard, 2003

Impact on 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

Impact on Behavior

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

(Oberlander & Symons, 2006)

PAIN EXPERIENCES FOR CHARGE

Pain Experience

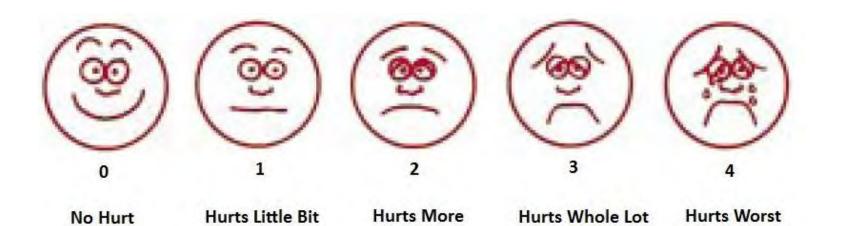
- Acute
- Chronic
- Surgeries/Hospitalizati ons
- Medical Procedures
- Therapies

- CHARGE
 Characteristics
- Cranial Nerves Migraines & Headaches
- Sleep Disturbances

Chronic Pain

	Days per Year in Pain	
Characteristic	M	SD
Surgery Pain	9.52	9.40
Jaw Discomfort	13.22	11.17
Migraine	13.50	13.51
Chronic Recurrent Otitis Media	22.88	32.18
Sinusitis	35.13	41.51
Constipation	52.25	58.38
Coughing	66.48	99.42
Muscle Pain	95.70	136.07
Abdominal Migraine	97.47	128.95
Hip/Back Pain	98.09	144.14
Breathing	108.67	131.82
Difficulty Swallowing	129.00	154.04
Gastroesophageal Reflux	169.29	133.70

Pain Intensity



Pain Intensity

	Pain Intensity			
Characteristic	M	SD	Range	
Migraine	2.67	.87	2-4	
Abdominal Migraine	2.45	1.10	1-4	
Constipation	2.38	.80	1-4	
Surgery Pain	2.34	.97	1-4	
Chronic Recurrent Otitis Media	2.24	.99	0-4	
Sinusitis	2.17	.82	1-4	
Gastroesophageal Reflux	2.06	1.14	0-4	
Breathing	2.00	1.03	1-4	
Hip/Back Pain	1.86	.95	1-4	
Muscle Pain	1.82	.87	1-3	
Coughing	1.61	.80	1-3	
Jaw Discomfort	1.56	.88	1-3	
Difficulty Swallowing	1.50	.83	1-4	

Surgeries & Medical

- Surgery
 - 1 to 63 procedures
 - Average 13

Study 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

IDENTIFYING & MEASURING PAIN

DIRECTIONS:

Please complete the following rating after observations of your child for one day when you believe your child is experiencing pain. For each item, circle the number that best describes your child's behavior during the pain episode.

If your child does not engage in a behavior when not in pain OR is not capable of performing an action, score this item as *"not at all."*

	Not at all	A little	Quite a lot	A great deal
VOCAL				
Cries	0	1	2	3
Moans/groans/screams	0	1	2	3
SOCIAL				
Cheerful	3	2	1	0
Sociable/responsive	3	2	1	0
Not cooperative (cranky, irritable)	0	1	2	3
Obstinate (e.g. doesn't respond to directions)	0	1	2	3
Withdrawn or depressed	0	1	2	3
Hard to console or comfort	0	1	2	3
Difficult to distract	0	1	2	3
FACIAL				
Frowns/has furrowed brow/looks worried	0	1	2	3
Squinting eyes/eyes wide open/eyes frowning	0	1	2	3
Mouth turned down	0	1	2	3
Lips puckered up, tight, pouting, or quivering	0	1	2	3
Grimaces/screws up face	0	1	2	3

	Not at all	A little	Quite a lot	A great deal
Grinds teeth/clenches teeth	0	1	2	3
ACTIVITY/CHALLENGING BEHAVIORS				
Less active or quiet	0	1	2	3
Restless/agitated	0	1	2	3
Self-injurious behaviors (Biting self, banging/hitting head)	0	1	2	3
Aggressive (e.g. hitting others, throwing objects)	0	1	2	3
Acts out/Misbehaves	0	1	2	3
Disturbed sleep	0	1	2	3
Change in eating habits	0	1	2	3
Resists being moved	0	1	2	3
Increase in OCD-like behaviors	0	1	2	3
BODY AND LIMBS/PHYSIOLOGICAL				
Stiffens/spasms/seizures	0	1	2	3
Touching or rubbing parts of the body more than usual	0	1	2	3
Guarding a part of the body	0	1	2	3
Specific body movement to indicate pain (e.g. arms down, curled up, head down)	0	1	2	3
Change in color (e.g., pale, splotchy, flush)	0	1	2	3
Sharp intake of breath/gasping	0	1	2	3

Stratton & Hartshorne, 2012: The CNVPA was created from parental input based on behaviors observed of children with CHARGE when they are experiencing pain and from the following references (used with permission from the authors of the NCCPC-R and the PPP): Breau, L., McGrath, P.J., Finley, A., & Camfield, C. (2004). Non-communicating children's pain checklist-revised (NCCPC-R). Halifax, Nova Scotia: Lynn Breau.

Hunt, A. (2003). Paediatric Pain Profile. Oxford, UK: RCN Institute.

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: Most Significant Items

- 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

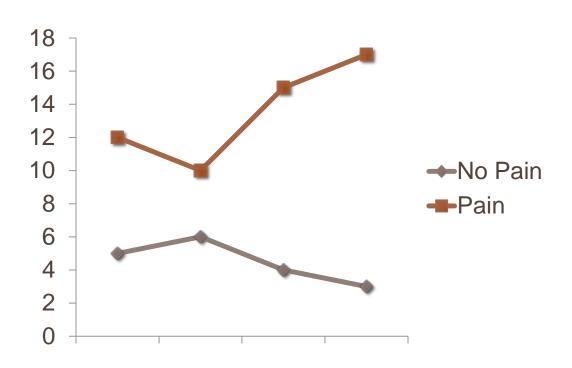
"But what about age?"

- 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

Measuring Pain

- Baseline:
 - CNVPA on good day
- CNVPA on a pain day



PAIN COPING

Understanding Pain

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



Pain Coping

"...one of the most significant behavioral contributions to outcomes, functioning, and adjustment in both children and adults."

(Boothby et al, 1999; Hermann et al., 2007)

Teaching to Communicate Pain

- PEC, Sign, Word
- Label
- Look for & react to pain experiences





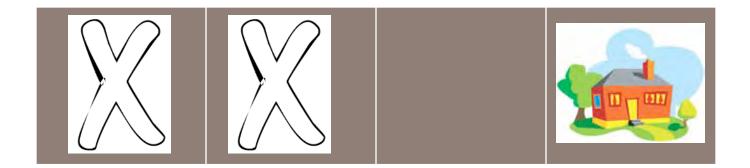
Preparing for Pain

- Be honest
- "Fix" versus "cut"
- Identify part of the body
- Change the environment: "Just lunch!"
- Ask for a Child Life or Play Specialist
- Online/In-person Hospital Tours



Preparing for Pain

- Pain in coming now!
 - Use Visuals or Touch
- Once appropriate, begin a count down for home



Distraction

Active Distraction for planned procedures

(Jameson, Trevena, & Swain, 2011)

"Shift attention to something engaging and attractive to hinder attention to painful stimuli thereby reducing pain, distress, and anxiety"

(Koller & Goldman 2012; Kleiber & McCarthy, 2006; Lambert, 1999)



Active Distraction Techniques

- Interactive Apps
- Electronic toys
- Handheld Videogames
- Virtual Reality (3D)

- Blowing
 - Bubbles
 - Party blowers
 - Paper balls
- Favorite items





Passive Distraction Techniques

- Movie/TV
 - Question characters
 - Find objects
- Guided Imagery
 - Magic Island: Relaxation for Kids (age 5-12)

- Music?
- Reading a story book?



Reducing the pain experience

- Use CNVPA to track progress over time
- Mitigation
 - Analgesics
 - Dietary change
- Redesign the environment
 - Reduce demands; change experience
- Teaching coping skills
 - Self-advocacy
 - Functional communication alternatives
- Parental Interaction with the medical environment

In Closing...

- 1. All behavior is communication.
- 2. Teach a functional way to communicate pain.
- 3. Consider pain **before** attempting to modify behavior.
- 4. Encourage medical/educational teams to use CNVPA to assist with ruling out pain.





Current projects:

- CHARGE vs. Autism
- Social Media & CHARGE Awareness
- Relationships & Divorce
- Individualized
 Education Plans



facebook.com/BulldogCHARGELab

@BulldogCHARGE

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Thursday, July 30, 2015 Breakout Session #2: Euphoria Ballroom 2:45-4:15pm

Aspects of Immunological and adrenal function of CHARGE syndrome

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

Presenter Information:

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

Presentation Abstract:

We studied immunological and adrenal function and their clinically related problems in 24 children with CHARGE syndrome. We found that otitis and airway infections, including pneumonia, frequently occurred in children with CHARGE syndrome. Immunological abnormalities such as decreased CD8+ T cells numbers and reduced antibody responses to vaccination were found. Central adrenal insufficiency was present in only one patient with CHARGE syndrome.

"Defense mechanisms: Immunological and Adrenal function in CHARGE syndrome"

About the presenter



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

Introduction to the study

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

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

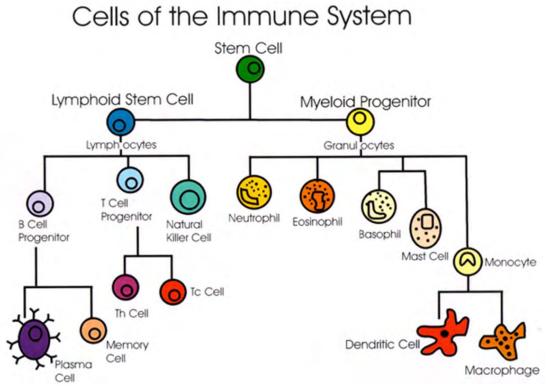


Figure 1. Cells of the immune system. All immune cells were explored in this study.

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

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

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

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

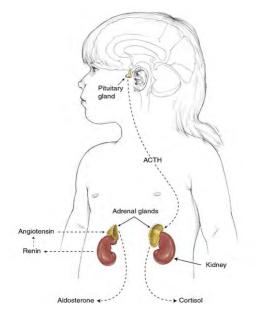


Figure 2. Anatomy of the adrenal glands

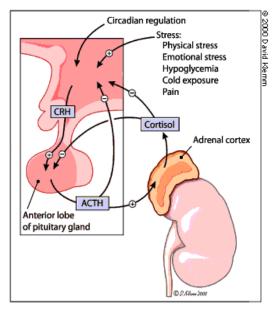


Figure 3. Regulation of the release of cortisol

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

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

What have we done in our study?

Participants

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

Immunological laboratory tests

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

Adrenal function test

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

What have we found in our study?

Frequent infections

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

Immunological abnormalities

Three main abnormalities were observed:

1. Decreased numbers of T-cells.

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

2. Incomplete formation of memory B-cells.

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

3. Reduced immune responses to vaccines.

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

Adrenal insufficiency is not common in CHARGE syndrome

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

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

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

Adrenal function

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

Acknowledgements

The following people were involved in conducting the study:

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

The study was supported by grants provided by the NutsOhra Foundation.

Contact information

For further information or questions, please do not hesitate to contact me. You can send an e-mail to: <u>charge@umcg.nl</u>



Thursday, July 30, 2015 Breakout Session #2: Euphoria Ballroom 2:45-4:15pm

Teaching Safe Sexuality: From the Early Years and Beyond

Kasee Stratton, PhD, NCSP Mississippi State University Jeremy Kirk, M.D, FRCPCH, FRCP Diana Princess of Wales Children's Hospital, Birmingham UK

Presenter Information:

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She currently runs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome* and the developer of the CHARGE Non-Vocal Pain Assessment. Dr. Stratton specializes in challenging behavior and improving adaptive skills.

Dr. Jeremy Kirk is a Consultant Pediatric Endocrinologist (Honorary Reader) at the Diana Princess of Wales Children's Hospital in Birmingham, UK. Jeremy has a special interest in hormonal and growth problems in CHARGE syndrome.

Presentation Abstract:

All you ever wanted to know about sex hormones, preparing for puberty, and sexuality in CHARGE! Dr. Kirk will cover investigation of pubertal problems and optimal hormone replacement, achieving the best results whilst avoiding unacceptable side effects. Dr. Stratton will present on the development of sexuality from identifying body parts to preparing for masturbation. Specific teaching tools will be presented to help your son or daughter with CHARGE better understand the changes to their body.



Thursday, July 30, 2015 Breakout Session #2: Euphoria Ballroom 2:45-4:15pm

The CHARGE Syndrome Research Lab at Central Michigan University

Timothy S. Hartshorne, Ph.D., Benjamin Kennert, Megan Schmittel, Rachel Malta, Hayley Hoesch, Gretchen Imel, Amanda Odren, Claire Latus-Kennedy

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 deafbind 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:

The CHARGE Lab at Central Michigan University began about 1999 when Tim Hartshorne began to study behavior in children with CHARGE. This presentation provides a bit of history, and then describes the research of the current lab members.

The CHARGE Lab

Central Michigan University Timothy S. Hartshorne, Ph.D.

A bit of History

- 1979 Hall identifies an association
- 1981 The association is called CHARGE
- 1993 First US conference nothing on behavior
- 1995 Bernstein presented on behavior in adolescents at 2nd US conference
- 1996 Workshop at 2nd Australasian conference on behavior by Williams, Hartshorne, and Hartshorne
- 1997 Bernstein presents data at 3rd US Conference

Personal History

- 1989 Jacob is born with CHARGE
- Odd, autistic-like behaviors in first two years. Introduced to the impact of deafblindness.
- Looked at parent-professional relationships and impact on parents
- As behavior problems became evident, figured out that a psychologist was needed

Lab Beginnings

- 1999 decided to be the psychologist who studies behavior in CHARGE
- Very confused by the behavior
- Abby Cypher assigned as a graduate assistant
- Hartshorne, T. S., & Cypher, A. D. (2004). Challenging behavior in CHARGE syndrome. *Mental Health Aspects of Developmental Disabilities*, 7(2), 41-52.

Initially the lab was staffed by graduate students who were somewhat arbitrarily assigned as research assistants

- Tina Grialou
- Nichole Dailor

Joanna Russ

- A junior interested in autism joined the lab in Fall, 2003.
- She died in an auto accident in March, 2004.
- Set up an endowed scholarship in her name to support undergraduates studying CHARGE



Moved into office space in 2003

• A junior psychology major joined the lab in Fall, 2004. She remained for 7 years.

Kasee Stratton, Ph.D.

• The lab became much more formalized.

Trying to understand the behavior

- It's because they are deafblind
- It's because they have communication problems
- It's because of vestibular difficulties
- It's because of early experiences
- Research on
 - Executive function
 - Autistic like behaviors
 - Attachment
 - Sleep disturbances
 - Psychiatric diagnoses and medication
 - Self-regulation

Lab Students

- Maria Ramirez
- Ben Kennert
- Megan Schmittel
- Rachel Malta
- Hayley Hoesch
- Mandy Odren
- Gretchen Imel
- Clair Latus-Kennedy

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Thursday, July 30, 2015 Breakout Session #1: Utopia A-B 1:00-2:00pm

Generation and characterization of Chd7-iCre transgenic mice as a tool for lineage tracing and gene deletion

Jennifer M. Skidmore, B.S, Donald L. Swiderski Ph.D., and Donna M. Martin, M.D., Ph.D., Departments of Human Genetics and Pediatrics and Otolaryngology, The University of Michigan, Ann Arbor

Presenter Information:

Ms. Skidmore is a long time research associate of Dr. Martin and the manager of the Martin lab. She is particularly interested in genetic interactions between CHD7 and its cofactors.

Dr. Swiderski is a Research Specialist in the laboratory of Dr. Yehoash Raphael in the Kresge Hearing Research Institute and an Adjunct Assistant Research Scientist in the University of Michigan Museum of Zoology. His interests include the genetic basis of skeletal development in the head and neck. He has contributed to publications on the roles of CHD7 and related genes in the development of the middle and inner ear.

Dr. Martin is an Associate Professor of Pediatrics and Human Genetics at the University of Michigan. In addition to working with children and families affected by numerous genetic disorders, including CHARGE syndrome, Dr. Martin devotes a significant amount of time to the study of CHD7 and related genes implicated in CHARGE pathogenesis. Her laboratory has developed several mouse models of Chd7 deficiency used to study CHARGE syndrome by investigators worldwide. Dr. Martin has published extensively on the role of CHD7 in inner ear and neural development. She is the Chair of the Scientific Advisory Board for the CHARGE Syndrome Foundation.

Presentation Abstract:

We have generated a mouse model of *CHD7* function that will allow researchers to determine the critical temporal and spatial requirements for *CHD7*, leading to the discovery of potential interacting partners for *CHD7* and novel CHARGE disease genes.



Thursday, July 30, 2015 Breakout Session #1: Utopia A-B 1:00-2:30pm

Studies of CHD7 function in CHARGEpatient derived induced pluriopotent stem cells

Peter C. Scacheri¹ and Donna M. Martin² ¹Department of Genetics and Genome Sciences, Case Western Reserve University School of Medicine ²Departments of Pediatrics and Human Genetics, University of Michigan

Presenter Information:

Dr. Martin is an Associate Professor of Pediatrics and Human Genetics at the University of Michigan. In addition to working with children and families affected by numerous genetic disorders, including CHARGE syndrome, Dr. Martin devotes a significant amount of time to the study of CHD7 and related genes implicated in CHARGE pathogenesis. Her laboratory has developed several mouse models of Chd7 deficiency used to study CHARGE syndrome by investigators worldwide. Dr. Martin has published extensively on the role of CHD7 in inner ear and neural development. She is the Chair of the Scientific Advisory Board for the CHARGE Syndrome Foundation.

Dr. Peter Scacheri is an Associate Professor in the Department of Genetics and Genome Sciences at Case Western Reserve University, and a member of the Case Comprehensive Cancer Center. He received his undergraduate training at Gettysburg College and his PhD in Biochemistry and Molecular Genetics from the University of Pittsburgh. He trained as a postdoctoral fellow at the National Human Genome Research Institute in Bethesda, MD. He is known for his studies on the role of gene regulatory elements in human diseases. Dr. Scacheri has co-authored more than 50 publications to date.

Presentation Abstract:

Two-thirds of individuals clinically diagnosed with CHARGE syndrome have heterozygous loss-of-function mutations in the gene encoding chromodomain helicase DNA-binding protein 7 (CHD7). In mouse embryonic stem cells, we previously showed that CHD7 binds to gene enhancer elements containing high levels of H3K4me1 and regulates transcription. CHD7 also localizes to the nucleolus and regulates transcription of ribosomal RNA. The findings suggest that dysregulated expression of genes normally regulated by CHD7 during development gives rise to the developmental defects observed in CHARGE syndrome. As part of an effort to translate these findings from mice to humans, we utilized state of the art reprogramming technologies to generate human induced pluripotent stem cells (iPSCs) from skin cells collected from two patients clinically diagnosed with CHARGE syndrome and positive for *CHD7* mutation. Progress on this study will be discussed.



Thursday, July 30, 2015 Breakout Session #1: Utopia A-B 1:00-2:30pm

Novel treatments for deafness and balance disorders

Yehoash Raphael, Ph.D. Kresge Hearing Research Institute, Department of Otolaryngology – Head and Neck Surgery, The University of Michigan, Ann Arbor and Donna M. Martin, M.D., Ph.D. Departments of Pediatrics and Human Genetics, The University of Michigan Medical School, Ann Arbor

Presenter Information:

Dr. Raphael's research is dedicated to protection, repair and regeneration of hair cells and neurons in the inner ear. The research group members are exploring methods based on gene therapy and pharmacological approaches to prevent loss of hair cells and neurons or to replace them. For hereditary inner ear disease such as CHARGE, we are exploring phenotypic rescue with gene transfer of the wild type gene. We are also developing means to enhance survival of auditory neurons in deaf ears and to induce their sprouting towards to cochlear prosthesis, in order to improve functionality of the electrode.

Presentation Abstract:

Our objective is to develop novel therapies for repair and regeneration of the cochlear epithelium and the auditory nerve in deaf ears. The objective of one set of experiments was to assess the influence of neurotrophin gene therapy on auditory neurons in deaf ears. Neurotrophin experiments were performed on guinea pigs deafened with an ototoxic lesion and on mice with a deafness mutation modeling human hereditary hearing loss. Adult guinea pigs were deafened by neomycin or kanamycin and furosemide. Adeno-associated viral vectors (AAV) with BDNF or NTF3 gene insert were injected into the perilymph one week later. In cochleae that were obtained three months later, the extent and pattern of nerve sprouting was assessed, along with spiral ganglion nerve survival. Similar experiments were performed using a mouse model for a connexin 26 (Cx26) mutation, in which cre-Sox10 drives excision of the Cx26 gene from supporting cells of the auditory epithelium. In this model the peripheral fibers of the auditory nerve die back followed by death of the neurons. Overall, the results suggest that gene therapy with either BDNF or NT-3 leads to peripheral auditory nerve fiber re-growth, and treatment with BDNF leads to enhanced SGN survival. In Gjb2-CKO mice injected with Ad.BDNF at 1 month of age spiral ganglion neurons in the basal cochlear turn were rescued. The goal of the other experiments was to test methods for integrating exogenous cells in the mature deaf cochlea, in preparation for stem cell implantation. Specifically, we tested whether deaf cochleae can be "conditioned" to "accept" implanted exogenous cells and promote their survival and integration. To condition the cochlea, we used guinea pigs deafened with neomycin and performed procedures aimed at transiently lowering potassium levels in endolymph and opening the apical junctions in the auditory epithelium. We determined that exogenous cells injected into scala media survived in the conditioned cochleae for at least 7 days, but in un-conditioned (control) cochleae they degenerated promptly. Together, our data show that the cochlea can be manipulated to enhance nerve survival and sprouting and to accept and maintain exogenous cells. This can be accomplished in in ears both environmentally and genetically caused cochlear pathologies.



Thursday, July 30, 2015 Breakout Session #1: Utopia A-B 1:00-2:00pm

Basic Science Research on CHD7 with a focus on inner ear development and function

Ethan D. Sperry, B.S., B.A., Diana F. Syam, M.Sc., Donna M. Martin, M.D., Ph.D., Donald L. Swiderski, Ph.D. and Yehoash Raphael, Ph.D., Departments of Human Genetics and Pediatrics, The University of Michigan, Ann Arbor

Presenter Information:

Mr. Sperry is an M.D./Ph.D. student in Human Genetics at the University of Michigan. He is interested in the genetic etiology of complex disorders, including CHARGE syndrome, and recently has been focusing on roles for CHD7 and SOX11 in development of the vestibular system. Previously, Mr. Sperry has published on the role of Chd7 in the development of the skeletal and cardiorespiratory systems. Mr. Sperry aims to work with children and families affected by genetic disorders, both in the clinic and in the laboratory.

Ms. Syam is a research associate who recently began training in the laboratory of Dr. Martin at the University of Michigan. She is interested in understanding how genetic disorders cause deafness and is investigating the role of CHD7 in the development and function of the cochlea.

Dr. Martin is Associate Professor of Pediatrics and Human Genetics at the University of Michigan. In addition to working with children and families affected by numerous genetic disorders, including CHARGE syndrome, Dr. Martin devotes a significant amount of time to the study of CHD7 and related genes implicated in CHARGE pathogenesis. Her laboratory has developed several mouse models of Chd7 deficiency used to study CHARGE syndrome by investigators worldwide. Dr. Martin has published extensively on the role of CHD7 in inner ear and neural development. She is the Chair of the Scientific Advisory Board for the CHARGE Syndrome Foundation.

Presentation Abstract:

Many individuals with CHARGE syndrome have difficulty with hearing and balance. Our laboratory has been studying mice with mutations in *Chd7*, the most common gene mutated in CHARGE. We will discuss recent data indicating roles for *CHD7* and associated genes in development and function of the inner ear, including the cochlea (hearing) and vestibular system (balance).



Thursday, July 30, 2015 Breakout Session #1: Utopia A-B 2:45-4:15pm

Direct visualization of gastrointestinal morphology and motility changes associated with *chd7* loss of function in a zebrafish (*Danio rerio*) model of CHARGE syndrome

Shelby L. Steele, Kellie Cloney, Department of Medicine, Dalhousie University, Halifax NS CANADA

Matthew Stoyek, Department of Physiology and Biophysics, Dalhousie University, Halifax NS CANADA

Frank Smith, Department of Medical Neuroscience, Dalhousie University, Halifax NS CANADA

Kim Blake, Department of Pediatrics, Dalhousie University, Halifax NS CANADA Jason N Berman, Departments of Pediatrics, Microbiology and Immunology, and Pathology, Dalhousie University, Halifax NS CANADA

Presenter Information:

Shelby's interest in zebrafish as a model organism began when she was a doctoral student at the University of Ottawa, where I studied the hypoxic and adrenergic regulation of the cardiovascular system in zebrafish larvae. In her current postdoctoral position at Dalhousie University, she has taken her expertise on the zebrafish cardiovascular physiology and translated it into a focus on modeling rare diseases involving multiple organ systems including the heart, eye, skin, kidney, and blood, in the zebrafish. This has mostly been due to her involvement in the IGNITE project, a collaboration of clinicians and basic researchers with the goal of finding novel therapeutics for orphan diseases. Based on her expertise and the previously published utility of the zebrafish to model CHARGE syndrome, and along with her postdoctoral advisor Dr. Jason Berman, they formed a collaboration with Dr. Kim Blake, a clinician at the IWK Health Centre and an expert on CHARGE syndrome, and Kellie Cloney, a Dalhousie medical student working on CHARGE syndrome as part of her longitudinal Research In Medicine project.

Presentation Abstract:

We have used the zebrafish *chd7* morpholino knockdown model to determine whether gastrointestinal (GI) motility characteristics are related to decreased food uptake and/or gastroparesis associated with CHARGE syndrome. We have observed that spontaneous gastrointestinal peristalsis is decreased in *chd7* morphant larvae compared to controls, and we are employing standardized gavage feeding techniques and RNA sequencing analysis to characterize the physical and genetic consequences of *chd7* loss of function in the GI tract.



Thursday, July 30, 2015 Breakout Session #2: Utopia A-B 2:45-4:15pm

Exploring the link between CHD7 and the COMPASS-like complex

Louis Gervais, PhD., Patricia Skorski†, Carolina Perdigoto†, Allison Bardin† †Institut Curie, CNRS/INSERM

Presenter Information:

Louis is a French scientist at the Curie Institute in Paris who is interested in the regulation of stem cell activity *in vivo*. In particular he is studying the role of chromatin factors on adult stem cell using the fruit fly, *Drosophila* melanogaster, as a model system. He started working on the fly CHD7 (Kismet) after identifying it in a genetic screen as being important for control of adult stem cells. Their belief is that this work will provide insight into the fundamental molecular mechanisms of CHD7/Kismet activity.

Presentation Abstract:

Despite its role in regulation of gene expression, CHD7 modes of action and interactions with other chromatin factors are not well identified. Using the *Drosophila* intestine as a model to study chromatin remodeling factors *in-vivo*, we identified a possible link between CHD7 (Kismet in flies) and the COMPASS-like complex, a histone-modifying enzyme defining enhancer transcriptional status. Our work aims to elucidate the mechanisms underlying CHD7/Kismet and COMPASS-like complex collaboration.

Mutations in CHD7 are a common genetic cause of CHARGE Syndrome. Although most of the disorders affecting individuals with CHARGE are due to defects during embryonic development, recent published work points toward essential functions of CHD7 in adult tissues. Therefore, it is essential to better understand the role of *CHD7* in adult tissues. In addition, despite a known role in regulation of gene expression, CHD7 modes of action and interactions with other chromatin factors are not well identified.

Using the *Drosophila* intestine as a model to identify new factors regulating tissue homeostasis and adult stem cell activity we found that clonal inactivation of the CHD7 orthologous (*kismet*) gene results in a strong increase in relative number and density of intestinal stem cells in the gut. This is due to an abnormal division rate of CHD7/*kismet* mutant stem cells compared to wild-type stem cells as illustrated by aberrant activation of signaling pathways known to drive proliferation of intestinal stem cells such as EGFR and Jun kinase signaling. Consistent with this, inactivation of these pathways suppresses *kismet* phenotypes indicating that Kismet is an important repressor of intestinal stem cell proliferation essential to maintain proper gut homeostasis.

In an attempt to test other chromatin remodeling factors *in vivo* for their role in the control of adult stem cell activity (proliferation and differentiation) we found that 3 components of Compass-like complex reproduce Kismet phenotypes. This genetic data suggests that Kismet and COMPASS-like complex act together in a similar process.

Consistent with a possible functional interaction of COMPASS-like and Chd7/Kismet, mutations in COMPASS-like components have been associated with the Kabuki syndrome an other congenital disease sharing many of the same medical and behavioral features with CHARGE syndrome.

The COMPASS-like complex is a histone-modifying enzyme essential in defining enhancers transcriptional status. Therefore, we are currently testing the relationship between CHD7/Kismet and the COMPASS-like complex at the level of enhancers in general and in particular in adult stem cells in a homeostatic tissue.



Thursday, July 30, 2015 Breakout Session #2: Utopia A-B 2:45-4:15pm

CHD7 controls cerebellar growth via Reelin

M. Albert Basson, PhD, King's College London

Danielle E. Whittaker^{1,2*}, Tian Yu^{1*}, Kimberley L. Riegman^{1*}, Sahrunizam Kasah¹, Blanca Pijuan Sala¹, Husam Hebaishi³, Ana Marques⁴, Apar Shah¹, Chris Ponting⁴, Fiona Wardle³, Imelda McGonnell², Cathy Fernandes⁵, & M. Albert Basson^{1,6} ¹King's College London, Department of Craniofacial Development and Stem Cell Biology

²Royal Veterinary College

³Randall Division, King's College London

⁴Department of Physiology, Anatomy and Genetics, University of Oxford

⁵MRC Social, Genetic & Developmental Psychiatry Centre, PO82, Institute of Psychiatry,

Psychology & Neuroscience, King's College London

⁶King's College London, MRC Centre for Developmental Neurobiology

Presenter Information:

Albert is a basic scientist with interests in development of the brain, developmental signaling pathways and epigenetic mechanisms. My laboratory creates and uses genetically modified mouse models to dissect the causes of genetic disease. Their work has provided insights into a number of conditions that affect development of the kidney, heart, immune system and brain. Our most recent work has been focused on identifying the causes of cerebellar malformations in CHARGE syndrome. Their aim is to understand the mechanisms whereby CHD7 regulates gene expression during different stages of cerebellar development.

Presentation Abstract:

CHD7 is expressed in proliferating granule neuron progenitors (GNPs) in the early postnatal cerebellum. Using conditional gene deletion strategies, we have deleted CHD7 specifically from these progenitors and identified defects in cell proliferation and differentiation resulting in cerebellar hypoplasia. We identified the Reelin gene as a functional target of CHD7 in these cells and observed mild motor deficits in some of the animals.



Thursday, July 30, 2015 Breakout Session #1: Utopia C-D 1:00-2:30pm

Promoting Early Concept Development through Adapted Books

Christine Spratling Georgia PINES

Presenter Information:

After graduating with a Bachelor of Science degree in Special Education for students who are deaf/hard-of-hearing, Christine started teaching in the public school systems in 1986. Since then, I have taught in a variety of settings from self-contained to itinerant and added a Master's Degree in interrelated special education and an EDS in educational leadership. About 6 years ago, I also began to work for Georgia PINES as a parent advisor in an early intervention setting. Georgia PINES serves families of young children with vision and/or hearing loss including those with CHARGE Syndrome.

In addition to working in the public school and early intervention settings, I have become involved with the Georgia deaf-blind project, the Georgia Sensory Assistance Project (GSAP). Like Georgia PINES, GSAP serves families of children with deaf-blindness including those with CHARGE. I have co-presented with GSAP staff at state conferences and workshops on topics such as literacy, concept development, and transition tools. When NCDB asked GSAP to participate in the development of the Literacy for Children with Combined Vision and Hearing Loss website, I was part of the team.

One of my biggest passions is early emergent literacy for all children. Since commercially available books and other literacy materials are often conceptually inappropriate or confusing for children with dual sensory losses, I have been adapting and making books for individual children of all ages based on their specific level of concept development, mode of communication, and sensory input preferences.

Presentation Abstract:

Concepts are the ideas that give meaning to our world. We constantly develop concepts based on how we experience the world around us. Children who have sensory loss(es) and additional health issues or impairments often have limited experiences and therefore develop concepts that appear counterintuitive to us. Adapted books are a powerful and fun tool to help children develop accurate concepts about their specific environment.

Promoting Early Concept Development

Through adapted books

International CHARGE conference - July 2015 Christine Spratling Georgia PINES & Georgia Sensory Assistance Project



"For people who can see and hear, the world extends outward as far as his or her eyes and ears can reach. For the young child who is deafblind, the world is initially much smaller."



"If the child is profoundly deaf and totally blind, his or her experience of the world extends only as far as the fingertips can reach.

Such children are effectively alone if no one is touching them. Their concepts of the world depend upon what or whom they have had the opportunity to physically contact."



Barbara Miles Overview on Deaf-Blindness DB Link October 2008



to our world.

"Concepts are the ideas that give meaning

We develop concepts based on our particular experiences."

Barbara Miles Developing Concepts with Children Who Are Deaf-Blind DB Link 2008

What happens to concept development when we have limited experiences:



What do you see?



What does the literature say about concepts:

Three groups of concepts:

1. Concrete concepts (tangible things)

2. Semi-concrete concepts (something that can be demonstrated but is not tangible)

3. Abstract concepts



- 1. how the world works
- 2. how the physical environment is arranged and how to navigate it
- 3. where things come from
- 4. how things are sequenced
 - 8

6 Areas of concept development:

- 1. Objects exist
- 2. Objects have permanence
- 3. Objects differ from one another
- 4. Objects have names or labels
- 5. Objects have characteristics
- 6. Objects have functions or use

1. Objects exist



2. Objects have permanence





3. Objects differ











4. Objects have names or labels

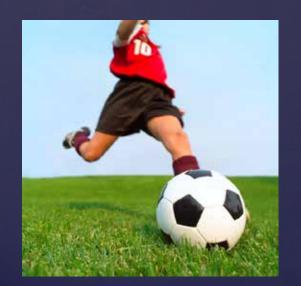
crystal ball, moth ball, meatball, eyeball, gumball, fire ball, snowball, football, eight ball, golf ball, spitball, oddball, goofball, sleaze ball, ball bearing, etc.

5. Objects have characteristics:

- * color
- * size
- * texture
- * weight
- * smell
- * etc.











O&M Concepts

- Body Image Concepts
- Spatial Concepts
- Environmental Concepts

According to Babara Miles (1999), children typically develop concepts in this order:

o self

- people and objects as extensions of herself or as things to be incorporated (put in the mouth)
- o movements
- people as distinct in themselves
- objects themselves

Difference between concepts and skills:

• Skill - the ability to do something



 Concept – the ideas that help us understand "why"



How to choose a concept to work on:

- Start with the child in mind.
- What is meaningful to the child right now?
- What is something that might help the child participate in day to day life?
- What is something that would help the child understand the world around her/him better?
- Remember, this should be a team approach ${\sf J}$

Some assessments for concept development:

School Inventory of Problem Solving Skills (SIPSS)
 Home Inventory of Problem Solving Skills (HIPSS)
 <u>http://www.designtolearn.com/products</u>

3. Functional Scheme:Functions Skills Assessment http://www.lilliworks.com

Strategies that help with concept development:

- Choose activities that happen regularly and are meaningful, such as daily living
- Activities that the child enjoys
- Attach language to all experiences
- Build on language the child already knows
- Remove confusing variables
- Generalize the concept across various situations

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Planning instructional activities for concepts:

CONCEPT	;
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Weeks of: ______ through _____

O&M Skill Development / Real Life Applications

1. Whole body:

Object-to-body:

3. Objects-beyond-the-body:

Concept Category (check one):

- how the world works (cause and effect, purpose and use of objects, where things come from, etc.)
- self-concept (I exist and have feelings, I can do things, I affect people and objects around me, etc.)
- how the physical environment is arranged and how to navigate it (O&M, presence and absence, positional concepts, routes and landmarks, barriers, etc.)

			-
Activity	Main Idea - what are we focusing on?	Signs	Other
Job Box A job box is an activity that follows a teacher directed procedure with a definite beginning and end. *	* an activity with a beginning and an end * structured activities that lead to independence	* * * *	
Concept Items These are any given number of items/toys that help Ariana understand the designated concept. This is more of a child centered activity.	* active learning * mutual, tactual attention	* * * *	
Pre-Literacy finger games, adapted books, book boxes, calendar boxes, etc. *		* * *	

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Weeks of: through ____

CONCEPT : Up/Down

O&M Skill Development / Real Life Applications

1. Whole body: Using stairs/steps, jumping, going up steps/down slide, swinging up/down, stand up/sit down, arms up, legs up, head up, etc.

2. Object-to-body: Brushing teeth, Brushing, Pulling up/down sock, pants, pull-ups,

3. Objects-beyond-the-body: I can throw things up and they will come back down.

Concept Category (check one):

- How the world works (cause and effect, purpose and use of objects, where things come from, etc.)
- Self-concept (I exist and have feelings, I can do things, I affect people and objects around me, etc.)
- How the physical environment is arranged and how to navigate it (O&M, presence and absence, positional concepts, routes and landmarks, barriers, etc.)

Activity	Signs		O&M	OT/PT
			support?	support?
Job Box A job box is an activity that follows a teacher directed procedure with a definite beginning and end. 1. Button Job Box	* work * finished * find	*down *time *up		
 Concept Items These are any given number of items/toys that help lvey understand the designated concept. This is more of a child centered activity. 1. Concept Box: "The Napping House" 2. Stacking blocks, jack in the box, pipe pulling up/down, balls, train/car going up and down tracks, 	* up/down * left/right *work *finished	*find *pull *ball *	Up/Down stairs/steps Up/Down Slide	Therapy room: up/down slide Bike, Swing Pant/ socks: up/down Jump, sit, stand, pull:up/down Brushing
Literacy / pre-Braille tracking a dotted/textured line from left to right and turning page with a rewarding activity at the end of each page 1. Hickory Dickory Dock 2. Chicka Chicka Boom-Boom	* book * touch * turn page * up	* down *finished *		

Notes:

Concept-based activity ideas for "Up and Down" <u>5 Little Monkeys Jumping on the Bed</u>

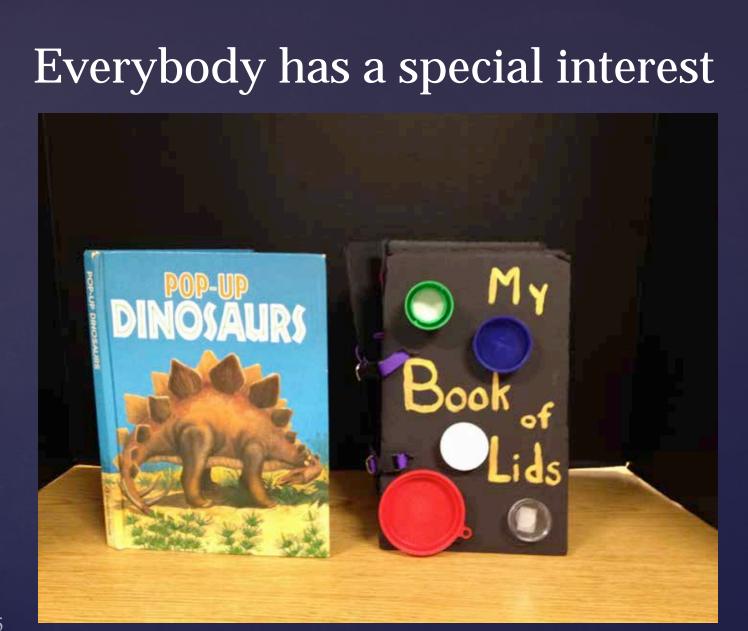
According to Barbara Miles, concepts are the ideas that give meaning to our world. People who can hear and see, experience the world as far as their eyes and ears reach. For our PINES babies who have a hearing or vision loss, the world becomes smaller depending on their sensory loss. For our babies who have a dual sensory loss and possibly additional health issues, the world reaches only as far as their fingertips reach. This has a huge impact on their concept development.

Millie Smith talks about how information is gathered through the senses, how babies with visual and/or multiple impairments access their world using their near and distance senses. According to her, this is the sequence how babies learn concepts:

- 1. Concepts about the learner's own body
- Concepts about people, actions, or objects touching the learner's body
- Concepts about people, actions, and objects beyond the learner's body

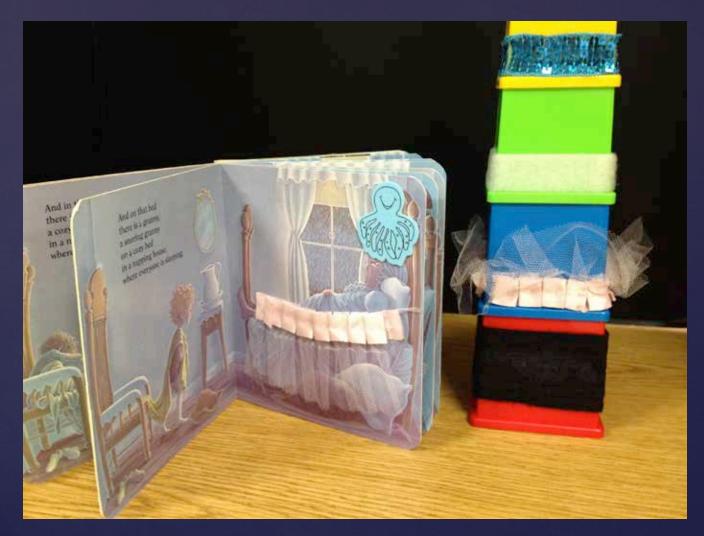
Concept Category	Activity Suggestions		
1. Baby's own body (whole body up and down experiences)	 * Bounce baby up and down on your lap, * Lift baby up and down. * Body parts can go up and down (arms, legs, hands, etc.) * Stand up and sit down. * Jump up and down, etc. 		
2. Objects touching the baby's body	 Clothes go up and down on the body, such as socks and pants. Zippers go up and down. Tickle up and down on baby's arms, legs, etc. Move the monkeys up and down baby's arms, legs etc. 		
3. Objects beyond the baby's body <mark>Book extension ideas</mark>	 * Use the monkeys to jump up on the sponge bed and fall down. * The monkeys on the clothespins can "jump" up and down on the book or fall down from the book. * Balloons, bubbles, balls, etc. go up and down. 		

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Concept book for "on top"

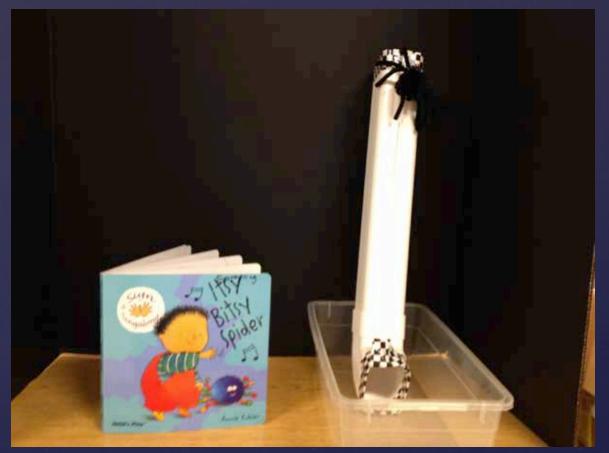




Concept book for "up and down"



Also for "up and down":



Concept book for "rolling"

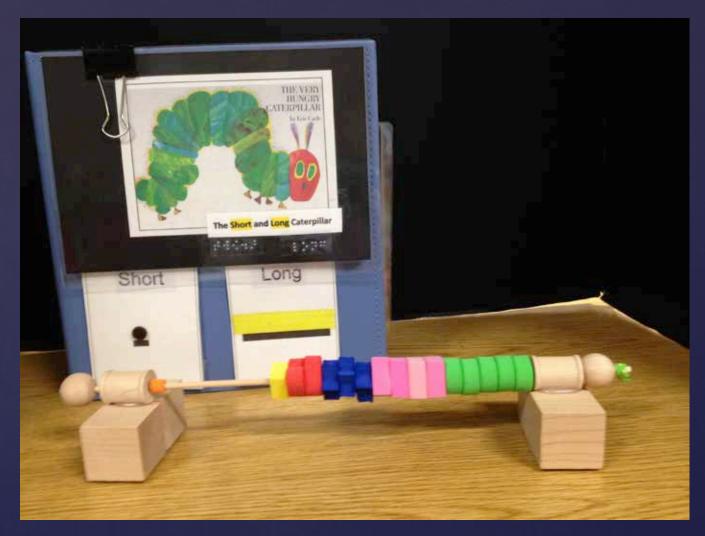


Concept book for "wet/dry"



30

Concept book for "long/short"



31

Resources:

1. Concept Development

http://www.sparkle.usu.edu/Topics/concept_developmen t/index.asp

2. **Developing Concepts with Children Who Are Deaf-Blind** Barbara Miles, M.Ed. & Barbara McLetchie, Ph.D. DB-Link February 1, 2008

3. Early Concept Development by Holly Cooper, <u>http://www.tsbvi.edu/preschool/1117-</u> <u>early-concept-development</u>

More Resources:

4. Understanding Deaf blindness : Issues, Perspectives, and Strategies --Alsop, Linda, M.Ed. (Ed.) Logan, UT: SKI-HI Institute, Utah State University. (2002)

5. What a Concept!

Jim Durkel, CCC-SPL/A and Statewide Staff Development Coordinator (with help from Kate Moss, Stacy Shafer and Debra Sewell) Texas School for the Blind and Visually Impaired Outreach

More Resources:

6. Remarkable Conversations: A Guide to Developing Meaningful Communication with Children and Young Adults Who Are Deafblind

by Barara Miles and Marianne Riggio, 1999



Thursday, July 30, 2015 Breakout Session #1: Utopia C-D 1:00-2:30pm

Hello to the Future!

Eva Seljestad and Wenche Andersen, Statped, Senior Advisers, Deafblindness and Dual Sensory Impairment, Oslo, Norway

Presenter Information:

Eva Seljestad and Wenche Andersen: Preschool teachers, teacher of hearing impaired children/ cand. paed. spec, working with CHARGE syndrome and children with combined hearing- and visual impairment. We visit and observe in schools and nursery schools; we talk with parents and pupils. We give courses where we highlight the challenges that the syndrome gives.

Presentation Abstract:

We will present some themes in a conversation between two girls having CHARGE syndrome. Our perspective is to highlight how they reflect upon the syndrome, and what they know about facts and myths about CHARGE. Another perspective, is how they manage to listen to each other, exchange information and ask following-up questions. When we analyze their conversations, we focus on both facts and more philosophical aspects of their understanding. This might give a valuable contribution to our counseling.

Presenters information

Eva Seljestad, Senior Adviser, Deafblindness and Dual Sensory Impairment, Statped, Oslo, Norway. Preschool teacher, audiopedagog, kindergarten/school for hearing impaired children, sign language. Working with dual sensory impairment and CHARGE syndrome for 15 years.

Wenche Helene Andersen, Senior Adviser, Deafblindness and Dual Sensory Impairment, Statped, Oslo, Norway. Preschool teacher, Cand. Ped. Spes. with focus on social and emotional problems. Working with dual sensory impairment and CHARGE syndrome for 15 years.

Abstract information: Background for attached presentation "Hello Future"

We have called our presentation; "Hello Future". This hopeful and optimistic expression, was uttered during a conversation we had with two girls, in January, this year.

We focus on, how to facilitate a conversation between the girls, 12 and 15 years old, on the subject, having CHARGE syndrome. Our perspective is to highlight how they reflect upon the syndrome, including facts and myths on CHARGE.

At this age, questions arise, like; "Who am I?" The main issue, in this period, is related to identity and self-esteem. You measure yourself in schoolmates, in "movie stars", your reflection in the mirror, in friends and significant others.

Having CHARGE syndrome, makes questions concerning identity more complex and challenging. When and how they are informed about the syndrome, might influence possible existential pain. Having CHARGE syndrome is a lifelong challenge. How you accept the diagnosis, greatly influences on your quality of life. Acceptance is the golden key.

Another point to consider, is how they listen to each other, exchange information and ask follow-up questions. When we analyzed their conversations, we noticed some communication challenges.

A lasting conversation, demands communication skills that make it possible to maintain focus. Often those skills are reduced due to dual sensory loss. Secondly, focus can be blurred, when attempting to keep your body under control, not being distracted by your "special interests" and to maintain attention.

Our underlying intention, in bringing these two girls together, was the hope that they, in the future, could be useful for each other as peers.

Keywords: CHARGE syndrome, facts and myths Communication and psycosocial challenges Children being peers to each other



Hello Future

Chicago

Professional day 31. July 2015

Senior advicers Eva Seljestad and Wenche Andersen

Statped sørøst, departement for deafblindness and Dual Sensory Impairment, Oslo, Norway

Comfortable talking face to face

- Girls maintaining stable position
- Girls sitting at eye-level
- Girls with relatively calm bodies
- No background sounds



Information about diagnosis

- Who told you that you have CHARGE syndrome?
- How old were you?



The letters in CHARGE

- The acronym CHARGE
- C?
- H?
- A?
- R?
- G?
- E?



CHARGE and learning

- «You learn less than others»
- Actually: « You have to practice a little bit more and a little bit longer»



Becoming peers?

- Shared experiences
- Lack of knowledge
- Lack of interest
- Great interest for hobbies, friends, parties (all night)
- A shared, common peer process begins?



Age is relative

Philosphical question:

- «Am I little?»
- «Are you little when you are 6 years old?»
- «What do you think yourself?»
- «I am much bigger than those under six, but much smaller than those over sixteen»



CHARGE in CHICAGO 2015

- «I am going to Chicago, to a place where everybody has CHARGE».
- «I am going to tell that everything will be OK in THE FUTURE».



HUMAN BEINGS

«NOBODY IS EQUAL».

• «EVERYBODY IS UNIQUE».



«Can we do this again?»

- Doing and talking is a good combination
- Making food and making statements/reflections
- Simple and safe surroundings for difficult and serious talks



Comfortable techno-talking

- Cellphone/ iPHONE
- ▶ iPAD
- ► PC

► SKYPE



Communication /psychosocial challenges

- Auditive and visual problems
- Control of the topics
- Articulation
- Consentration and energy
- Few friends
- Too many grown-ups
- Repeating the schedule over and over again



HELLO FUTURE !

- Hopeful and optimistic
- Peers are fine
- The future will be OK
- Acceptance is the golden key!





Thursday, July 30, 2015 Breakout Session #2: Utopia C-D 2:15-4:30pm

The Lived Experience of Parents of Children with Charge Syndrome in Advocating and Navigating Systems

Seth Harkins, EdD, Philip J. Rock Center & School, Glen Ellyn, IL; National Louis University, Chicago, IL

Presenter Information:

Dr. Seth Harkins is chief executive officer of the Philip J. Rock Center and School in Glen Ellyn, IL. He is a career educational administrator, having served in a variety of leadership roles in public and private education and state government. A former special education teacher, special education director, principal, assistant superintendent, and superintendent, Dr. Harkins has served students with disabilities, their families, and teachers in elementary, middle, and secondary school settings. Dr. Harkins earned his BA degree from Coe College, Cedar Rapids, IA, his MA in Special Education and MA in educational Administration and Supervision, from Northeastern Illinois University, Chicago, IL, and his Ed.D, from National Louis University, Chicago, IL. Dr. Harkins has taught graduate students in special education as an adjunct professor and assistant professor at National Louis University since 1983. His private consulting practice focuses on school leadership, management, organizational research, and program evaluation. From 1978 to 1998, Dr. Harkins was an impartial due process hearing officer for the Illinois State Board of Education. Additionally, he has been active in group relations conference work since 1985 and is a member of the A.K. Rice Institute for the Study of Social Systems. His community volunteer work includes: statewide leadership teams and task forces, past president and secretary of the Chicago Center for the Study of Groups and Organizations; legislative designee to the board of directors of the Illinois Community and Residential Services Authority; and board member of the Chicago Virtual Charter School.

Presentation Abstract:

Parents of children with complex disabilities are particularly challenged in advocating for their youth as they navigate the complex maze of special education and human service systems. This presentation examines the narratives of parents of children with Charge Syndrome and their experiences in developing and sustaining partnerships with professionals, as they and their children traverse early intervention services through public education, and transition to adult services. Effective leadership, advocacy, and collaboration strategies are examined.

Family Leadership and Empowerment: Parents of Youth with Charge Syndrome and Navigating Human Service Systems

Seth Harkins, EdD. Chief Executive Officer, Philip J. Rock Center and School Scott Boroff, Parent Allison Stahl, Parent Janay and Saudi Mohamed, Parents Maria Roeters. Parent Karen Windy, Philip J. Rock Deaf-Blind Specialist

Abstract

As a very low incidence disability, there is little in the professional literature about the lived experience of parents and families of these children with Charge Syndrome. The research model for this presentation is narrative inquiry and qualitative methodology. Audio recordings of interviews were triangulated with with Individual Education Programs (IEPs), medical reports, and special education case study evaluations. The presentation focuses on the narratives of parents of children with Charge Syndrome as they encountered human service systems and experienced turning points, which mobilized their personal authority for empowerment and advocacy. Parents of children with Charge Syndrome are particularly challenged in advocating on behalf their youth, as they navigate the complex social emotional realities and the maze of medical, early intervention, education and government human service systems. Because the syndrome involves complex sensory, medical, educational, family and social systems dimensions, much can be learned through the stories of parents as they advocate on behalf of their children. This research therefore examines the narratives of parents and their experiences in developing and sustaining partnerships with professionals, as they and their children traverse medical, early intervention, education, and adult service systems. Effective leadership, family empowerment, advocacy, and collaboration strategies parents and service providers are examined.

Presentation Outline

- I. Social Systems and Family Systems Perspective:
 - a. Benevolent Holding Environments
 - b. Unconscious Phenomena: Micro insults, micro invalidation
 - c. Grief, Mourning, Acceptance, and Taking Up Personal Authority
 - d. Parent and Family Empowerment and Advocacy
- II. Navigating Medical Services
 - a. Experiencing the Diagnosis
 - b. Objectification and Turning Points

- c. Partnering with Medical Professionals and Addressing Developmental Challenges
- III. Navigating Early Intervention Services
 - a. Experiencing Early Intervention Professionals
 - b. The Individual Family Service Plan as a Lived Experience
 - c. Developmental Challenges and Turning Points
 - d. Transitioning to Early Childhood Services
- IV. Navigating Special Education Services
 - a. Experiencing the Special Education Maze
 - b. The Individual Education Program (IEP) as a Lived Experience
 - c. Turning Points and the Challenge of Building Partnerships with Educational Professionals
- V. Looking Ahead to Transition and Navigating Adult Services
 - a. Dreams, Best Hopes, and Worst Nightmares
 - b. Transition from Mandated to Eligibility Service Systems
 - c. Transition to Post-secondary Adult Services
- VI. Dialogue and Conclusions



Thursday, July 30, 2015 Breakout Session #1: Utopia C-D 1:00-2:30pm

An educational model to reduce passivity and increase selfdetermination in deafblind learners resulting in improved connection, communication, and learning

Kimberly Lauger, Redtail Neurodevelopmental Center, Consultant for the Arizona Deafblind Project

Presenter Information:

As an RN caring for children with complex health conditions, Kimberly Lauger never dreamed she would later have her own child with CHARGE syndrome or how Dylan's birth would take her on an odyssey of further learning and specialization.

Driven to understand the complexities of Dylan's behavioral and learning challenges, she became a Certified HANDLE[®] Practitioner. Looking at behavior from a sensory rather than a cognitive perspective, Kimberly provides HANDLE evaluations, programs, and trainings for individuals and families from across the country at her Redtail Neurodevelopmental Center.

An Intervener Trainer and Consultant for the Arizona Deafblind Project, Kimberly is also a contributor to A Family's Guide to Interveners for Children with Combined Vision and Hearing Loss and the Open Hands, Open Access: Deaf-Blind Intervener Learning Modules.

Most recently, Kimberly has developed, coordinated, and implemented a team driven home and community based educational program for Dylan. Following Dylan's interests and measuring success through smiles and laughter, Dylan has shown improvements in communication, learning, relationships, and independence.

Presentation Abstract:

Emerging from one team's efforts to meet the needs of Dylan, a 17-year old boy with CHARGE syndrome, this session will cover an educational model that supports connection, communication, and learning in deafblindness. Committed to following Dylan's interests and measuring smiles and laughter instead of goals, his team was therefore encouraged to see the improvements in health, mood, communication, literacy, learning, and friendships that emerged, as he became an active participant in school and home.

An educational model to support improved connection, communication, and learning, through reduced passivity and increased selfdetermination in deafblind learners.

An excerpt from the teaching memoir, Learning from Dylan

Kimberly Lauger © 2015 All Rights Reserved



Overview

This article reviews an educational program that inadvertently led to a reduction in passivity and corresponding increase in self-determination and communication in a young man with CHARGE syndrome. The result? The emergence of the young man—my son—using symbols to express choices, to communicate needs through functional objects, and to have conversations through conventional and non-conventional means about the past, the present, and the future, making choices in his life and acting on them, and enjoying his first true friendship.

With no hearing and large colobomas, Dylan's sensory impairments are severe. Medically his CHARGE syndrome appeared less severe, but over time it has been the subtle neurological differences influencing his behavior, learning, and health that have been the most profound. In spite of focusing on providing access to language from Dylan's diagnosis at only 3 weeks of age, language has remained elusive for him, and of course that influences the rest of his development, with Dylan also meeting the diagnostic criteria for autism.

With Dylan's health and development in a downward slide, Dylan transitioned to a home and community based, but team led educational program for High School¹. At that point, all I wanted was for Dylan to be happy and to feel well again and maybe to gain independence in his Activities of Daily Living.

His itinerant educational team includes a Teacher for the Deaf who is also a Teacher for the Vision Impaired, a Communication Specialist, an Orientation and Mobility Specialist, and an Intervener. In the day to day, Dylan is supported by a full time Intervener and I fill the role of classroom teacher and program administrator.

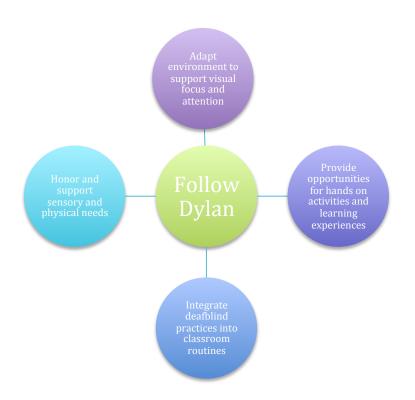
Formal goals for language, choice making etc. were left behind in favor of measuring smiles and laughter, while curriculum was based on Dylan's interests. Yet without changes at home Dylan's behavior there changed as he began to actively use his symbols to communicate a choice, to imitate others, to initiate signs, and to do things for his self.

If smiles and laughter were our focus as we worked with water bottles, balloons, power tools, and a new friend, why did expressive choices and other communicative shifts occur? We'd started with a philosophy and a basic educational model, but what was it in the day to day that made such a difference for Dylan? Was there a unique piece we hadn't yet accounted for? Was there something in our model that could be helpful to others?

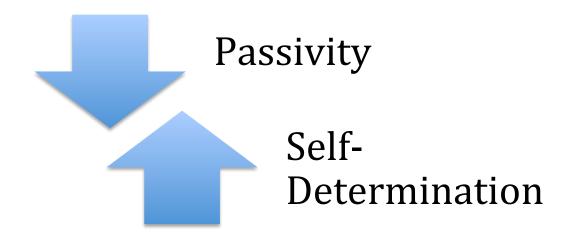
I know Dylan has experienced every element of our educational model throughout his school years. What was the difference at home? Looking back at his first year, I believe the synergistic implementation of the model as a whole with Dylan's responses guiding us inadvertently created a reactive and responsive environment, which led to Dylan's reduced passivity and increased self-determination.

¹ Dylan's program is paid for by the State of Arizona through an Empowerment Scholarship Account.

Figure 1 Dylan's School-Home Educational Model



Create a Reactive and Responsive Environment for



SCHOOL-HOME

I believe in all students having the opportunity to learn with their peers in the least restrictive environment. I also believe in all students having their own needs met. Due to Dylan's health and a desire for a deafblind specific education, his High School Program is at home, yet the model is relevant to all students who are deafblind regardless of placement. To help him differentiate between Middle School and school at home we sign SCHOOL-HOME.

With this handout, I invite you to explore Dylan's SCHOOL-HOME model in more detail, through excerpts from my memoir, *Learning from Dylan*. Although this is Dylan's story, I hope relevance for your child or student will emerge from the pages. Is there a missing piece for your student? Is there something you can increase the frequency of? Is there a cultural roadblock to implementation the team can address? Is the student's spark allowed to shine?

Teaching Philosophy

Emerging from deafblind educational principles and supported by the popular book, *The Spark*, by Kristine Barnett, we started SCHOOL-HOME with the following philosophies and vision.

Teaching Philosophies

Follow Dylan's passion – whatever sparks his curiosity, his enjoyment, and his learning Measure success through smiles and laughter Believe in Dylan's present knowledge and his capacity to learn Honor Dylan's contribution to the world Meet Dylan where he is at and follow his lead Enter his world and then offer a bridge to ours

Vision

We believe creating an Environment specific to Dylan's needs Routines specific to Dylan's needs Adaptations specific to Dylan's needs Lessons and activities specific to Dylan's interests ---

--- will lead to Dylan being able to express what he already knows and provide the opportunity for Dylan to expand his learning, paving the way to greater independence, self-respect, self-determination, enjoyment, and health.

Dylan's team is committed to following Dylan's spark and is excited to see where his spark will take us and what we will all learn along the way.

Educational Model

After 17 years of observing Dylan's responses to people, environment and interventions, and noting the impact on his daily functioning, I had a pretty good idea of what would be needed at SCHOOL-HOME to help Dylan learn to connect, communicate, and learn. Each piece was something I'd learned over the years to help Dylan through his ups and downs with CHARGE.

- A team and a philosophy with Dylan at the center and a willingness to follow his lead
- An environment that was set up to encourage visual focus and attention
- An environment that honored what Dylan's body told us he needed, and a willingness to provide the necessary sensory and physical supports to maintain engaged learning
- Deafblind principles and practices so embedded into our SCHOOL-HOME routine that we didn't even know we were doing them
- Lots of opportunities to learn through hands on activities and experiences

Follow Dylan's Lead

I believe all people learn and function better in an environment where they feel accepted and valued for who they are, where others listen to their point of view, where others honor their contribution, where others think they are capable, where others acknowledge their choices and preferences, and where others do not judge or blame them for their challenges, but offer supports instead. People thrive when they are seen, heard, respected, and understood. It was this type of an environment we wanted to create for Dylan. Of course this happened at school too, but at SCHOOL-HOME this was to be our primary focus.

To accomplish this, our plan was to start with Dylan's interests—water bottles—rather than a specific curriculum. This had proven to be successful for Dylan in earlier years and *The Spark* reinforced this plan. Smiles and laughter were to be our measure of success.

Every day I was prepared with our theme, with activities and materials ready, but it was Dylan who guided the course of the lessons. If I thought too far ahead I would panic, "I don't know what I'm doing. What should we do next?" But in following Dylan's lead, his responses each day guided us to ideas for the next. Should we move onto the next topic or activity, should we repeat a lesson, how could we expand it? These were all questions answered through following Dylan's lead.

Following a child's lead is a common deafblind principle, but I find it is often one of the most difficult to implement consistently. Fears of spoiling or enabling the child, fears of not teaching the child enough, and fears of relinquishing control to the child, have all been expressed.

Peer Pressure, Job Expectations, and Cultural Expectations can also get in the way, although many times the obstacle is really just in our head as we imagine what we think

others expect. There have been two times this Spring when Dylan has not felt well, that I pushed him forward, when everything in me wanted to honor his expression of no. Why? Because I believed it was what the other team member expected or that the other team member was in charge at that time or that I didn't want to be judged as enabling. Turns out they wanted to stop too, but didn't because they thought I wanted to push him on. How often do we disregard a student's communication of "Enough!" because of what we think someone else expects? There is no quick answer to this dilemma, but open respectful communication between team members is essential if the student's input is to be honored.

David Brown says it like this.

"Follow the child is not a recommendation that a child be left to do whatever they like all day without any adult intervention and interaction. Neither does it mean that potentially damaging self-injurious behaviors should be ignored and tolerated. The follow the child approach is primarily concerned with finding out, as quickly as possible, who the child is and how they operate, and in the process starting to build a relationship with minimal aversive episodes. People often think, "What can you do?" is the key question to pose to any child during an assessment, but with this group a better question to begin with would be "What do you do?" I may not ask the child these questions directly, but they will be in my mind as crucially important things that I wish to discover, and they will guide my behavior in a way that I hope will show the child that I am interested in these questions, which demonstrate my respect for them. This approach works, and nobody has ever shown me a better way to start to prepare for effective intervention and teaching. (Brown, 2014)

Set up Physical Environment to Support Visual Focus and Attention²

Through Dylan's Cortical Visual Impairment evaluation, his HANDLE®³ evaluation, and his success in a classroom set up to help the eyes to look and the brain to see, it was clear we would need to mindfully set up Dylan's school room. Sharing the space with my meeting room, meeting the needs of Dylan and my course participants would need to be considered.

Walls

• Contrast – I needed the white board in the front of the classroom for my presentations, but it was too high for Dylan to make use of, was a significant source of glare, and provided poor visual contrast for objects in front of it. To solve this we hung black fabric over it, later replacing it with a curtain that could be drawn back when I needed the board for my work. Sitting at the table in front of this curtain, Dylan had the high contrast visual environment he needed.

² Dylan has no hearing, but if a child does, the environment also needs set up to support the ears listening so the brain can hear. For kids with processing difficulties focusing on either looking or listening at one time can be helpful.

³ HANDLE - Holistic Approach to NeuroDevelopment and Learning Efficiency - www.handle.org

• Visual clutter – Dylan tends to be very aware of his physical environment for navigation, but for schoolwork and for sign language, he seems to pay most attention to what is within arms reach, often not seeming to be aware of what's in the distance. Even so, with blind spots, reduced acuity, and visual processing concerns, minimizing visual clutter - extra stuff on the walls and tables - was an important piece of reducing the effort his eyes needed to make in order to process what he saw. The bulletin Board —painted black by Dylan—has limited postings, which are specific to Dylan's interests for each unit. The art that typically adorns my office walls was moved to walls behind Dylan's workspaces.

Materials

- Slant boards were used for Dylan's schedule, his reading area, and for writing, each board at a different angle to make the specific learning media accessible. Slant board surfaces and Dylan's tables were also black, so his eyes could focus on his work in contrast to the table.
- Dylan uses co-drawn symbols and picture communication symbols, which we place on a black felt surface or on colored card stock with black lines to clearly separate each space. (I still haven't figured out how to get good contrast on his weekly calendar.)
- Computer screens need to have good visual quality and minimal glare. Dylan has done well with an interactive white-board for shared stories and access to the curriculum in other settings. This is one element we did not have as we began SCHOOL-HOME. We tried a regular computer monitor that did not seem to draw his visual attention as effectively as the white-board. Dylan's iPad is nice for visual relaxation. Dylan enjoys scrolling through movies of his school experiences, but the screen is too small for shared reading of literature.

Lighting

Use light to help the eyes see

- Use light to provide a high contrast environment. Think of what it's like to navigate in an unfamiliar dim room compared to one that is well lit. The eyes need light to see.
- Use light to direct the eyes towards a visual target, such as light coming from behind the person towards the book the teacher is holding up in the distance. Think of the spotlight shining on a performer. It needs to light up the performer, but not come between you and the performer.
- Light can also cause glare, which decreases the ability to see. Think of driving into the sun as the light hits your windshield.
- Different types of light impact the ability to see differently for each person. Natural light coming in from outdoors, full spectrum lighting such as from an Ott lamp, different color bulbs, and even the type of the bulb can influence how well people see and how they feel. A beam of light from outdoors, the flicker of fluorescent lights or the intensity of LED lights can distract or even cause distress. Think of what light you see best in and feel best in. Is it the same for others in your home or classroom?

Dylan's eyes are drawn to a light source—and movement—like a moth to a flame. Figuring out how to use light to provide contrast and direct visual attention without causing glare and visual distractions has been a work in progress and sometimes we have to choose visual attention over ideal lighting. Some of our solutions have been:

- Curtains to cover the glass doors behind Dylan's chair to prevent glare
- Close the blinds on the window to prevent visual interest shifting to the rays of light filtering in beside him
- Keep the overhead fan off
- Multiple light sources throughout the room, which caused an impression of the room being lit by a single source, decreasing his tendency to look for that one source, and yet providing enough light for good contrast.

Contrast, clutter, glare, movement, distance and other supports such as placement within visual fields and allowing time for the child to visually process and respond are all helpful in helping the eyes to look and the brain to see. Do these strategies sound familiar? Yes they line up with the ten characteristics of CVI as identified by Dr. Christine Roman-Lantzy. (Roman-Lantzy, 2007)

Why does this matter if your student doesn't have CVI? All brains must process visual input; some are simply more efficient than others. With CHARGE there are enough other interruptions to visual input that it certainly doesn't hurt to adapt the environment to make it easier for the brain to process what visual input does make it through. In a signing environment where language is dependent on processing visual input, these supports can be essential for students with CHARGE. You might find it helps the other students too.

Honor and Support Sensory and Physical Needs

I can only describe Dylan's nervous system at age eight as out of control with night parties, random episodes of running, hitting, kicking, and throwing, numerous tics, etc. Dylan's transformation through an approach called HANDLE®, was life changing for our family. The ability to regulate his sleep, his emotions, his body, and even the use of his eyes dramatically changed the quality of our day-to-day lives. Communicating through actions and finger flicking near his eye, or spinning objects on his nose, he still appeared autistic to others, while I celebrated how engaged and connected with the people around him he was. Able to visually attend to sign language, his receptive language improved. Fascinated by this sensory approach that made behavior seem logical, I studied and became a Certified HANDLE Practitioner. Also an element in Dylan's successes during school, HANDLE would need to be a part of our SCHOOL-HOME routine.

HANDLE is more than HANDLE activities, it is a synergistic coming together of developmental principles, that lead to a way of thinking, a way of looking at human function—behavior—through a sensory lens. It is a way of looking for patterns in actions and responses that might indicate where supports can be provided.

For Dylan, HANDLE Activities done as part of his school routine, help him organize his sensory systems for improved focus and processing. Observing Dylan through my HANDLE eyes, HANDLE also provides a framework for analyzing what supports Dylan needs to reduce the demands on his already challenged sensory systems.

A few HANDLE Principles⁴

• Gentle Enhancement®

From a HANDLE Perspective, incorporating respect for an individual's perceptions, for their tolerance of stimulation, passive and active, internal and external, in order to reduce responses of stress is key. Gentle Enhancement respects the person and his perceptions. (Bluestone, 2004) Following this principle, Dylan's team pauses or shifts in response to any state change – a physiological response that an internal stress response has been triggered. State changes can be a change in breathing, muscle tone, or skin color, complaints or appearance of not feeling well, eyes losing focus, and even red ears. (Bluestone, 2004) Getting wild or silly, running, jumping, spinning, or throwing, or the opposite actions of folding in away from people or going to sleep can also be state changes. State changes simply mean it is time to stop or shift the activity or expectation or increase supports. This fits with my work in deafblindness called *The Box of Deafblindness*© (Lauger, 2013)where we adjust our interactions with a person who is deafblind based on if the person is opening or closing the lid to their box, that is engaging or shutting down.

• Behavior as Communication and Non-judgment.

As I observe Dylan, I trust what his body tells me he needs. I do not judge his function to be good or bad, I seek to understand it. Understanding is what helps me recognize what I can do to help. I might provide sensory supports or shift the environment, the expectation or the activity to help Dylan function more effectively and efficiently—that is with the least amount of stress and the most energy left over.

With underdeveloped semicircular canals—part of the vestibular or balance system—plus limited vision and hearing, people with CHARGE often need time to become horizontal or to have other physical supports that create an awareness of where the body is in space. Lying down or a light pressure to his hips removes the demands on Dylan's vestibular system, freeing his eyes up to focus more efficiently. Dylan will guide us to where he needs this input to stay focused or if he needs to get horizontal.

Without these supports he may get up and leave, may push people away, or get wild, all seemingly "bad" behaviors. For Dylan's team keeping ourselves in a neutral emotional state about the nature of the behavior prevents us from attaching negative intent to it and frees us up to see what Dylan's body is telling us he needs.

⁴ For more about the principles embedded within the HANDLE Approach see http://thehandleinstitute.wildapricot.org/More-about-the-HANDLE-Acronym

It is our effective response to what his body is telling us that allows the lesson to continue without upset.

• Presume Competence

Although SCHOOL-HOME often happens from the floor, I have to admit I love it when Dylan is upright through his entire school day. Why? It lets me know he is feeling well *and* that we have appropriately matched our expectations with his body's ability to meet that expectation. It also raises his level of perceived competence by others.

The reality is Dylan's health is such that he often needs to lie down in order to learn. This does not mean he doesn't have the capacity to learn, it simply means he has more brainpower available for focus and learning when he can take some of the pressure off of his body. Lying down doesn't mean SCHOOL-HOME is over; it means we bring school to the floor. Weighing objects, charting his data, talking about planets, launching rockets, or reading and writing can all happen from the floor. With engaged learning or connection as our goal, we simply adjust to what Dylan's body needs to maintain that connection.

Another area with societal presumptions about competence is based on a person's ability to use formal language to communicate. Many people with CHARGE syndrome and other conditions have difficulty with expressive communication. This does not mean they do not have the capacity to learn or that they "don't know anything".

I tell people Dylan has representations for many things in his head, he just doesn't have words attached to them. I wonder if he did have the words, what complexities of quantum physics he would teach me from all the looking at and creating of wave patterns through his Dasani water bottle and other toys. I did have someone who knows quantum physics come up and tell me Dylan was a genius and was being very intentional with the wave patterns he was making and that it was not just stimming. I have no way of knowing if he is right and Dylan is mapping set points on wave patterns, or his level of intelligence. I'm not that intelligent. I do know Dylan has great capacity for learning and loves science. What matters to me more than any measured level of Dylan's cognition or a formal autism diagnosis is that Dylan is treated as if he has the capacity to learn and something to contribute. With those expectations in place he does both.

Of course not everyone is a HANDLE Practitioner, yet it is possible for any one person to interact from a place of respect and belief in competence and to adapt our expectations, actions, and the environment based on what a child's behavior tells us about what supports they need to continue successfully.

Information about the multiple sensory involvements in CHARGE abounds. Not only can there be vision and hearing loss, but also changes in smell, taste, balance, touch and proprioceptive awareness—the coming together of all of this sensory information that

helps an individual be able to flexibly adapt to the world as it changes around them. With these changes, communication, learning, and behavior can be challenging. Understanding the sensory impact of these challenges can help us learn to "read" the language of behavior and increase our knowledge of how to respond. Finding information from whatever source you can that helps not only explain why these sensory challenges occur, but how to support people with them can be an important element in supporting engaged learning and reducing challenging behaviors.

Integrate Deafblind Practices into Classroom Routines

Teaching deafblind practices at Intervener training for many years, I had some idea of what it would take to teach Dylan at home, but I also knew how difficult it was to put these interventions into practice; to have the environment set up to facilitate attention, to have lessons planned in advance so materials could be prepared, and pre-teaching done, yet to maintain the flexibility to follow the child's lead to a different teachable moment or on a different path to the same outcome. I knew how quickly Dylan lost interest and participation if there were distractions—could I stay focused enough not to be that distraction? Dylan needed a slow pace with lots of wait time, while my mind was always going. Could I move from Dylan's mom and Intervener trainer to Dylan's classroom teacher?

Through my years as an Intervener trainer and a parent advocate I have come to believe that every deafblind strategy is important. Yet thinking of them as individual tools to be pulled from a toolbox as needed is highly inefficient and often times inadequate. For me, too much brain-power is lost if I need to stop and think what tool to use and in that split second of thinking the opportunity can be gone as Dylan's attention is lost. I knew for our program to work we would need to *integrate* deafblind practices into our SCHOOL-HOME routine so seamlessly they would become automatic for me—as well as for him.

There are many resources for learning deafblind principles and practices. My hope is in reading how we implemented them in our program ideas will be generated that will help others who struggle with the inefficiency of thinking of which tool to use when.

Intervener

Leaving school did not mean Dylan lost the opportunity to have an Intervener—a one to one with special training in deafblindness—with him. Dylan is capable of many things, but without an Intervener he does not have access to the information, communication, and relationships that connect him to what is happening in the world around him. Through the process of intervention, Dylan's Intervener facilitates this access. (Alsop, 2012)

Four Components of Intervention

John McGinnis from Canada has identified four main components of intervention. (McGinnis, 1999) Many deafblind principles can be included in these four categories.

• Anticipation

The first time I experienced a deafblind simulation, I was blindfolded with earplugs in and noise cancelling headphones on. Taken by a stranger to an auditorium and made to sit down, I was promptly left alone. What was I expected to do? How long would I be here? Would they come back for me? I could feel the vibration of feet pounding on the stage and someone brushed up against me as they went by. Were we supposed to get up and go on our own? I sat frozen in my seat, too afraid to do the wrong thing, until at last my partner came back, and guided me back to the classroom. How well would you learn in that scenario?

Anticipation is the first piece of the brain's processing a response to a stimulus. Anticipation is necessary before choices can be made. Anticipation increases efficiency of learning. If a person is handed a pencil and a bubble sheet, their brain might think, "Oh, I know what to do with that." Brainpower is then freed up from thinking about what to do with the materials and is instead available for thinking about the answers to the questions. Anticipation reduces stress. For the brain, it's all about survival. Anticipation tells the brain, "Oh I've experienced something like this simulation before. I will be okay."

Schedules and Calendars are ways to keep track of events allowing for anticipation and planning and preparation for the event. Familiar items on the schedule are less stressful than unfamiliar? Have I met that person before? Have I done that activity before? I can recall my daily schedule without looking at it, but I function more efficiently when it is written down, where I can glance over at it, in the midst of business, not having to use unnecessary brainpower to try to recall what's next.

Routines lay the foundation for efficient learning. With repetition the brain's response to a stimulus becomes automatic, which means it can be done without thinking about it. The more elements of a classroom schedule, ritual, or activity are routine, the more energy the brain has to think about the new learning. Dylan's SCHOOL-HOME is filled with routines within routines. To open SCHOOL-HOME, Dylan puts his fidget away, closes the curtains, and turns on the lights. To close SCHOOL-HOME he reverses the process. Each day Dylan does Schedule, Reading, Work, and Writing with cues to represent these taken from his schedule to his visual timer, so he can monitor the passage of time. When Quinn comes over they do four activities. Dylan knows when time is up on the fourth activity SCHOOL-HOME is finished.

Relationships also allow for anticipation. Getting to know someone, becoming familiar with their responses, and their expectations, eases stress as the brain learns, I can trust this person, I know what to do with them, and they will keep me safe.

• Motivation

Motivation is a physiological process and is dependent on anticipation. You have to know what the rocket does before you are motivated to launch it. For new activities the motivation can be pleasing the Intervener or trusting the Intervener will keep them safe. Once familiar the activity itself can be the motivator. A big motivator for Dylan is his friend. He anticipates her arrival and is motivated to do whatever she does.

In the past, finding what motivated Dylan has been a challenge, as he seemed to be uninterested in so many things. In the past even opening presents seemed to be a chore, rather than something that was exciting and motivating. As Dylan unwrapped presents this year, he immediately wanted to open the box and once the paper was off he wanted to get started right away with the science project inside. His Intervener, Deb, and I laughed as we considered the possibility that perhaps Dylan has been motivated by presents all along. We just haven't provided him gifts with enough challenge for his inquiring scientific mind. I have to admit, before SCHOOL-HOME I would not have thought Dylan—who seems to see best an arms distance away—would be fascinated by the Solar System, loving anything to do with planets, and stars, the moon, and the sun, rockets, space shuttles, astronauts and so on.

In SCHOOL-HOME we plan our day two ways. One by us deciding the theme and then pairing it with an activity based on things that motivate Dylan, his likes and interests. The other way is starting with something that Dylan seems motivated by and then us pairing that with language and concepts. So far we haven't run out of ideas, something I was definitely afraid of as we started out.

• Communication

The literature on CHARGE emphasizes the importance of the development of an effective communication system for a child and how important this communication is for the child's ultimate "success." (Hartshorne, 2010) As parents we are of course aware of this importance. I haven't met a parent of a child with sensory impairments yet who is not concerned about how to communicate with their child and who hasn't done his or her very best to help their child learn to communicate. I haven't met an IEP team yet that doesn't have some focus on communication goals.

Parents, educators, and therapists work to help our children learn to express them selves, but in spite of our best efforts, expressive language and/or the pragmatics of language can continue to be a challenge for our children and for us.

We learned American Sign Language when Dylan was a baby thinking this was the best way to provide language right away. Since then we've used touch cues, objects, tactual symbols, real photographs, picture symbols, co-drawn symbols, and tactual and visual ASL and still Dylan functions primarily at a pre-linguistic level. Continuing to use Dylan's co-drawn symbols and experience stories supported with signs, the emergence of Dylan's confidence in him self as a communicator has been one of the most exciting elements of Dylan's SCHOOL-HOME model. And yet communication does not happen in isolation. I believe it is all the elements of Dylan's SCHOOL-HOME educational model utilized in an integrative whole that have contributed to this progress.

Megan is Dylan's Communication Specialist. Yes, she's a Speech-Language Pathologist, but since our goal is not "speech" I prefer Communication Specialist. Megan is not focused on drilling Dylan in repeating signs. She is a master at engaging with Dylan in pre-symbolic language, as she lays the foundation for Dylan learning the foundational elements to build formal language.

What does that mean? It means she uses rhythm, and repetition, and patterns, and turn taking, the shapes of signs, and Dylan's interests to co-write stories and share experiences. Megan has been doing this with Dylan for years before SCHOOL-HOME.

Doe, Dylan's first Intervener, also co-wrote shared stories with Dylan. These were an awesome way for them to communicate in the moment about a shared experience, but Dylan did not show much interest in reading them with another person later.

Deb and I continue to co-write with Dylan everyday incorporating the principles Megan has taught us. Dylan recently has progressed to using co-writing to talk about what is going to happen, what is happening, and what did happen, as well as reading the books with others. If you'd told me a year ago, that I – the person who fails at art – would be drawing with Dylan to communicate, I'd have said, "You're crazy." But now I find I need to have paper and markers everywhere, so I can converse with Dylan, other than at school.

Kristina is Dylan's teacher. She is certified as a Teacher for the Deaf and a Teacher for the Visually Impaired. Kristina works with Dylan from the top down, providing a solid formal American Sign Language model for Dylan.

Kristina read's children's literature with Dylan related to our theme, and creates patterned PowerPoint experience stories to share with Dylan. On Science Saturdays, when Dylan's friend Quinn joins us, Kristina first reads a story with Dylan, modeling the language for Quinn who does not know ASL, and then Quinn reads the story with Dylan. In each reading, Dylan copies the signs made by the reader.

This year, Dylan enjoyed reading The Night Before Christmas for the first time, *and* for the first time signed words such as "reindeer" and "Santa" on the appropriate page, before Kristina modeled it. Reading was definitely not a goal I'd been directly focused on, but here it was.

Ed, Dylan's Orientation and Mobility Specialist, is a child of Deaf parents and a native signer. He works with Kristina in modeling vocabulary and concepts in ASL for the team and is one of Dylan's favorite communication partners. Dylan and Ed have a long history of exploring the world together in a hand under hand model. When Ed shows up, Dylan's hands go up, an invitation for Ed to please talk with me.

Three different approaches to communicating with Dylan; pre-symbolic foundational skills, formal language model, and hand under hand, all three necessary for Dylan to learn not only how to imitate signs, but how to learn how to use language in daily life.

• Confirmation

People need to know their actions are noticed, their voices heard. Dylan can miss subtle acknowledgements, such as a quick nod of the head, or a thumbs up or an ordinary smile. Dylan's expressive communications can also be subtle and are easily missed by others. Without confirmation people may become more passive and stop initiating communication.

Dylan first learned to use the signs for stop and go, while on a horse. As he signed go, the teacher had the horse immediately go. As he signed stop the teacher had the horse stop. He could feel the response of the horse, rather than needing to see or hear the response from a person.

High fives, signing perfect, even signing oops to confirm a mistake are all frequent occurrences in Dylan's day as are exaggerated facial expressions to help Dylan see the response. In a session with Dr. Jan van Dijk, he had participants in the room all raise their hands and wave—Deaf clapping—when Dylan did something. Dylan changed from withdrawn—head down in his chair—to engaged—upright and smiling, looking at what else was going on in the room from this simple exercise.

Of course there are numerous other strategies integrated into our program. Wait time, beginning-middle-end, pacing and so on. When thought of as individual tools, I might think, "Should I use wait time or beginning-middle-end?" for example, when of course Dylan needs both. Together each deafblind principle and practice comprises the sum of deafblind education. With the whole of deafblind education woven into the routines of our day and our interactions with Dylan, he has all the educational supports he needs as a student with no hearing and limited vision.

Provide Opportunities for Hands on Activities and Learning Experiences

Incidental learning is the learning that happens without even thinking about it. The things your brain picks up without even trying to. Most of this information comes in through the eyes and the ears and is limited in individuals who are deafblind.

The typical educational model relies heavily on the incidental information children learn before they ever come to school, and the incidental information they pick up on what is going on in school, to prepare them for secondary learning. That is the information learned by what the teacher tells you. Deafblind learners miss much of that too.

For the typical deafblind learner, they learn best by things they can touch and feel and manipulate. All classes have occasional opportunities for this type of learning experience, particularly with projects and science experiments. The deafblind learner needs this ratio switched. Occasional reliance on incidental information and the majority of the information taught through direct hands on learning experiences. (Alsop, 2012) In addition they need to learn this information in natural environments. To learn about winter, snow, and cold, it is helpful to experience snow and cold rather than to look at snow in a book.

For a student in a general education class the Intervener can help adapt the materials to bring in direct learning as the other students listen to a lecture. At SCHOOL-HOME we have the freedom to create direct learning experiences, using objects, subjects, and activities interesting to Dylan, to teach whatever concepts we plan to teach. Heavy/light, hot/cold, up/down, planets, sun, stars, pretend/real and on it goes. Without needing to transition to the next subject or next class at a set time, Dylan gets to direct the length of the time he needs for an activity. Today in a science project Dylan found interesting, he was able to explore the materials for one hour and forty-five minutes, moving from no prior knowledge to complete mastery.

Follow Dylan

As we've discussed each element, adapt the physical environment to support visual focus and attention, honor and support sensory and physical needs, integrate deafblind practices into classroom routines, and provide opportunities for hands on activities and learning experiences, it is clear that for each element to be successful we must follow Dylan. It is Dylan who lets us know what adaptations need to be made in each area in response to his changing health and sensory processing needs. As physical health improves he may need less supports. Conversely as demands increase he may need more supports. There is a movement among the elements all shifting in response to Dylan.

In spite of knowing this model works for Dylan, at times we find ourselves questioning, when is it honoring and when is it enabling as we follow Dylan's lead. For example, Dylan has had a lot more health problems his second year of SCHOOL-HOME. On days where it appears he is not feeling up to SCHOOL-HOME we offer him the choice of REST-HOME. He's missed so much school this year that sometimes we wonder, is he just out of routine? Are we not doing a good job matching curriculum and activities with his interests? Is he just taking the easy way out and hanging out at home? We analyze what we're doing, problem solve possible solutions and give each other pep talks on trusting Dylan rather than resorting to our previous educational background or cultural expectation or personal belief that says, "Push him." Yet when we follow Dylan's lead the answer is always clear. It is not enabling. When he himself feels up to it he picks the SCHOOL-HOME cue and willingly walks over. If we "overly encourage" Dylan to make it to school, the days are not as productive as he bravely tries to rise to our expectations in spite of not feeling well.

Conversely when he does not feel well, and his choice of REST-HOME is honored, how much does that contribute to Dylan's perception of being respected and understood and his belief that he can make choices in his life and act on them?

Create a responsive reactive environment to decrease passivity and increase self-determination.⁵

As you can see, following Dylan is the heart of this program. It is not about just doing what he wants. It is about trusting the feedback he gives us about what he needs in order to connect, communicate, learn, and I might add contribute. At school or SCHOOL-HOME it is when all of these elements have been implemented together that Dylan has been the most connected, with the most communication, the most learning, and the best health.

Physiologically humans need the perception of control. It is a survival skill. What can I do to make my body feel safe and comfortable in the world? Is this room safe? Can I get out if I need to? Am I comfortable with the lights, the sounds, and the smells? Can I change things if I want to? To feel safe one needs to have the ability to make a choice and to act on it. The more opportunities there are for choice with the ability to influence the environment in the desired way, the more the individual develops a sense of competence, strengthening the sense of self and self-determination. Fewer opportunities for choice lead to a sense of not being capable and passivity and learned helplessness sets in.

I was aware of the dangers of learned helplessness by doing too much for Dylan when he was very young and did my best to minimize this impact and yet Dylan was very different in his responses and in how quickly he "gave up" from a very early age. Even as a baby Dylan rarely even cried and certainly not in a way that seemed to be "for attention."

I recall one time trying to wait him out so he would pull his pants up by himself. The pants were pulled up to the level of his thighs as he walked around the house with his knees pulled together to keep them from falling down—the ultimate sagger. This went on for hours with no indication that it bothered Dylan as he waddled around the house or that he had any interest in if I was "noticing."

One time of my rushing and putting his shoes on for him as I tried to get the other four children out the door and to school on time—instead of waiting the 30 minutes it took Dylan—and it was like he'd never learned how to do it himself.

Reactive or responsive environment

⁵ Directive or passive environment

An environment where most things are done for the child or to the child with deafblindness with few opportunities for choice making, problem solving, communication or interaction.

An environment that provides opportunities for the child to make choices, solve problems, communicate, and develop a sense of self.

http://www.sparkle.usu.edu/glossary/index.asp?cat=int

Typically, once a skill is mastered humans resist someone taking that skill over. Imagine a child pushing you away as they say, "I can do it myself." or a person hesitant to give up the keys to the car as they age. These responses are typical from a self-determined individual. Dylan clearly did not respond that way through much of his life, he certainly does now.

As the first year of SCHOOL-HOME progressed and we celebrated the smiles and laughter and the opportunity for Dylan to express what he knew—holding the basket as he set the brick in on that side of the scale, perhaps thinking, "Yes, I know it is heavy and I don't want the basket to crash down," —life at home changed too. Dylan changed.

We have used picture symbols since preschool. He understands the symbols receptively, but in spite of our efforts at school and home, he did not use them expressively to make a choice or to tell us his preference. His home Intervener continued to use the same symbol system, when suddenly one day he just started pulling off the symbol of what he wanted to do and handing it to her. Certainly not something he did when we attempted a reward system to get him to hand over a cue years earlier. It had to come from within. Do you get the power of that for him? *At 17 he was finally able to tell us through a symbol, not an action, what his preference was.*

He began to be bothered if we wanted to hurry him along. I imagine him saying, "Chill out Mom, I've got this."

We began to see the initiation of signs—again something he has been exposed to since infancy, but never used expressively. I love how he gets out of the car, an average of 15 minutes to get his glasses on just right, adjust his hat and shoes, and prepare his body to move. Once out of the car he turns and closes the door, signing "CLOSE" and then "FINISHED," before turning to walk towards the house, opening and closing the gate to the courtyard and then the door to the house, and finally heading to his playroom where he puts his hat and shoes and glasses away, all on his own.

Dylan's bed has a rail. He doesn't need the rail anymore, but it is part of his routine. If we don't put it up, he grabs our hands and puts them on the rail to indicate put it up. We tell him "Good night. I love you. See you in the morning." then turn off the light and close the door. A few nights, as I was going to bed, I saw light under his door. I wondered if I'd forgotten to turn the light off. Then one night I heard a thump. Rushing to Dylan's room, there was Dylan sitting on the floor where he'd fallen coming over the rail—his night time feeding still connected to his G-tube—as he was sneaking out of bed to turn the light on. For 17 years he had slept in the dark. *For how much of that time had he wanted the light on before he finally did something about it?*

Following our SCHOOL-HOME routine, with Dylan's cues, schedule, co-drawing, and signs for communication, focusing on water bottle mobiles, sprinklers, boats, and stick-horses, and filling and pouring, and weighing them, as well as the concepts and language that went along with them, our focus was on measuring smiles and laughter and improving health. Without intending to set up a reactive, responsive environment, that is what we had done. As the amount of time Dylan had direct, meaningful control of his world increased, Dylan's passivity went down and his self-determination went up. And with that shift the entire world is now open to him.

Summary

Science supports the view of control as essential for survival and a normal adaptive function.⁶ HANDLE principles purport that the greater the vulnerabilities a person has the greater the need for control they have. But grown ups don't like children to be in control, it makes us feel out of control. And so we respond in strong ways, often attempting to take away more control and choice, only to find situations escalating out of hand. I propose instead that increasing opportunities for choice and control might decrease undesired behaviors as well as supporting engaged learning.

Dylan's educational model suggests by following the child's lead, setting up the environment so the eyes can look and the brain can see, honoring and supporting sensory and physical needs, integrating deafblind practices into classroom routines, and providing opportunities for concrete learning experiences, a reactive responsive environment is formed, leading to reduced passivity and increased self-determination—that is the ability to make choices and exercise control over one's own life.

⁶ Born to Choose: The Origins and Value of the Need for Control. "Belief in one's ability to exert control over the environment and to produce desired results is essential for an individual's well being. To choose is to express a preference, and to assert the self. Each choice – no matter how small – reinforces the perception of control and self-efficacy. The presence or absence of control has a profound impact on the regulation of emotion, cognition, and physiology. If people did not believe they were capable of successfully producing desired results, there would be very little incentive to face even the slightest challenge. Opportunities to exercise control may be necessary to foster self- efficacy beliefs. Individuals with little experience of acting as an effective agent will likely have little belief in their ability to produce desired results leading to feelings of helplessness and depression. Individuals who do not perceive control over their environments may seek to gain control in any way possible, potentially engaging in maladaptive behaviors. Lack of control over the environment is believed to be a major cause of the abnormal stereotypic behaviors, failure to thrive and impaired reproduction commonly observed in animals raised in captivity." Lauren A Leotti et al. (Leotti, 2010)

A Day in SCHOOL-HOME

From the house to the school, he walked across the courtyard, opening and closing doors behind him. Entering school he put his fidget away, closed the curtain, turned on the lights, then sat in his chair for the morning schedule. Deb and I proudly smiling in the background as Dylan did the entire open SCHOOL-HOME routine on his own.

After writing today's schedule, Dylan and Deb went on to review the week, pointing to the co-drawn picture cues and talking about Dylan's experiences. YESTERDAY, MEGAN, BOOKS-READ. – Oh how he'd read them, seven co-drawn books about rockets and planets —books he'd written with Deb or me—that for the first time he was able to enjoy reading with someone who hadn't been there. TOMORROW, ED, VISIT, WATER-BOTTLE-MOBILE, GIVE. He got stuck here, signing, ED, ED, ED. 'SATURDAY, WHO?' Deb asked. Dylan pointed—yes, pointed—to the Quinn cue and fingerspelled her name, more indications of Dylan's emerging skills as a reader and he'd just answered a question.

Next in Dylan's SCHOOL-HOME routine is reading. Dylan walked toward the computer where we had a storm story up on screen, but then turned towards the window, looking first to me and then to Deb and then towards the window. At first I thought he was confused if he should sit next to me or next to Deb, but no, he was drawing our attention to the co-drawn stories on the shelf next to me. With Deb's and my jaws hitting the floor, Dylan picked up his writing board with the stories on it and took it over to the writing table. We thought he might want to read these stories again, like he'd done with Megan yesterday, instead of reading the computer story, but no he placed these stories on the table, reached behind and got out a piece of paper, ready to write a new story. I guess writing was going to happen before reading and work today.

Three things about this tiny scene were completely out of the ordinary. First was Dylan breaking the school day routine to do something else. Up to this point the only times Dylan initiated a change to the routine would be when he got up and went to rest on the floor in the activity room. Second was Joint Attention. Dylan using his eyes to draw our attention to his writing board—letting us know he was thinking about writing —was a completely novel experience to me. And if that wasn't amazing enough, Dylan picking up his writing board and moving it himself, certainly was. For 18 years, Dylan has acted on people and objects to get what he wants. Instead of going to get his own writing board he would have taken my hand and placed it on the writing board, then nudged my hand in the direction of the table. Without a doubt we were going to Follow Dylan's lead and write, but what should we write about? The lesson planned for the day, of course.

Bringing over the new science project—a tornado in a bottle—Dylan and Deb co-drew what it was and how to do it. Pouring water from a pre-filled pitcher into the bottle, the water spun in tornado fashion, but with clear water in a clear container could Dylan see it? Offering Dylan a box of food coloring, he knew right what to do. Reaching in and selecting red, Dylan added it to the water. Ahh, that was better, but was it enough? Placing a black board behind the bottle, creating contrast and getting rid of distraction, Dylan was

mesmerized watching the water go around and around. Adding objects, Dylan watched as they were drawn into the tornado.

Then we were on to dumping the water out, filling it from water in the sink, and bringing it back to the table, choosing a new color, learning to turn it on —himself—not by guiding our hand to do it. Back and forth pour, fill, spin, pour, fill, spin, Dylan doing more and more on his own with each repetition. Two hands holding tight, eyes looking at the top, carrying the full container back to the table. Lid off, color in, turning it on, hands on the outside, feeling it move, look, spin, around and around. Grab the paper, hurry, let's co-write what I saw.

Again and again, one hour and 45 minutes of the same project. With each repetition came another opportunity for Dylan's scientific exploration. What color to use? How much food coloring to add to get just the right color to see the spin the best? Which objects spun the best? Which objects got stuck? Learning how to do each step, ending with total independence from start to finish on the project. And with all the repetitions of co-drawing he asked for as he worked, for the first time taking the marker from Deb, Dylan completed the picture, filling in the water spinning in the bottle, without hand-under-hand support.

As Dylan worked, I watched and cried, giddy with excitement, celebrating the emergence of Dylan as a self-determined individual—an equal participant in his education. I celebrated the freedom of being able to honor Dylan's request to write, and the freedom to allow Dylan the amount of time on the project he wanted. I celebrated the emergence of his ability to read and his new love of writing. But mostly I celebrated Dylan, loving the opportunity to see him happy and eager and engaged with learning.

Lights off, curtains open, Dylan walking back across the courtyard with fidgets in hand, I slid exhausted to the floor, my body feeling as limp as a noodle, as hidden tension from years of searching for answers left me.



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Thursday, July 30, 2015 Breakout Session #2: Utopia C-D 2:15-4:30pm

Identifying Effective Positive Behavioral Supports for Young Adults with CHARGE Syndrome

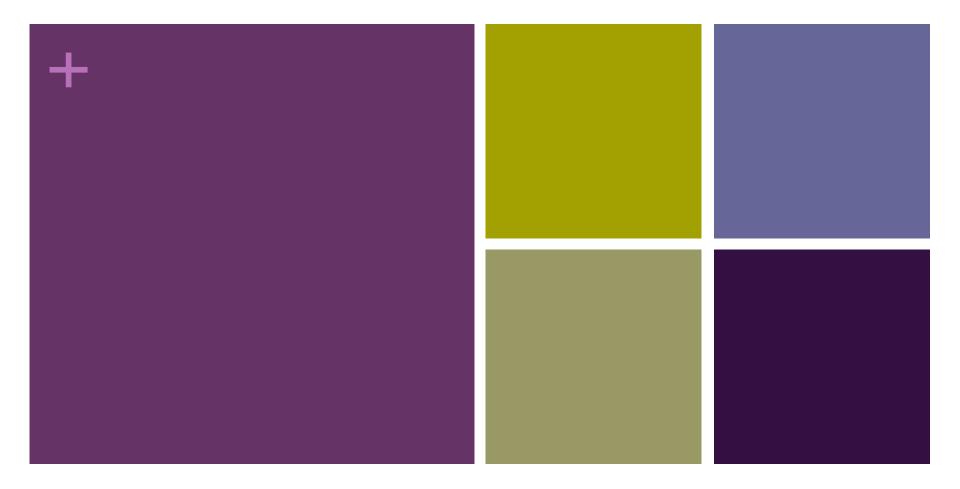
Susan M. Bashinski, Ed.D., Associate Professor, Special Education, Missouri Western State University Susan M. Bruce, Ph.D., Professor, Special Education, Boston College

Presenter Information:

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

Presentation Abstract:

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



Strategies to Support Positive Behaviors in Students with CHARGE Syndrome

Susan M. Bashinski, Ed.D., Missouri Western State University Susan M. Bruce, Ph.D., Boston College Review of Literature: PBIS

Positive Behavior Intervention Supports apply behavioral principles to support emotional regulation

Learners with CHARGE syndrome:

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

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

Review of Literature (con't.)

n PBIS is proactive rather than just reactive

n PBIS looks beyond the learner

- n Considers sensory integration needs
- n Considers environment
- In Uses information from Functional Behavior Assessment (FBA)on purposes of behavior to build the Positive Behavior Intervention Support Plan
- n Cites alternative/replacement behaviors that must be taught (such as communication) (Horner, 2000)

Action Research

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

(Bruce & Pine, 2010; Hendricks, 2009)

Research Questions

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

Young Adult Student Participants

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



Our study focused on three areas of PBIS:

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

Data Sources

- n Individual behavior charts (3 focus areas)
- Classroom journals (for teachers and paraprofessionals to record ideas about each student's behavior)
- **n** Videotaped observations that captured proactive and reactive strategies (transcribed)
- n Ongoing development of a Positive Behavior
 Intervention Support Profile for each learner.
 (Completed profiles were member checked by teachers)

Data Analysis

n Constant comparative, elements of grounded theory

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

(McHatton, 2009)

Findings 8 Major Themes/Super-Categories

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

<u>Theme #2</u>: Support students to cope with anxiety

- n Structure (Theme #1) helps
- n Keep students informed
- **n** Gross motor exercise, rough & tumble play (3 students)
- n Calm spaces

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- **n** Consider influence of sensory sensitivities (Theme #3)
- n Support students to learn & identify their individual strategies:
 - Gail: Use of mature behavior sheet, turning down hearing aids when overwhelmed, establishing physical distance from irritations

Support students - cope with anxiety (con't.)

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

Support students - cope with anxiety (con't.)

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

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

- **n** Behavior = Repetitive talk about Wizard of Oz
- **n** Staff must be aware of manipulations to get into such conversations
 - n She might say "Call me Dorothy"
 - n She might say "Do you like shoes?" Who wears red shoes?" This leads to Dorothy conversation.
 - n She might say, "I like yellow." This leads to talking about the yellow brick road.

n Adult use of language: Staff talk about real/not real

Theme #8: Adult Language Supports Positive Behavior

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

Adult Language Supports Positive Behavior (con't.)

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

Adult Language Supports Positive Behavior (con't.)

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

Journal Excerpts on Adult Language Supports with **Gail**

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

Discussion

n Common needs (such as defined and organized spaces, sensory sensitivities)

n Unique needs-to each student

- n Must know each student well-their likes / dislikeslevel of detailed knowledge held by teachersamazing
- Adults could act in specific ways to prevent negative behaviors

Discussion

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

Study Limitations

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

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Thursday, July 30, 2015 Breakout Session #2: Utopia C-D 2:15-4:30pm

What shall professionals know – and how can we teach them in a sustainable way for working with people with CHARGE Syndrome?

Dr. Andrea Wanka, stiftung st. franziskus heiligenbronn, Germany

Presenter Information:

Andrea is a teacher for the deafblind who finished her PhD on early interaction and communication between parents and their children with CHARGE Syndrome in 2011. Since then she has been working in a large centre for the blind, deaf and deafblind and is responsible for children, youths and adults who are deafblind. For Andrea, the national and international networking is very important to share knowledge and learn from each other. This made us in the EU build up a network of professionals working in the field of deafblindness called "PropäK".

Presentation Abstract:

An international working group is developing a staff training program. One module in this program is on CHARGE syndrome. The main focus of the working group is based on the selection of the topics (separated into learning areas) with simultaneous considerations of new learning theories (like principles of teaching based on neurology) to make sure that staff gets sustainable knowledge while taking part in the program. Contents and the way of imparting will be presented.



Thursday, July 30, 2015 Breakout Session #2: Utopia C-D 2:15-4:30pm

Making it Happen: Intervener Services For Children and Young Adults Who Have Vision and Hearing Losses

Linda Alsop, M.Ed., SKI-HI Institute, Center for Persons with Disabilities, Utah State University and Beth Kennedy, M.Ed., DeafBlind Central: Michigan's Training & Resource Project

Presenter Information:

Beth Kennedy has worked in the field of deafblindness for twenty-five years. She worked at Perkins School for the Blind, in the Deafblind Department, and graduated from the teacher training program at Boston College before taking a job consulting for the Florida deafblind project. Beth is currently the Director of DB Central: Michigan's Training & Resource Project and teaches American Sign Language at Central Michigan University. While she addresses a variety of topics as a consultant, she has worked on and supported many teams involving a person who has CHARGE Syndrome. She is currently leading a team to develop her fourth Open Hands, Open Access (OHOA) intervener training module for the National Center on Deaf-Blindness (NCDB), and has made notable progress in increasing the number of credentialed interveners working in Michigan.

Linda Alsop is the Director of Deafblind Programs at the SKI-HI Institute/Center for Persons with Disabilities at Utah State University in Logan, Utah. She has extensive experience working directly with children with deafblindness (including those with CHARGE) and their families. She developed the SPARKLE model of parent training and resources in deafblindness, and numerous service programs, curriculum, and training materials that are being used by families, educators, interveners, and others around the country. She acts as a national consultant in deafblindness, and is actively involved in national advocacy efforts related to intervener practices in the United States. She developed and implemented the first online higher education training program in the country that trains interveners to work with children and youth who are deafblind and prepares them to receive the National Intervener Credential. She is actively involved in efforts to establish interveners as a professional discipline that is recognized as a related service under IDEA.

Presentation Abstract:

Interveners are individuals who work one-to-one with children and youth who are deafblind and who have training and specialized skills in deafblindness. The practice of using interveners as individualized supports for children with combined vision and hearing loss in educational settings is growing across the country, and this is creating an ongoing need to provide information and training to parents about the critical role that interveners play in the education of their children. You know that an intervener is what your child/student needs- but how do you make it happen? This presentation will provide parents and other members of the team with important information for making the case for intervener services.

Making it Happen: Intervener Services for Students Who Are Deaf-Blind

Interveners are for people who are Deaf-Blind:

- People who have combined vision and losses may qualify as "Deaf-Blind."
- Deafblindness is a disability of access- access to visual and auditory information about people and things in the environment.
- Students who are Deaf-Blind must have direct, hands-on experiences in order to learn. Learning information and/or skills secondarily or incidentally is difficult to impossible.
- The unique learning and support needs of children who are Deaf-Blind pose a challenge to most educational settings because they are generally not designed to provide constant direct, hands-on learning.

Interveners have training and a specific role:

- An intervener is a person who: 1. has training and specialized skills in deafblindness and 2. works consistently one-to-one with a child who is deafblind.
- The role of the intervener is to: 1. facilitate access to environmental information usually gained through vision and hearing, 2. facilitate the development and/or use of receptive and or expressive communication skills, 3. develop and maintain a trusting, interactive relationship that can promote social and emotional wellbeing.
- The National Intervener Task Force formed on January 17, 2002. The work of the Task Force includes: 1. collecting data and disseminating information on child impact (of intervention), 2. raising awareness about interveners and advocating for intervention, 3. impacting systems change, 4. providing parent education and encouraging parent involvement, and 5. advocating for intervener training.
- Interveners can now take coursework through an online program through Utah State University, complete a practicum, and compile a portfolio in order to receive the National Intervener Credential from the National Resource Center for Paraeducators. The competencies for interveners were approved by the Council for Exceptional Children in 2008.
- Interveners formed the National Intervener Association (NIA) to: 1. promote quality intervener services, 2. recognition of interveners on local, state, and national levels, 3. promote awareness of the impact an intervener can have for a student who is deaf-blind, 4. provide support for interveners in the field.
- There are now approximately 103 interveners who have completed the USU coursework, and about 70 credentialed interveners across the United States.
- In Michigan, there are 18 students who have the support of an intervener. Six of those interveners hold the national credential, three more are completing

portfolios, and nine others are currently taking or have completed the coursework.

Advocating for an intervener:

- Your state's deaf-blind project may be able to assist. To find the project in your state, and to access additional resources on interveners/intervention, visit <u>www.nationaldb.org</u>.
- Deafblindness is a disability of access. While interveners are not yet included in IDEA, the mandates afforded by the Americans with Disabilities Act (ADA) regarding access may be helpful.
- House Resolution 4040, the Alice Cogswell and Anne Sullivan-Macy Act, includes intervener services. To learn more visit <u>https://www.congress.gov/bill/113th-congress/house-bill/4040</u>.

More information is available:

- To learn more, visit <u>www.intervener.org</u>.
- Find them on Facebook.



Expressive Communication Skills of Children with CHARGE Syndrome

Alexandria Cook and Charity Rowland, Ph.D. Oregon Health and Science University

Presenter Information:

Alexandria Cook graduated from Reed College with a B.A. in psychology and is currently a research assistant in the Design to Learn Lab at Oregon Health & Science University (OHSU). Her research interests include motivational and neurological development of children and adolescents of all developmental capabilities, including risk taking and the transition to adulthood. She offers support and research expertise to the Communication Matrix and the associated Community of Practice. Through these projects she has begun to mine the extensive data available in the Communication Matrix database on the communication skills of individuals who experience rare disorders.

Dr. Charity Rowland directs the Design to Learn Projects at Oregon Health & Science University in Portland, OR. Trained in developmental and experimental psychology, she has conducted extensive research on communication development in individuals with complex communication needs. She is the author of the Communication Matrix. The Communication Matrix assessment has been used by parents and professionals to document the many ways through which individuals with CHARGE Syndrome and other rare disorders can communicate.

Presentation Abstract:

The Communication Matrix is an assessment tool for children and adults with complex communication needs that is used world-wide to show how individuals at the earliest stages of communication development express themselves. Data entered into the website (<u>www.communicationmatrix.org</u>) are mined to provide detailed information about behaviors used to communicate and messages expressed. Over 169 children with CHARGE Syndrome are represented in the associated database. Extensive information on their communication skills is summarized in this poster.



Common Psychotropic medications used in CHARGE syndrome and what we know about them

Claire Latus-Kennedy and Timothy S. Hartshorne, Ph.D. Central Michigan University CHARGE Syndrome Research Lab

Presenter Information:

Claire is from Grand Rapids, Michigan and a second semester freshman at Central Michigan University, majoring in Neuroscience and Psychology and minoring in Communication Disorders. At CMU she is a member of the CHARGE Syndrome Research Lab and has been looking at psychotropic medications and their use with children who have CHARGE syndrome.

Presentation Abstract:

This poster will provide information regarding psychotropic medications that are often used with children who have CHARGE syndrome. The poster will include information about the drug, its side effects, and what research supports its use with children.



"Wow, that Sounds Familiar": Parent-Child Playgroups for Families of Children with Deaf-blindness

Julie Maier, California Deaf-Blind Services

Presenter Information:

Julie Maier is an educational specialist with California Deaf-Blind Services (CDBS) and provides technical assistance to children and youth with deaf-blindness (including CHARGE Syndrome), their families, and educational teams. Julie is also a faculty member in the SFSU Department of Special Education and has served as a research assistant, course instructor, and fieldwork supervisor in the Moderate-Severe Disabilities program since 1999. Julie entered the field in 1987 teaching individuals with deaf-blindness in community-based adult program and later in inclusive schools in Berkeley, CA. She has authored and co-authored many published writings on the topics of collaborative teaming, social supports for students who use AAC, self-determination, and capacity building perspectives and practices. She has presented at local, state, and national conferences on topics related to natural supports, inclusive education, family partnerships, collaborative teaming, and ability awareness. Julie feels very fortunate to have been trained and mentored over the past several years by David Brown, her former colleague at CDBS.

Presentation Abstract:

Julie will present the components and results of a two-year collaboration between CDBS and a local early intervention program to implement biweekly playgroup meetings with parents and their children with deaf-blindness. Facilitators provided information, modeling and support to a diverse group of families with children with unique developmental profiles and support needs in an environment that allowed for discussion, demonstration and practice of new skills. Details about the implementation and outcomes of the playgroup sessions will be shared.

"Wow, that Sounds Familiar": Parent-Child Playgroups to Train, Support, & Build Connections

What? A collaboration between the deaf hard of hearing early intervention staff at the Center for Early Intervention on Deafness (CEID) in Berkeley, CA and staff from California Deaf-Blind Services (CDBS) in San Francisco Bay Area to lead playgroups for children who are deaf-blind and their parents as means to provide parents with information, coaching and support related to the unique needs of their children.

When? The first and third Thursday of every month from 3-5 pm since Sept. 2013

Where? The toddler classroom at CEID and occasionally at community locations such as a park, a local farm, a children's museum, therapeutic horse riding facility.





Who participated? Up to six families attended early on, including three children with CHARGE, and four families continue to attend. Our youngest child joined us at 6 months and the oldest was close to 5 years old at the time. One father and 4 mothers consistently attend the playgroup.



Why? Providing early intervention and support to families of infants and young children who are deafblind is critically important. This population requires the expertise of early interventionists, including both a hearing and vision specialists, as well as adults and community members who are deaf-blind and can provide insights into living with deaf-blindness. One of the greatest challenges in the field of education for young children with deaf-blindness is reaching families to provide information specific to deaf-blindness in a way that is respectful of and accessible to the needs of families. With the collaborative involvement between CEID staff and CDBS staff, these CEID playgroups helped parents of children with deafblindness learn different types of communication and language techniques including tactile and co-active signing, sensory processing interventions, methods for maximizing visual and auditory abilities, handunder-hand support and how to provide multisensory input, among many other things.

How? CEID applied for and was awarded a two-year state early start grant that covered any necessary transportation costs, cost of materials for art projects and take-home projects, paid for one support staff to assist during playgroup. CEID provided time for early intervention specialist prepare for and to facilitate group meetings. CDBS staff prepared written resources and articles and attended at least one session per month.



A typical playgroup looked this:

- Arrival and casual checking in
- **Greetings Circle** that included using children's photos mounted in tactile photo frames that each family made and a greetings song done at the children's pace.
- While children played or were held in parents' laps or on a blanket with toys the CEID and CDBS staff led an interactive discussion about that week's topic. Strategies were shared, stories were told, and questions were asked.
- Group activity at the table or on the rug, such as a hands-on art project, sensory
 project, creating experience books. Most weeks we also began to spend time with
 service dog from CEID, which was a favorite activity for some children. During the
 group activity the discussion on the week's topic and other concerns continued as
 CEID and CDBS staff modeled strategies and techniques, such hand-under-hand
 support, follow the child, use of touch or object cues, encouraging use of child's
 functional vision, etc.
- Goodbye circle and song and staff gave parents a take-home packet with more information, ideas and materials. Each packet included information about the week's topic and announcements of upcoming playgroup events and other fun family events happening in the local area; a short article about the topic for parents who wanted to learn more; and a "homework" assignment which was a suggested activity for the parents to do together with their child to practice the skills and concepts we had discussed that week (e.g. make a story bag with tactile objects for a favorite bedtime story, make scented bubble bath soap, select object cues for several
- During the second year we began to meet in community settings for **field trips** once every month or two and also invited guest speakers to come to our some of our meetings (e.g. adults with deaf-blindness, parents of older children with deaf-

blindness). Parents had told us at the end of the first year that these were two things they would like to add to our sessions.

Some topics we've covered

O Greetings and name signs

daily home routines)

- O Establishing and following routines
- O Follow the Child
- O Concept Development
- O Encouraging use of functional vision
- O The Other Senses
- O Literacy and experience books
- O IFSP and IEP process and advocacy

- O Using play to promote interactions
- ${\rm O}$ Interveners
- O Planning family outings
- O Self-regulation
- O Including your child in holiday traditions and family gatherings
- O Self-determination
- O Family resiliency and leadership



Outcomes We Had Hoped For

- Increased knowledge of the range of the impact of deaf-blindness on communication and social, emotional, and cognitive development.
- Opportunity to provide individualized coaching and support to families.
- Parents increased use of the skills and strategies demonstrated and suggested by facilitators.
- Parents increased confidence in sharing information about deaf-blindness with others.
- · Parents increased knowledge of local resources and support services.

What We Learned

- Knowledgeable, informed parents are POWERFUL parents.
- Parents were eager to receive information and did use the practices we demonstrated.
- Parents had a lot to share about their child and their family's story with the playgroup facilitators and each other and seemed to bond over their common experiences and challenges.
- We were able to individualize the information and strategies we shared because the group was a manageable size and the consistency of attendance by several families.
- As months passed, parents began to advocate more. First for additional information, next for resources that would allow them to advocate more effectively for their child with local support and funding agencies and educational programs.
- The parents appeared to appreciate and value what other parents had to share and recognized each other as another important source for support and resources.
- Collaboration among early intervention service providers is fruitful and makes an impact.

What we heard from parents



We have learned from the playgroup that with a child with special needs you need to think outside of the box to do normal everyday things. Get creative and have fun. We have had so much fun going on field trips to the petting zoo and the local park. It helped teach us how to get Mackenzie more involved in each experience. During the holidays class we learned about how to get your relatives to interact with our child. Family is very important to us and we want everyone to connect with Mackenzie. The tips we learned in class have made our family closer. Our most memorable class was when we had a guess speaker, Ms. Haben Girma. She gave us hope that Mackenzie can do or be anything that she wants to do in life. Mackenzie is already an independent little girl and I look forward to where she will go. We give credit to the training we have received from the deaf-blind playgroup for her development. We look forward to going to the playgroup to see what tricks of the trade we will learn next. The bonds that we have developed with the other parents are great. We know that we have someone to bounce ideas off of and that they would be there to listen to us and understand where we are coming from. ~Shilo, Mackenzie's mom

"Initially, I didn't know what to expect from the deaf blind playgroup at CEID. I was hoping it would give me some "Mommy and Hank" time. It has certainly done that and more. It has become a small social network and support group as well a resource for invaluable information and techniques that can help my son now and in the future. Also, Hank's favorite part is spending time with Nan, the service dog at CEID. It's so wonderful to see him smile around Nan. I also enjoy our field trips exploring the amenities throughout the Bay Area that are available and accessible to Hank. I'm very grateful for the playgroup and look forward to it every couple of weeks." ~ Julie, Hanks' mom





I have learned so much about how to help Isabel learn, grow, and develop from her teacher, Kimberly, and all the people who contribute to the group--from the folks at CDBS to the guide dog Nan to the translators in the classroom--this group has made a significant difference in our lives. As typically developing children learn so much from sight and sound, figuring out how to help my child who wears glasses and hearing aides was daunting. Isabel is six and while she still can't speak she is learning to communicate and interact with others from the help with the playgroup. One of the things I really appreciate about the playgroup is that it is an effective combination of learning and fun. Kimberly always sets up a theme for the month and gives us "homework" projects to carry the lessons home. This helps me share learning with Isabel's Dad who can't attend the group. It also enables me to further think about how to help Isabel. One of the great things we talked about was creating an experience book about everyday things or a special event to help our children understand what they are going to do or what they have done. I have created them not only for Isabel but also for a child who is also in her special needs aguatic program--something I would have never known how to do except for this playgroup. We also do fun play dates, such as going to Roberts Regional Park, which has a barrier free playground. We have future plans to attend an equine group so our kids can interact with ponies. I appreciate being able to do such outings with other special needs children as it makes me feel not so isolated or different.

And this leads to one of the most important things about the playgroup: support from other parents. Having a special needs child with multiple disabilities and medical needs is hard. I love my daughter with all my heart, but there are times when it feels overwhelming. Being able to talk with other parents and share both the pain and joy of raising our children is incredibly helpful to me. Plus we have fun with new friends! The deaf-blind playgroup has truly helped my daughter and me. From learning, playing, and developing, Isabel and I have grown a lot from being a part of this group.



Parental Decision Making in the Removal of Gastrostomy Tubes

Rachel Malta & Timothy S. Hartshorne, Ph.D., Central Michigan University CHARGE Syndrome Research Lab

Presenter Information:

Rachel is a second-year specialist student in the School Psychology graduate program at Central Michigan University. Additionally, she works with Dr. Tim Hartshorne as part of the CHARGE Syndrome Research Lab and is constantly amazed at how much more there is to learn about CHARGE syndrome and how these incredible individuals overcome many challenges. Her current research is focused on how parents make medical decisions for their child with CHARGE syndrome, specifically the decisions regarding the removal of g-tubes.

Presentation Abstract:

Feeding issues for individuals with CHARGE syndrome are frequently treated using a gastrostomy tube to bypass oral feeding and maintain adequate nutrition. However, as oral feeding increases over time, parents often make the decision to remove their child's tube. Because of the complexity of the process, parents need support and accurate information from medical professionals and the CHARGE community to ensure they are empowered to make the best decision for their child.

Parental Decision Making in the Removal of Gastrostomy Tubes or Buttons

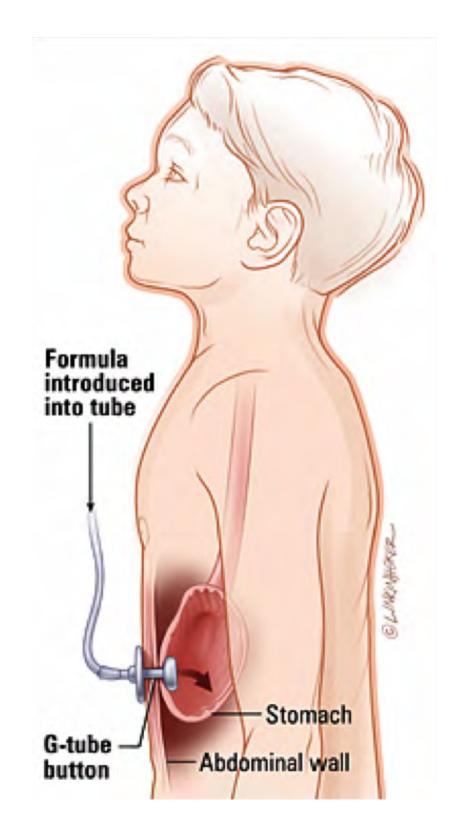
Rachel Malta & Tim Hartshorne, Ph.D. • Central Michigan University

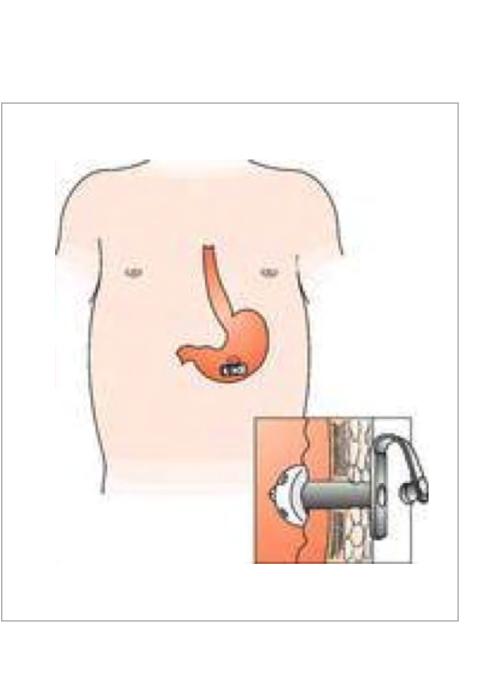
Parental Decision-Making in Medical Situations

- There are a number of studies which look at factors impacting parent decision-making for children with a variety of medical conditions. Results show:
- Parents often gain support, discern values, and obtain guidance from professionals, family members, and members of the community through online support groups and forums (Jackson, Cheater, & Reid, 2008).
- Parents desire a role in the decision-making process alongside the professionals involved and prefer to have their autonomy respected (Lipstein et al., 2012)
- Three common themes for making an informed decision emerged across studies (Jackson, Cheater, & Reid, 2008):
 - A need for information
 - Control over the process
 - Connection with others in comparable situations
- Despite 200 studies cited in various meta-analyses, only 3 looked at gastrostomy tube placement. None of the studies examined the removal of tubes.

What are Gastrostomy Tubes (g-tube) and Buttons?

- A gastrostomy tube is an apparatus that is surgically inserted through an opening in the abdomen directly into the stomach. Food, liquids, and medications can be fed directly to the individual through this tube, bypassing the entire feeding by mouth/swallow process.
- In the event of long-term feeding through a feeding tube, a button may be inserted into the opening where a tube was placed. The button is closer to the body and can be opened and closed for easier feedings.
- The button is designed so that there is no external tube consistently attached to the individual, lessening the chances that it will be caught on clothing or disturbed during play.





G-tubes and buttons provide a means for directly providing food, liquids and medicines directly into a patient's stomach. These medical devices allow for the bypassing of the processes of feeding by mouth and swallowing when either of these is too difficult or dangerous.

Advantages of G-Tube/Button

- Post insertion, parents reported a reduction in vomiting, less stressful meal times, better parent-child communication, and adequate weight gain (Avitsland et al., 2013)
- Bypass of swallowing leads to reduction or elimination of aspiration (inhalation of food or fluids into the lungs) which can lead to aspiration pneumonia and difficulty breathing. (Åvitsland et al., 2013)

Disadvantages of G-Tube/Button

- Potential for irritation surrounding the insertion site (Goldberg et al., 2010)
- Leakage surrounding the stoma, or opening in the abdomen (Goldberg et al., 2010)
- Parent reports indicated a potential for infection at the stoma (Goldberg et al., 2010)
- Can be pulled out or dislodged during play or behavioral episodes

Mothers and Feeding Tubes

Why is this topic important?

- others around them.

Research Questions

- the G-tube?
- opinions?
- successful attempt?

Emotions also play a part in the difficulty surrounding the decisions involving gastrostomy tubes/buttons (Guerriere, McKeever, Llewellyn-Thomas, & Berall, 2003).

According to Brotherton & Abbott (2012), because food and liquids are needed to sustain life and mothers are considered the primary caregiver in families:

• Feelings of guilt and inadequacy may occur when they view their inability to feed their child "normally" as a failure, leading to a delay in the initial insertion and acceptance of the g-tube for their child

 Mothers may have feelings of loss and grief when unable to breastfeed and bond with their child the way they expected over feeding times

Because feeding by mouth and swallowing are common difficulties for individuals with CHARGE Syndrome, it is crucial to find a way to sustain nourishment/nutrition.

Parents of individuals with CHARGE face many decisions in the treatment and care of their child, yet there is often conflicting advice and information from professionals and

While there is information in the medical communities as to why the G-tube and/or button is important and what surrounds the insertion of the tube, there are not always clear decision points established for their removal.

Knowing who and what parents consult for information regarding these decisions would help to clarify the process for future parents or those who may make this decision.

Standard steps of the process (common professionals that helped or were consulted, any steps necessary for transition/preparation of removal)?

What sources do parents consult when deciding to remove

Does social media play a large role in the decision? What percentage of parents used social media to gather advice/

Approximately how many parents choose to remove the G-tube/button against medical advice and how many of these attempts were successful?

Are there any common characteristics (sex, age, severity of symptoms) that appear to increase the likelihood of a

Qualitatively, were there any common events that prompted the decision to remove?

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The Brain in CHARGE

Mandy Odren & Timothy S. Hartshorne, Ph.D., Central Michigan University CHARGE Syndrome Research Lab

Presenter Information:

Mandy Odren is an undergraduate student studying neuroscience and psychology at Central Michigan University. Her interest in CHARGE syndrome research stems from working with Dr. Tim Hartshorne's son. She is a senior planning to attend a physician assistant program after graduation.

Presentation Abstract:

This poster provides a brief overview of the literature on the brain anomalies seen in CHARGE syndrome and considers potential future research for a better understating of the neurophysiology associated with CHARGE syndrome.



College of Humanities & Social & Behavioral Sciences

Department of Psychology

CHARGE lab

THE CNS IN CHARGE SYNDROME

Mandy Odren and Tim Hartshorne Central Michigan University

Cranial Nerves

- I. Olfactory Mild hyposmia to complete anosmia
- II. Optic Vision issues
- III. Oculomotor Vision issues
- IV. Trochlear Vision issues
- V. Trigeminal Chewing and swallowing issues, headaches
- •• VI. Abducens Vision issues
- VII. Facial Facial palsy, increased salivation
- •• VIII. Vestibulocochlear Vestibular issues, sensorineural hearing loss
- IX. Glossopharyngeal Swallowing issues
- X. Vagus Swallowing issues, abdominal headaches
- XI. Accessory Shoulder and neck problems
- XII. Hypoglossal Tongue problems

Blake KD, et al. 2008. Cranial nerve manifestation in CHARGE syndrome. Am J Med Genet Part A 146A:585-592

From the Literature

- Cortex

- Cerebral/cortical atrophy
- Dysgenesis of the frontal lobes
- Gyral abnormalities
- Eissencephaly
- Nodular heterotopias
- Ventricles
 - ventriculomegaly
 - ¤ Hydrocephalus
 - ventricular dilation
- Anterior pituitary hypoplasia
- Hippocampal hypoplasia
- Olfactory system
 - Arhinencephaly
 - Hypoplasia of olfactory bulbs
- Craniosynostosis

- Hemorrhagic/ischemic lesions
- Basioccipital hypoplasia and basilar invagination
- Midline defects
 - Holoprosencephaly
 - Corpus callosum agenesis
 - ¤ Meningoencephalocele
 - a Agenesis of the septum pellucidum
- Posterior fossa
 - cerebellar (vermis) hypoplasia
 - Dandy-Walker malformation
 - cerebellar heterotopias
- Brainstem

 - Brainstem hypotrophy

Problems with the Literature

- Central Nervous System Malformations in the CHARGE Association. A. Lin, et al.
 - ∝ Done in 1990
 - Dut of 144 patients only 47 (33%) had examinations of the brain
 - n 30 postmortem examinations
 - n 17 CT scans
 - ¤ Before CDH7 gene was found

From the Netherlands

- Cortex

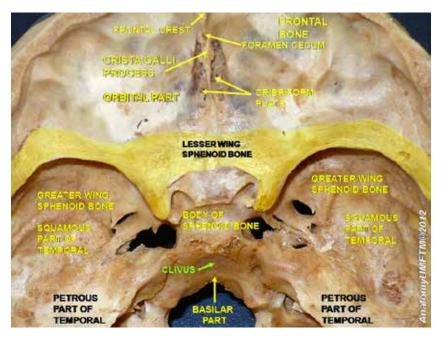
- Cerebral/cortical atrophy
- Dysgenesis of the frontal lobes
- a Gyral abnormalities
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- Ventricles
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 - ¤ Hydrocephalus
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- ·· Anterior pituitary hypoplasia
- Hippocampal hypoplasia
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- -- Craniosynostosis
- Hemorrhagic/ischemic lesions

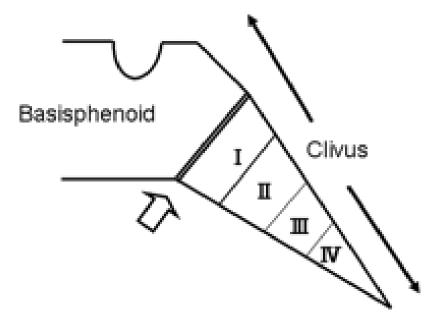
- Basioccipital hypoplasia and basilar invagination
- Midline defects
 - ➡ Holoprosencephaly
 - Partial corpus callosum agenesis
 - <u>
 ∞ Meningoencephalocele</u>
- Posterior fossa
 - cerebellar (vermis) hypoplasia
 - Dandy-Walker malformation
 - **¤** Foliation defects
- Brainstem
 - *∞* Agenesis of the 7th nucleus
 - Brainstem hypotrophy

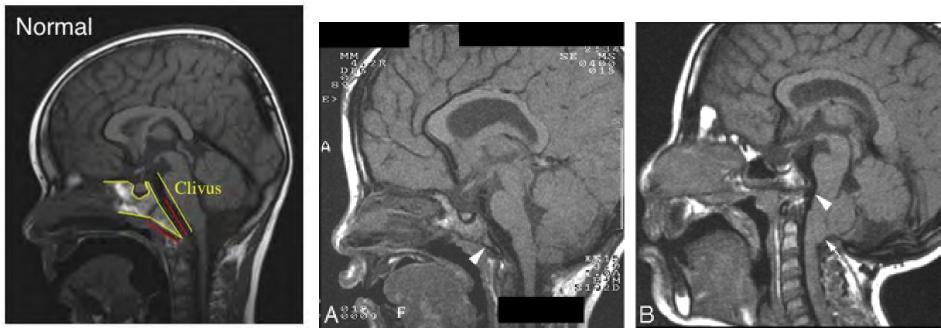
De Geus, C. Neurology in CHARGE syndrome. 2015 [powerpoint slides].

The Clivus in CHARGE

- How it started
 - Fujita et al., Abnormal basiocciput development in CHARGE syndrome. AJNR Am J Neurorad 2009;30(3):629-34
- What is the clivus?
- Clivus abnormalities
 - ¤ Clinical relevance
 - Associated anomalies in CHARGE
 - Small, malformed clivus
 - n Basilar invagination
 - n Chiari I malformation with syringomyelia
- Conclusion
 - ¤ 91% abnormal clivus (size and/or morphology)
 - Clivus anomalies has a diagnostic value







Fujita et al., Abnormal basiocciput development in CHARGE syndrome. AJNR Am J Neurorad 2009;30(3):629-34

Current Research

- Balance

- ¤ Cerebellum and ataxia
 - SARA/pediatric balance scale
 - Reevaluation of MRI
- Cerebellum and foliation defects
- Myopathic phenotype
 - n "Sloping shoulders"
 - <mark>n</mark> Goal
 - n Challenges
 - n Plans

Sources

- Basson, et al. Epistatic interactions between Chd7 and Fgf8 during cerebellar development: Implications for CHARGE syndrome. *Rare Diseases.* 2014; E28688-E28688.
- -- Brandsma et al., Ataxia rating scales are age-dependent in healthy children, Dev Med Child Neurol. 2014 Jun;56(6):556-63.
- Blake KD, et al. 2008. Cranial nerve manifestation in CHARGE syndrome. Am J Med Genet Part A 146A:585-592
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- Yu et al., Deregulated FGF and homeotic gene expression underlies cerebellar vermis hypoplasia in CHARGE syndrome. Elife, Vol. 2, e01305, 24.12.2013.



The Development of Play in CHARGE Syndrome

Megan Schmittel, M.S. and Timothy S. Hartshorne, Ph.D., Central Michigan University- CHARGE Syndrome Research Lab

Presenter Information:

Megan is a student in the School Psychology Doctoral Program at Central Michigan University who currently works with Dr. Tim Hartshorne. She is a member of the CHARGE Syndrome Research Lab and is interested in the 'B' in CHARGE Syndrome. Her research is specifically focused on the development of social play in children with CHARGE Syndrome.

Presentation Abstract:

Play is a fundamental aspect of development. As children age they progress through different stages of play supporting their social, emotional, and cognitive development. Many factors affect play and the way a child progresses through play. Conditions that may inhibit the development of play include sensory impairment, motor problems, communication delays and cognitive impairment. Studying play in individuals with CHARGE can help to develop interventions for these children to aid in their development.

The Development of Play in CHARGE Syndrome

Megan Schmittel and Timothy S. Hartshorne, Ph.D. • Central Michigan University



Why is play important?

- Aids in cognitive development (Fisher, 1992).
- Allows practice of social skills (Fisher, 1992).
- Allows practice of self-regulation strategies (Barnett, 1984).

Why study the development of play in children with CHARGE Syndrome?

- Determine effects of play on overall functioning
- Help develop interventions to aid in overall development

Impact of Physical Restriction on Play

- Those with a physical impairment engage in restricted play.
- Those with gross motor physical impairments spend less time in group play, social play, and outdoor play and more time in transition, looking on, and non-play (Kennedy-Behr, Rodger, & Mickan, 2013)

Impact of Hearing Impairment on Play

 Those with a hearing impairment experience the same stages of play as typically developing peers, but the amount of time spent in particular stages is varied compared to hearing peers (Spencer & Meadow-Orlans, 2004).



- At 12 months, hearing impaired children spend less time in representational play (Spencer & Meadow-Orlans, 2004).
- At 9 and 18 months children with a hearing impairment engage in similar play as hearing peers (Spencer & Meadow-Orlans, 2004).
- Children with a hearing impairment engage in less symbolic play in earlier stages of life than hearing peers, but go on to engage in the same amount of symbolic play at later ages (Spencer & Meadow-Orlans, 2004).
- Children with a hearing impairment engage in pretend play in terms of developmental stage rather than chronological age, so those with a hearing impairment may engage in play stages at later ages compared to hearing peers (Morelock, Brown, & Morrissey, 2003; Brown, Rickards, & Bartoli, 2001).

Impact of Vision Impairment on Play

- Those with a vision impairment engage in less risky play (Celeste, 2006).
- Those with a vision impairment have difficulty seeing play, which inhibits the use of modeling to teach play (Celeste, 2006).
- Children with a vision impairment progress through same stages of play, but at later ages (Ferguson & Buultejens, 1995; Troster & Brambring, 1994).
- Children with a vision impairment spend more time in the exploratory stage of object play (Troster & Brambring, 1994).
- Children with a vision impairment use more symbolic use of language rather than objects in symbolic play (Ferguson & Buultegjens, 1995).
- Children with a vision impairment engage in more solitary play (Celeste, 2006).
- The level of vision loss affects interaction with peers (Skellenger, Rosenblum, & Jager, 1997).

Impact of Cognitive Impairment on Play

- Children with a cognitive impairment engage in more solitary play (Leach, Pratt, & Roberts, 1990).
- Children with a cognitive impairment experience similar development of symbolic play as typically developing peers. (Wing et al., 1977).



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CHARGE Syndrome & Characteristics of Autism Spectrum Disorder: Examining Similarities and Differences

Hallie Smith, M.S., Doctoral Candidate, School Psychology, MacKenzie Sidwell, M.S., Specialist Student, School Psychology, and Kasee Stratton, Ph.D., NCSP, Assistant Professor, Licensed Psychologist, School Psychology, Mississippi State University

Presenter Information:

Hallie Smith and MacKenzie Sidwell are graduate students in School Psychology at Mississippi State University and members of the Bulldog CHARGE Syndrome Research Lab. Ms. Smith and Ms. Sidwell have previously worked closely with children with Autism Spectrum Disorder prior to beginning research on CHARGE Syndrome in 2013 under the direction of Dr. Kasee Stratton, assistant professor of school psychology, licensed psychologist, and CHARGE researcher. Ms. Smith and Ms. Sidwell are currently completing a research study investigating the similarities and differences between Autism and CHARGE.

Presentation Abstract:

Do individuals with CHARGE syndrome also have Autism Spectrum Disorder (ASD)? Understanding the similarities and differences between CHARGE and ASD can be complex for parents and professionals. We will feature the similarities, the differences, and the challenges with assessment (both educational and psychological) for Autism among individuals with CHARGE. Further, preliminary results from a current research study on CHARGE and ASD will be presented.



How do we raise awareness for CHARGE? A study of Facebook and Twitter Outcomes

Daniel L. Gadke, Ph.D., NCSP, BCBA, Assistant Professor, Hailey Ripple, B.S., Doctoral Candidate, School Psychology, Somya Mohanty, Ph.D., Assistant Research Professor, Arthur Cosby, Ph.D., William L. Giles Distinguished Professor, Mississippi State University

Presenter Information:

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She currently runs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome*, the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Stratton specializes in challenging behavior and improving adaptive skills.

Presentation Abstract:

The world of social media offers a unique outlet for raising awareness of CHARGE Syndrome. Our study involved the use of a Social Media Tracking and Analysis System (SMTAS) in a big data laboratory to collect data on Twitter "tweets" on CHARGE Syndrome. Twitter Awareness data will be presented along with current Facebook trends. Strategies to increase CHARGE awareness will be included.



The Bulldog CHARGE Syndrome Research Lab at Mississippi State University

Kasee Stratton, Ph.D., NCSP, Hailey Ripple, MacKenzie Sidwell, Hallie Smith, Mady Sully, Reeva Morton & Dominik Keller, Students, Mississippi State University

Presenter Information:

Dr. Kasee Stratton is an assistant professor of school psychology at Mississippi State University. She is also a licensed psychologist and nationally certified school psychologist. She currently runs the Bulldog CHARGE Syndrome Research Lab at MSU. Dr. Stratton has been researching and presenting about CHARGE since 2005. She is an author of two chapters in the book, *CHARGE Syndrome,* the developer of the CHARGE Non-Vocal Pain Assessment, and has presented in the U.S., Australia, New Zealand, and Denmark on CHARGE. Dr. Stratton specializes in challenging behavior and improving adaptive skills.

Presentation Abstract:

The Bulldog CHARGE Syndrome Research Lab at Mississippi State University conducts research and helps to spread awareness of CHARGE. The lab is composed of undergraduate students and graduate students in school psychology. The lab is under the direction of Dr. Kasee Stratton. Our goal is to investigate areas that are important to families, professionals, and individuals with CHARGE. Additionally, the lab seeks to create a diverse group of advocates and future professionals who are well informed of CHARGE Syndrome.



Take Time For Yoga

Rebekka Valian Teacher of the Deafblind, W. Ross Macdonald School for the Blind, Deafblind Unit

Presenter Information:

Rebekka Valian is a teacher of students with deafblindness, some with CHARGE Syndrome. Rebekka is also a yoga teacher so combines her passions when teaching Adapted Yoga for Persons with Deafblindness.

Presentation Abstract:

Creating time and space for yoga can be invaluable to your student's growth. Adapting the class for your individual student's preferences and strengths can be easy if you keep a few guidelines in mind, if you are a yoga teacher as well. I'd like to share the film showing one such class. My yogi is a young man, sixteen years old, with CHARGE. He is in a school for students with deafblindness. Watch the magic unfold.



The role of CHD7 in differentiation of neural progenitor cells

Hui Yao, Ph.D, Donna M. Martin, M.D., Ph.D Departments of Human Genetics and Pediatrics, The University of Michigan, Ann Arbor

Presenter Information:

Dr Yao is a postdoctoral research fellow in the laboratory of Dr. Martin in Pediatrics and Human Genetics. She is interested in studying stem cells to explore underlying mechanisms of neuronal developmental disorders, including CHARGE syndrome.

Dr. Martin is Associate Professor of Pediatrics and Human Genetics at the University of Michigan. In addition to working with children and families affected by numerous genetic disorders, including CHARGE syndrome, Dr. Martin devotes a significant amount of time to the study of CHD7 and related genes implicated in CHARGE pathogenesis. Her laboratory has developed several mouse models of Chd7 deficiency used to study CHARGE syndrome by investigators worldwide.

Presentation Abstract:

CHD7 encodes a chromodomain helicase DNA-binding protein that is mutated in CHARGE Syndrome. *CHD7* is highly expressed in developing human and mouse embryos, and in progenitor cells that are affected in CHARGE. Our laboratory reported that loss of *Chd7* impairs proliferation, self-renewal and neuronal potential of mouse brain-derived neural stem cells, suggesting a mechanism for the neurological and sensory defects in CHARGE. We will present recent data exploring CHD7 function in the differentiation of neuronal cells from human neural progenitor cells. Results from this study will help us identify the underlying signaling pathways involved in CHD7-mediated neuronal differentiation, and may help guide the design of regenerative therapies for individuals with CHARGE.



Genetic Testing in Patients Suspected of Having CHARGE Syndrome

Cheryl Scacheri, MS, LGC GeneDx, Inc. Chris Lauricella, MS, CGC; Toni Lewis, MS; Rebecca Fowler, MS, CGC; Jennifer Siegel, MS, LCGC; Jane Juusola, PhD, FACMG; Sherri Bale, PhD, FACMG

Presenter Information:

Cheryl Scacheri has been in the genetics field for 23 years and has worked for the genetic testing company, GeneDx, for seven of those years. She earned her master's degree in genetic counseling from the University of Pittsburgh and bachelor's degree from Drew University in New Jersey. In 2005, she attended the CHARGE syndrome conference with her husband, Peter Scacheri, PhD, where both were excited to hear Dr. Connie van Ravenswaaij present about the discovery of the CHD7 gene. Cheryl spends most of her efforts in genetics education, something which she is very passionate about. Her expertise is in genetic testing and exome sequencing for rare disorders.

Presentation Abstract:

Genetic testing for patients with features of CHARGE syndrome may provide helpful information to families and health care providers. Our laboratory has analyzed the CHD7 gene in over 1,000 samples that were referred for sequence analysis and/or deletion and duplication analysis. We have also performed whole exome sequencing (WES) on samples from several patients with some features of CHARGE syndrome. In some of these cases, CHD7 mutations were identified. In others, however, other genes were likely to be the underlying cause of their CHARGE-like features. Aside from CHD7, thus far, no single gene appears to have a recurring association with CHARGE syndrome. However, the diverse genetic diagnoses provided by WES may be helpful to some patients, particularly those who are negative for CHD7 mutations and present with atypical findings.



CSCDP: CHARGE Syndrome Clinical Database Project - Fun Facts

Meg Hefner, MS, Kevin Ballard MCS MHI, Joanne D'souza BS, Saint Louis University

Presenter Information:

Meg is a genetic counselor with more than 30 years' experience with CHARGE syndrome, as a founding Board member of the CHARGE Syndrome Foundation, writer and editor of the Management Manual for Parents, American Journal of Medical Genetics special issue on CHARGE syndrome, the CHARGE Syndrome book (Plural Publishing) and many other lay and professional publications. She is a recipient of one of the first Stars in CHARGE award and has presented at every International CHARGE Syndrome Conference.

Presentation Abstract:

This poster will discuss the CHARGE Syndrome Clinical Database Project project and illustrate some of the data collected. One of the authors (Kevin Ballard) will sign people up and get them started entering information in the database at conference.



Advice from siblings of persons with CHARGE

Hayley Hoesch & Timothy S. Hartshorne, Ph.D. Central Michigan University CHARGE Syndrome Research Lab

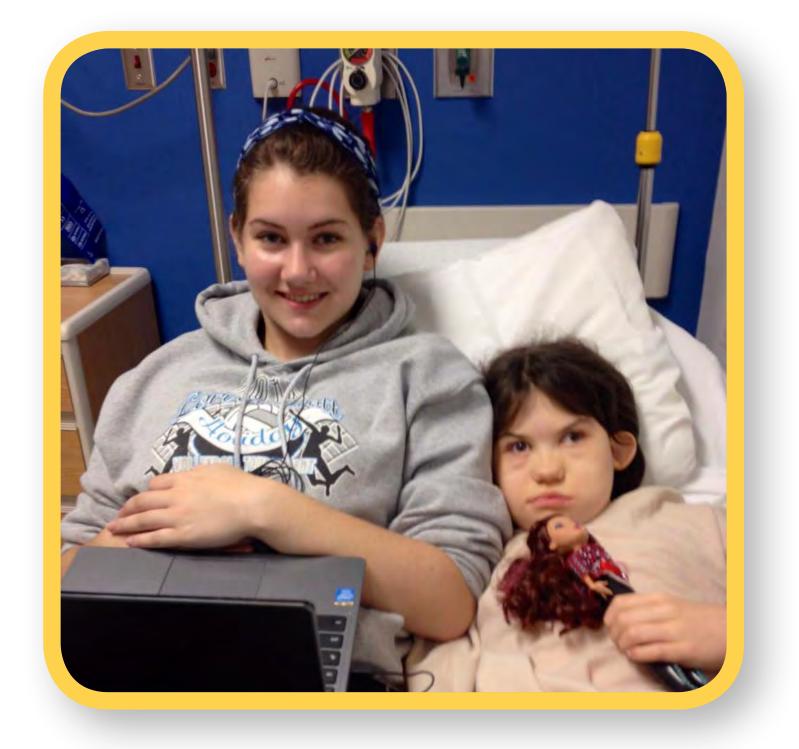
Presenter Information:

Hayley is a research assistant in the Central Michigan University CHARGE Lab headed by Timothy S. Hartshorne. Her research interests are centered around the social aspects of the lives of children with CHARGE, specifically the relationships between siblings.

Presentation Abstract:

Research conducted by the CHARGE Syndrome Research Lab on the experience of siblings of children with CHARGE included an open ended question "What advice would you give to others who have a sibling with CHARGE?" We have compiled the answers and present them on this poster.

Advice from siblings of persons with CHARGE to others who have a sibling with CHARGE



Be positive. Don't be depressed, because it's not a bad thing to have a sibling with CHARGE.

Be aware of their problems and fears, goals, and aspirations. Feel free to tell people about your sibling and what holds them back in life.

Don't be afraid to bring your friends around. In most cases they are interested more so than scared. If you act at ease, so will those around you.

Never stop trying to create a relationship with your **sibling.** It will be hard and frustrating at times, but he/ she is a part of your family forever.

Learn sign language. Try to find something you can do together like video games or rock climbing. Don't let your parents treat siblings differently.

Have better communication skills e.g., sign language.

Go with the flow. Enjoy them while you have them there.

Don't get mad at them because they may not understand that they are doing something wrong. The worst thing you can do is neglect your sibling who has CHARGE. I have found it very rewarding to become close to my sibling with CHARGE. He is my world and I think about him all the time.

Be patient, understanding, and put yourself in their shoes.

Be happy with who you are! Be happy with what you have! Be happy for your brother or sister that has the chance to change people <u>because</u> of <u>who they</u> are. CHARGE doesn't define them. It's just a part of their life.

Just keep goin'.



Learn about CHARGE, learn to talk about it with others, and embrace the positive effects that having a sibling with CHARGE has on your life.

Take it day by day. Remember they are still a wonderful, smart, and energetic part of the family. They don't express it the same way we do, but they understand and can teach us a lot! Our family has grown 10X closer since my brother was born.

Accept who they are, not what they're not, the more you understand them, the more you grow to love them.



Try not to be embarrassed by them - it wasn't their choice to be born with CHARGE. **Try** not to feel isolated from others or become withdrawn. Love them unconditionally.

Help out as much as you can and take responsibility.

They might be embarrassing sometimes and people might stare, but oh well.

Feeling bad/selfish for having a life disability free doesn't get you anywhere; if they're happy, be happy for them!

Some days will be awesome, and other days the crap will hit the fan. Just take everything one step at a time, and you'll do fine.

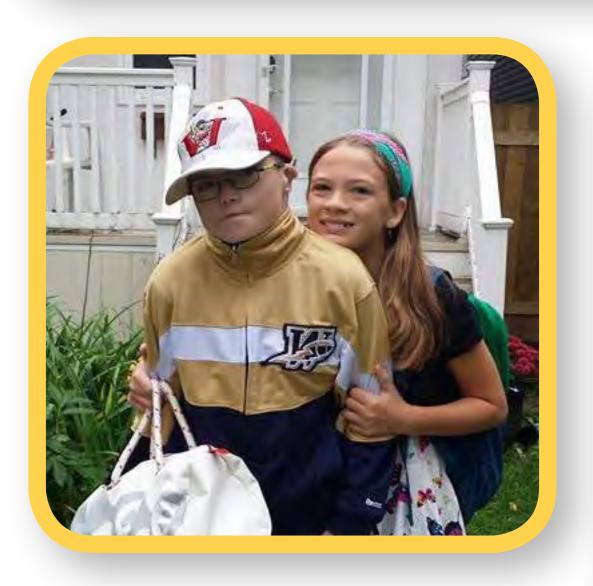
Be their friend.

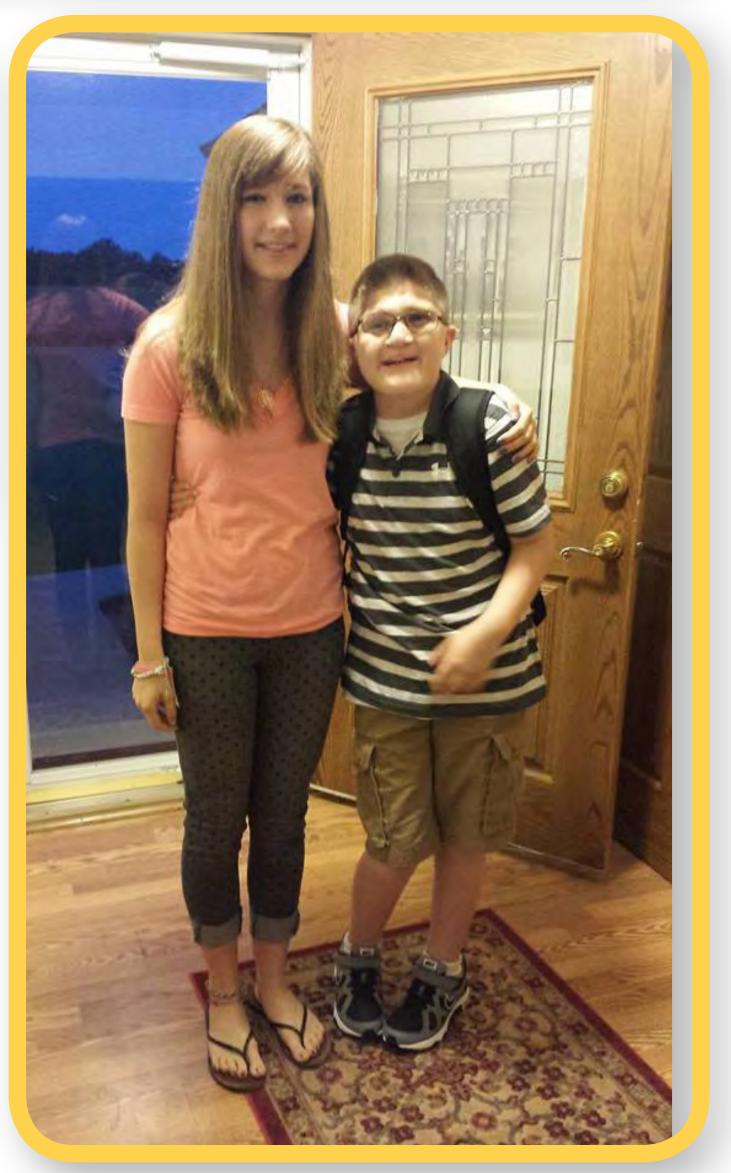
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Hayley Hoesch & Timothy Hartshorne **Central Michigan University**





Embrace the journey. I was a senior in high school when my youngest brother was born. I missed a lot of his early development. But in 1996, I had a child with a genetic syndrome and what experiences I had as a sibling truly helped prepare me to be the mother of a child with disabilities. Life is GOOD! :)





Role of CHD7 in nerve formation

Fumiaki Imamura, PhD., Pennsylvania State University

Presenter Information:

Fumiaki Imamura, is an expert neuroscientist, and has a long-standing interest in the molecular mechanisms regulating brain development. Especially, his study is focused on development of the olfactory system. CHARGE syndrome patients often show olfactory dysfunctions. It is considered that impaired CHD7 function in developing olfactory tissues results in abnormal development of the olfactory system. He is currently studying the role of CHD7 in brain development, especially formation of neural circuit formation, by focusing on the olfactory system. His current research aims to identify molecules regulated by CHD7 in developing brain. His long-term career goal is to translate my research results and expertise in a way that will benefit CHARGE syndrome patients. He hopes to develop my research results into drug discovery for CHARGE syndrome patients by finding the molecular targets and methods for the treatment of disrupted neural circuits in the future.

Presentation Abstract:

Abnormal nerve formation in the brain is part of the prominent defects in CHARGE syndrome patients. Given that disruptions of neuronal circuit cannot be surgically treated, what is desired is the molecular targets that can prevent/fix the disruption. I will present my ongoing research project aiming to identify molecules whose expression is regulated by CHD7 in developing neural circuit in the brain.



Camp Abilities Brockport: A Counselor's Experience

Gretchen Imel, Tim Hartshorne, PhD., Central Michigan University CHARGE Syndrome Research Lab and Lauren J. Lieberman Ph.D

Presenter Information:

Gretchen is an undergraduate student at Central Michigan University who works with Dr. Tim Hartshorne as a member of the CHARGE Syndrome Research Lab. She is working towards a Bachelor's of Science degree with a double major in Therapeutic Recreation and Psychology. Therefore, she is interested in researching adapted recreation for individuals with sensory impairments along with recreational inclusion strategies so individuals with such impairments can participate in any activity that they wish. She is also a caregiver/intervener for Jacob Hartshorne, an individual diagnosed with CHARGE syndrome.

She has the amazing opportunity this summer to volunteer as a counselor at Camp Abilities in Brockport, New York from June 28th to July 4th. From this experience she hopes to support parents who have a child diagnosed with CHARGE syndrome in finding the resources necessary to promote physical activity among individuals with sensory impairments.

Presentation Abstract:

Recreation and leisure activities are a critical dimension of the quality of life for all people, a vital component of personal expression and interaction that can open paths to adventure, confidence, and health. However, some children have never run a mile, batted a ball, jumped off a diving board, or been asked to play in a soccer game – all because they have some sensory impairment. Nonetheless, almost any sport can be modified using technology or adaptive equipment to accommodate participation at any ability level



CHARGE Syndrome & Parental Relationships

Dominik Keller, Mady Sully, & Kasee Stratton, Ph.D., NCSP, Mississippi State University

Presenter Information:

Dominik Keller and Mady Sully are undergraduate and graduate students, respectively, at Mississippi State University and members of the Bulldog CHARGE Syndrome Research Lab. Ms. Keller and Ms. Sully are currently interested in the family relationship and family dynamics for individuals with CHARGE. Their research was spurred by multiple parents asking about the divorce/separation rate among families of a child with CHARGE. Their investigation is under the direction of Dr. Kasee Stratton, assistant professor of school psychology, licensed psychologist, and CHARGE researcher.

Presentation Abstract:

It is documented in the research literature that parents of a child with a disability often experience similar divorce/separation trends to the general population; however, families with a child with Autism experience an increase in divorce/separation. Researchers in CHARGE are frequently asked about the divorce/separation rate among families of a child with CHARGE; however, they are left to speculation, as this has never been investigated. Our poster will present literature related to divorce and disability and results of a preliminary study on this topic as it relates to CHARGE.



Assessing attachment in mother-child relationships in rare syndrome contexts

Nicole Vian, Milano-Bicocca University, Department of Human Sciences for Education and Health Sciences Department, School of Medicine

Presenter Information:

Nicole Vian, Milano-Bicocca University, Department of Human Sciences for Education and Health Sciences Department, School of Medicine

Presentation Abstract:

The role of attachment proves to be one of the mediation factors in the relationship between child trauma and the overcoming of the trauma itself. The short and long term effects of traumatic experiences linked to numerous hospitalizations, as lived outside the family, should be mediated by the mental models developed by the subject in relation to her/his own attachment relationships with adults caregivers. Thus it is important to highlight the mother--child attachment patterns in rare syndrome contexts, so as to implement intervention plans aimed at increasing mothers' awareness of their caring style and at improving mother--child dyadic relationship.

Nicole Vian

Professor Adjunt, PhD Student University of Milano Bicocca, PhD Student, University of Milano-Bicocca Department of Human Science for Education "R. Massa", Piazza Ateneo Nuovo 1, Milano, Italy Nicole Vian, Corso Cairoli 32, 28921 Verbania VB Italy nicole.vian@gmail.com nicole.vian@unimib.it mobile: 0039.3937130187

Professor of Italian Sign Language (*LIS*) at University of Milan Bicocca, Department of Human Sciences for Education "Riccardo Massa". Support teacher specialist for handicapped psychophysical graduate to Catholic University in Milan. Worked as a special education teacher from 2005 to 2010 in a Public School with a special method in Milan: Primary School "Rinnovata Pizzigoni"; support teacher at Primary School and interpreter of Italian Sign Language . Trainer of Italian Sign Language interpreters and technical aids and visual and hearing impairment related .

In this field, former professor at the University of Applied Sciences of Italian Switzerland and lecturer of workshops on Sign Language at the Faculty of Education at the University of Milan Bicocca. It deals with education and schooling of children with Charge Syndrome since 2003, in contexts homecare, hospital and school.

Currently PhD student in Communication Sciences and training in the Department of Medicine University of Milan Bicocca: "San Gerardo" Hospital in Monza. (Multimedia Health Communication Laboratory: MHCL). Recently published with Pietro Celo three volumes for starting to read and writing of deaf children: "Guanti Rossi" Milan in 2014, the booklet: "Lo zoo Andrea" and the French version for deaf children in Burkina Faso "Gants Rouges".

For years, my work is structured around the children's needs with the Charge Syndrome, of their household especially of school education and the integration of the family and the child. Currently as a PhD Student: my research focuses on the investigation of mother-child with a rare syndrome, in particular with the Charge Syndrome, with a specific attention to narrative as a cure and overcome the grief.

Stefano Ardenghi

PhD Student, University of Milano-Bicocca Department of Human Science for Education "R. Massa", Piazza Ateneo Nuovo 1, Milano, Italy Stefano Ardenghi, Via G. L. Bernini, 7 - 20900 Monza (MB), Italy s.ardenghi@campus.unimib.it stefano.ardenghi@hotmail.it mobile: 0039.3405450929

Since January 2013, PhD in Science of Education and Communication - curriculum: "Wellness of the person, health and intercultural communication" at the University of Milan-Bicocca.

In 2010 he received with honors bachelor's degree in Psychological Sciences and Techniques (curriculum: Counselling and Mental Health) with a thesis on body image disturbances in people frequenting gyms (supervisor: Prof. Chiara Ripamonti). In July 2012 he received, with honors, master's degree in Clinical and Developmental Psychology and Neuropsychology with a thesis on the analysis of risk factors and symptoms in a sample of women with postpartum psychological distress (supervisors: Prof. Lucia Leonilde Carli and Prof. Maria Grazia Strepparava).

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Association between intellectual, cognitive, affective skills and Sign Language in a child with a rare disease: case study

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INTRODUCTION:

1. LIS as a competence that helps to develop socialization:

As known *Piaget* was the first one who correlates cognitive skills and linguistic skills. *Bates, Camaioni, Volterra* (1986) individuated four pre-requisites for the language appearance: 1) symbolic playing; 2) combinatorial playing; 3) imitation game; 4) the use of instruments. Language appears to be an instruments that evolves in a close relation with the psychic subject's organization and with the relational one. Researches shows the deaf subjects exposed to LIS till the first age, develop a linguistic competence equal to hearing language voice subjects.

2. Attachment's relation as a place of affective experimentation:

Deaf children and CHARGE syndrome children have the same necessities to consolidate the innate competence of communication and to achieve new ones that lead them to interact with the caregivers. Attachment's models are affective regulation's strategies where emotions have different function of assess:

1) the environmental surroundings; 2) the organism's state; 3) the availability of the caregivers.

The affective communication is the link to convey the first relational and attachments model between caregiver and son. The capability of recognizing own emotions, is influenced by the different type of emotive availability that the subject has experimented towards his own caregiver.

3. Attachments and disability:

A family after a disability's diagnosis cross 9 phases:

1) shock: it represents a phase of daze; 2) refusal: when the shock is too heavy, the refusal takes over; 3) rationalization as a refusal of the disability's form; 4) rage: it can be shown towards the son, doctors and partners; 5) guilty: there is a guilt trip for the belief of being responsible towards the son; 6) shame: caregiver can prove a sense of shame for the son; 7) fear: it can be the fear of not being able to succeed in checking the situation and the fear for the future; 8) anticipating pain: when the caregiver is prospected into the future; 9) acceptance: caregivers start to accept the situation.

After acceptance, the answer of adaptation comes in 3 phases: 1) the appearance of a stressing agent (for example, the diagnosis); 2) organism's alarm reaction; 3) mobilization to face up to the stressing agent through coping strategies.

METHODOLOGY:

The child is followed at home and at school from 2006 (him chronological age 3 years).

The methodology uses: 1.Three public relations released dall'AIAS of Membership (2006-2007-2009 years); 2. Three evaluations of Scala Griffith (2006-2007-2009 years); 3. Progressive Matrices Raven 's non verbal intelligence (2014); 4.Goodenoug test (2015); 5. Test for Understanding emotions TEC (2015); 6. Functional Diagnosis (2008-2009 and 2014-2015).

CONCLUSIONS:

The Sign Language(SL), even if not early, come only Communication System could having fostered and encouraged individual skills language, non verbal communication, meta-cognitive, emotional and intellectual. The results obtained encourage ADDITIONAL insights to evaluate the association between LS and individual skills cognitive - affective of deaf subjects with rare disease.

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Review of Two Research-Based Methods for Improving Sleep of Children with Developmental Disabilities

Benjamin Kennert and Timothy S. Hartshorne, Ph.D., Central Michigan University CHARGE Syndrome Research Lab

Presenter Information:

Ben Kennert is a third year doctoral student in the school psychology program at Central Michigan University and the most senior member of the CHARGE research lab currently, working under Dr. Tim Hartshorne. Ben first began researching CHARGE Syndrome three years ago when he met Dr. Hartshorne, who is a professor in my graduate school program. Ben became very interested in this research, and enjoyed thinking about and working through the many challenges that are related to CHARGE Syndrome. When Ben met his first group of people with CHARGE Syndrome at the conference in Scottsdale, Arizona, he immediately fell in love with the CHARGE community. He has never met another group with so many challenges, yet so much motivation and determination to "CHARGE On!" Since then he has met many children and families of the CHARGE community, and have learned a lot and had a lot of fun along the way. His interest is in the area of behavior, with his research focusing on behavioral difficulties that are important to children with CHARGE syndrome and their families. His current research focuses on sleep problems among children with CHARGE Syndrome. He has also been involved in research projects involving self-regulation and quality of life of individuals with CHARGE Syndrome.

Presentation Abstract:

Sleep problems are common among children with various developmental disabilities, including CHARGE Syndrome. However, very little research has been done on treatments for these sleep problems, especially for problems related to falling asleep and staying asleep, the most common sleep problems for children with CHARGE. This presentation will review two treatments that have been shown as useful among children with developmental disabilities, and their potential usefulness with CHARGE: Melatonin treatment and positive bedtime routines.

Review of Two Research-Based Methods for Improving Sleep of Children with **Developmental Disabilities BENJAMIN KENNERT AND TIMOTHY HARTSHORNE CENTRAL MICHIGAN UNIVERSITY**

How Sleep Works

- u Two phases: REM and NREM
- u 4 phases of NREM corresponding to deepness of sleep and changing brain waves
- u Controlled by circadian rhythms (i.e., our biological clock)
 - Kept in rhythm by the superchiasmatic nucleus within the hypothalamus, which is connected to the nerve fibers of our eyes
 - u Signaled by Melatonin, a hormone produced by the pineal gland when light enters our eyes
- u Sleep begins in stage 1 of NREM, and cycles through the sleep stages several times throughout the night
- u About 60 minutes after falling asleep we reach deep sleep stages
- u Brief periods of waking throughout the night

Overview of Sleep Difficulties

- Many different types of sleep difficulties: Initiation (falling asleep), maintenance (staying asleep), sleep-breathing, etc.
- Some evidence shows that sleep difficulties are more common among children with various developmental disabilities
- u Associated with several poor outcomes (Sung et al., 2008):
 - u Poorer quality of life
 - u Poorer caregiver mental health
 - u Poorer daily functioning
 - u Higher likelihood on lateness or absence from school
- Associated with behavioral problems as reported by caregivers (Hartshorne et al., 2008)
- u May reduce a child's motivation and concentration (Durand, 1998)

Sleep Difficulties and CHARGE Syndrome

u May be predisposed for sleep problems for several reasons:

- u Poor vision
- u Behavioral traits
- u Early experiences (e.g., hospital stays)
- u Craniofacial abnormalities
- u Self-regulation difficulties
- u Pain
- u Anxiety
- u 57.5% of children with CHARGE Syndrome presented with a clinically relevant sleep problem in one study (Hartshorne et al., 2008).
 - u Most common type were sleep initiation and maintenance problems
 - u Sleep problems linked to behavioral problems and poorer caregiver well-being
- Half of adolescents and adults with CHARGE continue to report sleep difficulties significant enough to have an impact on their lives (Blake et al., 2005)
- u Some focus on obstructive sleep apnea in the research, and medical treatment (e.g., Trider et al., 2012), but little research on the more "behavioral" sleep issues

Melatonin Treatment

- u Melatonin is a hormone naturally produced in the brain
- It is considered an "evidence-based" treatment for sleep difficulties, and shows promising evidence among children with developmental disabilities.
 - For example, in a study among 107 children with an Autism Spectrum disorder, following Melatonin treatment 25% of children no longer had sleep difficulties, 60% improved sleep but still had some difficulties, 14% had no change, and only 1% experienced worse sleep (Andersen et al., 2008)
- Because it signals the brain when to fall asleep, Melatonin may be more useful for initiating sleep, but may be less helpful with maintaining sleep throughout the night

Positive Bedtime Routines with Circadian Rhythm Management

- u Positive Bedtime Routines:
 - u 4-6 calm activities are consistently done prior to bedtime (e.g., take a bath or read a book)
 - u Usually sleep hygiene kept in mind, or "setting the conditions for sleep."
 - u For example, turning lights off, limiting noise, avoiding caffeine before bed, etc.
- u Circadian Rhythm Management
 - u Scheduled times for waking up and for eating throughout the day
 - u Intention is to reset the body's biological clock (circadian rhythms)
- u The combination of positive bedtime routines before bed, and regular waking and eating times during the day, may help regulate the sleep cycle. A variety of evidence exists to support the methods separately, and in combination.

Upcoming Study

- u Explore these potential interventions for sleep initiation and maintenance problems for children with CHARGE Syndrome.
- u Research Questions:
 - Can melatonin treatment be useful in reducing sleeping difficulties for children with CHARGE Syndrome related to initiation and maintenance of sleep?
 - Can a structured, positive bedtime routine with circadian rhythm management be useful in reducing sleeping difficulties for children with CHARGE Syndrome related to initiation and maintenance of sleep?
 - Will a combination of melatonin treatment and a positive bedtime routine, with circadian rhythm management, be more effective in reducing sleeping difficulties for children with CHARGE Syndrome related to initiation and maintenance of sleep than these treatments separately?
 - u What are the factors predicting successful outcomes for these treatments among children with CHARGE Syndrome?

Upcoming Study

- u Multiple baseline design with 2 groups
 - One group will move from baseline, to intervention A (melatonin), to combination of intervention A and B (melatonin with positive bedtime routine and circadian rhythm management)
 - U Other group will move from baseline, to intervention B (positive bedtime routine and circadian rhythm management), to combination of intervention A and B (melatonin with positive bedtime routine and circadian rhythm management)
- Participants first sent an initial survey to gather information about sleep problems and what is being done, or has been tried, in the past. This will be sent with a validated sleep screening scale in order to validate that a sleep problem is present.
- u 6-8 participants with sleep initiation and maintenance problems will be recruited, from the group of participants who returned the initial survey and rating scale.
- u Participants will complete 3 phases each: 1) baseline, 2) treatment, and 3) combined treatments.
- u Bedtime behaviors will be recorded by participants and parents/caregivers using a sleep diary
- u If you are interested in the study, please write your email address down at the conference!