Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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 (www.communicationmatrix.org) 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.

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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

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

Blake, K.D., MacCuspie, J., Hartshorne, T. S., Roy, M., Davenport, S. L. H., & Corsten, G. (2009). Postoperative airway events of individuals with CHARGE syndrome. *International Journal of Pediatric Otorhinolaryngology, 73,* 219-226.

Hartshorne, T. S. (2014, January). The needs of the person with CHARGE syndrome. *DbI Review, 52,* 48-51.

Hartshorne, T. S., & Cypher, A. D. (2004). Challenging behavior in CHARGE syndrome. *Mental Health Aspects of Developmental Disabilities, 7*(2), 41-52.

Hartshorne, T. S., Grialou, T. L., & Parker, K. R. (2005). Autistic-Like Behavior in CHARGE Syndrome. *American Journal of Medical Genetics, 133A,* 257-261.

Hartshorne, T. S., Hefner, M. A., & Davenport, S. L. H. (2005). Behavior in CHARGE Syndrome: Introduction to the series. *American Journal of Medical Genetics, 133A,* 228-231.

Hartshorne, T. S., Heussler, H. S., Dailor, A. N., Williams, G. L., Papadopoulos, D., and Brandt, K. K. (2009). Sleep Disturbances in CHARGE Syndrome: Types and Relationships with Behaviour and Caregiver Well-Being. *Developmental Medicine and Child Neurology, 51,* 143-150.

Hartshorne, T. S., & Hissong, K. N. (2014). CHARGE Syndrome: An Introduction for Speech-Language Pathologists. *SIG 16 Perspectives on School-Based Issues, 15*(2), 94-102. doi:10.1044/sbi15.2.94.

Hartshorne, T. S., Nicholas, J., Grialou, T. L., & Russ, J. M. (2007). Executive function in CHARGE syndrome. *Child Neuropsychology, 13,* 333-344.

Hartshorne, T. S., Stratton, K. K., & van Ravenswaaij, C. M. A. (2011). Prevalence of genetic testing in CHARGE syndrome. *Journal of Genetic Counseling, 20,* 49-57.

Kennert, B., Hartshorne, T. S., Wanka, A., Dix, H., & Nicholas, J. (2014, July). Self-regulation of cognition in CHARGE syndrome. *DbI Review, 53,* 14-18.

Kennert, B., Ramirdz, M., Hartshorne, T. S ,Deuce, G., & Nichoals, J. (2015, January). Self-regulation of meotion in CHARGE syndrome. *DbI Review, 54,* 26-30.

Ramirez, M. A., Hartshorne, T. S., & Nicholas, J. (2014, January). Self-regulation in individuals with CHARGE syndrome. *DbI Review, 52,* 43-47.

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

Thelin, J. W., Hartshorne, T. S., & Hartshorne, N. S. (2000). Audiologic and educational issues in CHARGE Syndrome. *Journal of Educational Audiology, 7,* 34-41.

Wachtel, L. E., Hartshorne, T. S., & Dailor, A. N. (2007). Psychiatric diagnoses and psychotropic medications in CHARGE syndrome: A pediatric survey. *Journal of Developmental and Physical Disabilities, 19,* 471-483.

Williams, G. L., & Hartshorne, T. S. (2005). Understanding balance problems in children with CHARGE syndrome. *Deaf-Blind Perspectives, 12* (2), 5-7.

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Mouth over-stuffing during meals in individuals with CHARGE syndrome

Alexandra Hudson, MD Candidate 2018, Department of Medicine, Dalhousie University, Halifax NS
Dr. Kim Blake, Department of Pediatrics, Dalhousie University, Halifax NS
Meghan Macdonald, MD Candidate 2017, Faculty of Medicine, Dalhousie University

Presenter Information:

Alexandra is a medical student at Dalhousie University in her first year of study. Prior to attending medical school, she completed my Honors Bachelor of Science at the University of Toronto in human biology, physiology, and psychology. She is currently studying CHARGE syndrome and its associated eating issues, under the supervision of Dr. Kim Blake at the IWK Hospital in Halifax, Nova Scotia.

Presentation Abstract:

Individuals with CHARGE syndrome commonly experience issues with eating[12]. Many individuals experience issues with over-stuffing their mouth or pocketing food in their cheeks, which may have consequences such as aspiration[3]. These issues may be associated with structural anomalies, motor impairment, and/or oral sensory impairment. This qualitative study interviewed caregivers to help better describe the mouth over-stuffing phenomenon in individuals with CHARGE syndrome, and assessed its resulting impact on caregivers themselves.

Mouth Over-Stuffing and Pocketing of Food in Cheeks in Children with CHARGE Syndrome

Description of Study

- There is minimal information concerning mouth over-stuffing or pocketing of food in cheeks during eating in children/adults with CHARGE syndrome and the resulting feeding and swallowing issues
- This study would like to describe this phenomenon by interviewing parents of children or adults with CHARGE syndrome who have experienced these eating behaviors

Eligibility

- Parents of children or adults with a confirmed diagnosis of CHARGE syndrome who over-stuff their mouth and/or pocket food in their cheeks during meals
- No age limit on CHARGE participants

Contact

- Alexandra Hudson, Student Researcher, at dalmed.charge@gmail.com or (902) 877-8727

Poster Presentation Highlights

Preliminary Results

- Interviewed 17 parents (to date) of children and adults aged 2-32 years
- Parent’s have highlighted issues from these feeding behaviors
  - Choking
  - Previous eating therapy has not addressed these issues
  - Requires careful supervision when eating
  - Increased time to finish eating

Things to Avoid

- Foods to avoid – e.g. Bread, hard vegetables, dry food
- Behaviors to avoid – e.g. Don’t put too much food on their plate at one time

Parent’s Tips & Tricks

- Reminders to chew and swallow
- Use water or a liquid chaser when eating
- Have both puree and solid food options at a meal
- Use smaller spoons
- Use an iPad or a TV show to distract while eating
- Cut food into really tiny pieces

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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

- Two phases: REM and NREM
- 4 phases of NREM corresponding to deepness of sleep and changing brain waves
- 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
  - Signaled by Melatonin, a hormone produced by the pineal gland when light enters our eyes
- Sleep begins in stage 1 of NREM, and cycles through the sleep stages several times throughout the night
- About 60 minutes after falling asleep we reach deep sleep stages
- 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
- Associated with several poor outcomes (Sung et al., 2008):
  - Poorer quality of life
  - Poorer caregiver mental health
  - Poorer daily functioning
  - Higher likelihood on lateness or absence from school
- Associated with behavioral problems as reported by caregivers (Hartshorne et al., 2008)
- May reduce a child’s motivation and concentration (Durand, 1998)

Sleep Difficulties and CHARGE Syndrome

- May be predisposed for sleep problems for several reasons:
  - Poor vision
  - Behavioral traits
  - Early experiences (e.g., hospital stays)
  - Craniofacial abnormalities
  - Self-regulation difficulties
  - Pain
  - Anxiety
- 57.5% of children with CHARGE Syndrome presented with a clinically relevant sleep problem in one study (Hartshorne et al., 2008).
  - Most common type were sleep initiation and maintenance problems
  - 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)
- 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

- 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

- Positive Bedtime Routines:
  - 4-6 calm activities are consistently done prior to bedtime (e.g., take a bath or read a book)
  - Usually sleep hygiene kept in mind, or "setting the conditions for sleep."
    - For example, turning lights off, limiting noise, avoiding caffeine before bed, etc.
- Circadian Rhythm Management
  - Scheduled times for waking up and for eating throughout the day
  - Intention is to reset the body's biological clock (circadian rhythms)
- 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

- Explore these potential interventions for sleep initiation and maintenance problems for children with CHARGE Syndrome.
- 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?
  - What are the factors predicting successful outcomes for these treatments among children with CHARGE Syndrome?

Upcoming Study

- 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)
  - 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.
- 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.
- Participants will complete 3 phases each: 1) baseline, 2) treatment, and 3) combined treatments.
- Bedtime behaviors will be recorded by participants and parents/caregivers using a sleep diary
- If you are interested in the study, please write your email address down at the conference!

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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).

References

Barnett, L.A. (1984). Research note: Young children's resolution of distress through play. *Journal of Child Psychology and Psychiatry, 25,* 477-483.

Brown, P.M, Rickards, F.W., & Bortoli, A. (2001). Structures underpinning pretend play and word production in young hearing children and children with hearing loss. *Journal of Deaf Studies and Deaf Education, 6,* 15-31.

Celeste, M. (2006). Play behaviors and social interactions of a child who is blind: In theory and practice. *Journal of Visual Impairment & Blindness, 100,* 75-90.

Ferguson, R. & Buultjens, M. (1995). The play behaviour of young blind children and its relationship to developmental stages. *British Journal of Visual Impairment, 13,* 100-107.

Fisher, E.P. (1992). The impact of play on development: A meta-analysis. *Play & Culture, 5,* 159-181.

Kennedy-Behr, A., Rodger, S., & Mickan, S. (2013). A comparison of the play skills of preschool children with and without developmental coordination disorder. *Occupational Therapy Journal of Research: Occupation, Participation, and Health, 33,* 198-208.

Leach, D., Pratt, C., & Roberts, C. (1991). Classroom and playground interaction of students with and without disabilities. *Exceptional Children, 57,* 212-221.

Morelock, M.J., Brown, P.M., & Morrissey, A. (2003). Pretend play and maternal scaffolding: Comparisons of toddlers with advanced development, typical development, and hearing impairment. *Roeper Review, 26,* 41-51.

Skellenger, A., Rosenblum, L., & Jager, B. (1997). Behaviors of preschoolers with visual impairments in indoor play settings. *Journal of Visual Impairment & Blindness, 91,* 519-530.

Spencer, P.E. & Meadow-Orlans, K.P. (1996). Play, language, and maternal responsiveness: A longitudinal study of deaf and hearing infants. *Child Development, 67,* 3176-3191.

Troster, H.H, & Brambring, M.M. (1994). The play behavior and play materials of blind and sighted infants and preschoolers. *Journal of Visual Impairment & Blindness, 88,* 421-443.

Wing, L., Gould, J., Yeates, S.R., & Brierley, L.M. (1977). Symbolic play in severely mentally retarded and in autistic children. *Journal of Child Psychology and Psychiatry, 18,* 167-178.

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2:15 - 4:30pm
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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.

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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.

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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).

In the academic year 2012/2013, he won the competition for admission to the PhD degree in Science Education and Communication (XXVIII cycle) at the Department of Human Sciences for Education "Riccardo Massa" of the University of Milano Bicocca.

From January 2013, he has studied Procedures and criteria for selection of candidates for the degree programs of the School of Medicine, main cognitive and non-cognitive predictors of success in Degree of Medicine and the change in the representation of the doctor-patient relationship in the students of the School of Medicine

Currently student of Cognitive-Behavioural Therapy at the "Centro di Terapia Cognitiva" in Como.

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

Nicole Vian°, Stefano Ardenghi•

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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.

BIBLIOGRAPHY:

I., Fabio, R. A., Marchetti, A. (2008), *Understanding emotions and mental states from faces and pictures in Rett syndrome*, In: M. Balconi (Ed.), Emotional face comprehension. Neuropsychological perspectives (pp. 205-232),Issekutz KA, Graham JM Jr, Prasad C, Smith IM, Blake KD. 2005. An epidemiological analysis of CHARGE syndrome: Preliminary results from a Canadian study. K.D. Blake, I.M. Russell-Eggitt, D.W. Morgan, J.M. Ratcliffe, R.K. Wyse, Who's in CHARGE? Multidisciplinary management of patients with CHARGE association, Arch. Dis. Child. 65 (1990) 217—223. Lacombe D (1994): Facial palsy and cranial abnormalities in CHARGE association. Am J Med Genet 49:351–352; L.E. Vissers, C.M. van Ravenswaaij, R. Admiraal, J.A. Hurst, B.B. de Vries, I.M. Janssen, et al. Mutations in a new member of the chromodomain gene family cause CHARGE syndrome, Nat. Genet. 36 (2004) 955—957; Lewis C, Lowther J. 2001. CHARGE Association: Symptoms, behaviour and intervention. Educ Psychol Pract 17:69–77. Lieberman, A.F., Pawl, J.H. (1990). Pennebaker, J. W., Seagal, J. D. (1999). Forming a story: The health benefits of narrative. Journal of clinical Psychology, 55(10), 1243-1254. Stanton, A. L., Danoff, B. S. (2002). Emotional expression, expressive writing and cancer. In S. J. Lepore & J. M. Smyth (Eds.), The writing cure: How expressive writing promotes health and emotionl well-being (pp. 31-51). Stern, D.N. (1995).

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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: Inclusive Recreation

Gretchen Imel and Tim Hartshorne, PhD • Department of Psychology • Central Michigan University

CHARGE Syndrome

CHARGE Syndrome is a recognizable (genetic) pattern of birth defects which occurs in about one in every 15,000 births. It is an extremely complex syndrome, involving extensive medical and physical difficulties that differ from child to child. Most have hearing loss, vision loss, and balance problems which delay their development and communication along with swallowing and breathing problems. Individuals diagnosed with this syndrome may spend many months in the hospital and undergo many surgeries and other treatments. All are likely to require medical and educational interventions for many years (International CHARGE Syndrome Foundation).

Importance of Recreation

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. This is true for individuals diagnosed with CHARGE Syndrome. Nonetheless, almost any sport can be modified using technology or adaptive equipment to accommodate participation at any ability level (www.atra-online.com).

CMU
CENTRAL MICHIGAN UNIVERSITY

Challenge Mountain

Challenge Mountain is a non-profit organization dedicated to enriching and improving lives for the mentally and physically challenged through outdoor recreation, including adaptive downhill skiing and snowboarding during the winter months (www.cmski.org).

Adaptations for Skiing:
-Guides for blind skiers
-Bi-skis (sit down ski)
-Two-track skis
-Ski bike
Benefits for CHARGE Syndrome:
-Increase proprioception awareness (one's ability to feel the position of different body parts and the effort that goes into moving them)
-Improve coordination
-Improve balance; increase core muscle tone
-Improvement in cardiovascular system

Camp Abilities – Brockport, NY

Camp Abilities Brockport is a one-week developmental sports camp for children and teens who are blind, visually impaired, and deafblind. The mission of Camp Abilities Brockport is to empower children and teens with visual impairments to be physically active and productive member of their communities, as well as to improve the health and well-being of people with sensory impairments. Activities include canoeing/kayaking, gymnastics, one-touch self-defense program, rollerblading, tandem biking, beep baseball, goalball, horseback riding, rock climbing, swimming, track & field, and showdown.

After their summer experience at camp, the children are more able, independent, and confident in their abilities. Camp Abilities Brockport serves as a vehicle to assist children with disabilities to go out and become involved in their communities. Once this occurs, the individuals in the community gain respect for individuals with visual impairments and increase the understanding and respect about individuals with different abilities (www.campabilities.org).

Equestrian Therapy

Equestrian therapy is the use of horses and equine-assisted activities in order to achieve goals that enhance physical, emotions, social, cognitive, behavioral, and educational skills for people who have disabilities (www.equestriantherapy.com).
Benefits for CHARGE Syndrome:
-Experience the independent movement of the pelvis and shoulders, which are essential for enabling healthy walking gait
-Improved balance, coordination and muscle strength/control
-Improved postural control
-Stimulates sensory integration
-Improved gross and fine motor skills
-Improved self-esteem and self-confidence
-Socialization and improved interpersonal skills

Ethan Wolfe Recreational Assistance Program

Provides individuals with CHARGE Syndrome the opportunity to participate in recreational activities. This program is need-based and funded through a generous gift from The Wolfe family and friends. During Ethan Wolfe's 12 years of life, he rode an adaptive bike, attended special recreational camps, participated in swimming lessons, and music class. David & Jody Wolfe, together with the CHARGE Syndrome Foundation, wish to honor Ethan's memory by providing other individuals with CHARGE with similar, fun activities and equipment that enriched his life (http://www.chargesyndrome.org/documents/EthanWolfeRecreationalAssistanceProgram.pdf).

The Sky's the Limit!
12th International
CHARGE Syndrome
Conference
Chicago 2015

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

“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 deaf-blind 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 deaf-blindness learn different types of communication and language techniques including tactile and co-active signing, sensory processing interventions, methods for maximizing visual and auditory abilities, hand-under-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.

FREE!

Deaf-Blind Parent/Child Playgroup

Every Other Thursday, 3:00-5:00pm

CEID Campus - 1035 Grayson Street, Berkeley, CA 94710

FIRST 5 ALAMEDA COUNTY

CEID

Who qualifies?

What will we do?

- Learn about communication techniques specific to deafblindness, including tactile signing and object communication.
- Acquire techniques to support your child's cognitive, language, social and emotional development.
- Practice sensory processing activities.

For information or to refer/register, contact:
Kimberly Tarasenko, Teacher of the Deaf

Typical Playgroup Session

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 daily home routines)

- 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

○ Greetings and name signs
○ Establishing and following routines
○ Follow the Child
○ Concept Development
○ Encouraging use of functional vision
○ The Other Senses
○ Literacy and experience books
○ IFSP and IEP process and advocacy
○ Using play to promote interactions
○ Interveners
○ Planning family outings
○ Self-regulation
○ Including your child in holiday traditions and family gatherings
○ Self-determination
○ Family resiliency and leadership

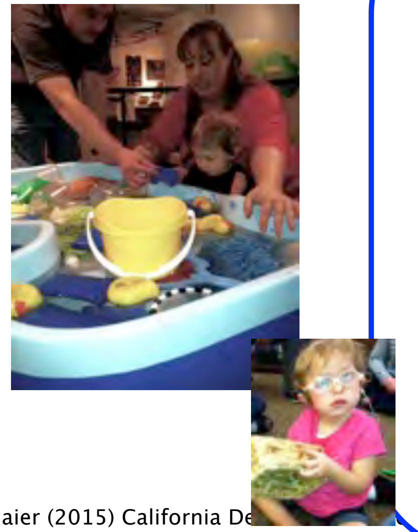
Outcomes We Had Hoped For

- 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 aquatic 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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Communication Matrix-Growing a Virtual Community of Practice to Support Students with Complex Communication Needs

Amy T. Parker, Ed.D. & COMS,
National Center on Deaf-Blindness
The Teaching Research Institute
Western Oregon University
Charity Rowland, Ph.D., & Alexandria Cook,
Oregon Health Sciences University

Presenter Information:

Dr. Amy Parker has nearly 20 years' experience in working with people who are deaf-blind as an employment specialist, independent living teacher, in-home parent trainer and advocate. She received her doctorate in special education with an emphasis in deaf-blindness and a certification in orientation and mobility in 2009 through an OSEP funded leadership and enrichment fellowship. She also happens to be the sister of an adult who happens to be deafblind and has multiple disabilities who reminds her about what's important in life every day. She lives with her husband and two children in Monmouth, Oregon and loves to travel.

Presentation Abstract:

The Communication Matrix (CM) has been used to support individuals with complex communication needs and teams for many years. Until recently, there has not been a way for individuals who are using the CM to share what they are learning with other teachers, parents and practitioners. To address this problem, our project has developed a virtual community of practice to facilitate use of the CM and to grow knowledge through dialogue.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Modeling CHARGE syndrome in zebrafish: A look at the innervation and function of the gastrointestinal system

Kellie Cloney, Department of Medicine, Shelby L. Steele, Department of Pediatrics, Matthew Stoyek, Department of Physiology and Biophysics, Roger P. Croll, Department of Physiology and Biophysics, Frank M. Smith, Department of Medical Neuroscience, Kim Blake, Department of Pediatrics, Jason N. Berman, Departments of Pediatrics, Microbiology and Immunology, and Pathology, Dalhousie University, Halifax NS, Canada

Presenter Information:

Kellie is a medical student at Dalhousie University in my second year of study. Prior to attending medical school, she completed a Master's degree in developmental biology. She studied the molecular pathways involved in craniofacial development, in chicken embryos. After she completed my Master's degree, Kellie continued to research the molecular mechanisms of skull development in zebrafish. Once in medical school, she sought research in a similar field to my previous work but with more of a clinical application. This lead me to connect with Dr. Blake and Dr. Berman who were about to begin collaborating on a project using zebrafish to model CHARGE syndrome. She has been performing her research in collaboration with Dr. Shelby Steele, a postdoctoral fellow in the Berman lab.

Presentation Abstract:

This research focuses on modeling aspects of CHARGE syndrome in zebrafish, by knocking down the expression of *chd7*. We labeled the nerves of the zebrafish and imaged the gastrointestinal tract. Using this method, we investigated the morphology of the gastrointestinal (GI) system as well as the cranial and enteric nerves that innervate the GI tract. The results of this study have potential to impact the management of patients with CHARGE syndrome.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

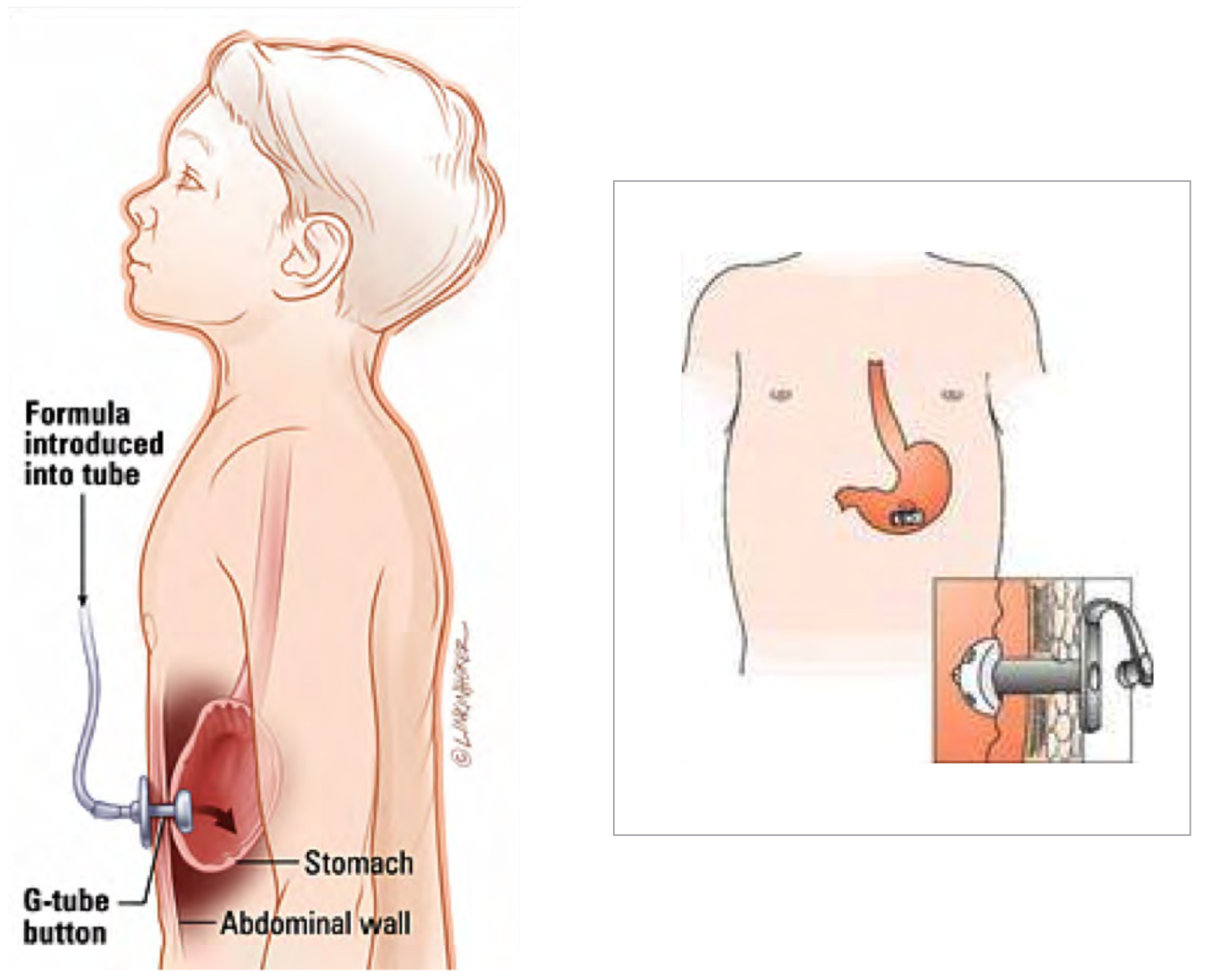
Rachel Malta & Tim Hartshorne, Ph.D. • Central Michigan University

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 (Åvitsland 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

- 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

Why is this topic important?

- 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 others around them.
- 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.

Research Questions

- 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 the G-tube?
- Does social media play a large role in the decision? What percentage of parents used social media to gather advice/opinions?
- 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 successful attempt?
- Qualitatively, were there any common events that prompted the decision to remove?

References

Arvedson, J. C. (2008). Assessment of pediatric dysphagia and feeding disorders: clinical and instrumental approaches. *Developmental Disabilities Research Reviews, 14*(2), 118–27. doi:10.1002/ddrr.17

Åvitsland, T. L., Birketvedt, K., Bjørnland, K., & Emblem, R. (2013). Parent-reported effects of gastrostomy tube placement. *Nutrition in clinical practice: official publication of the American Society for Parenteral and Enteral Nutrition, 28*(4), 493–8. doi:10.1177/0884533613486484

Blake, K.D., Hartshorne, T. S., Lawand, C., Dailor, A. N., & Thelin, J. W. (2008). Cranial nerve manifestations in CHARGE syndrome. *American Journal of Medical Genetics,* 146A, 585-592. doi: 10.1002/ajmg.a.3217

Dobbelsteyn, C., Peacocke, S.D., Blake, K., Crist, W., & Rashid, M. (2008). Feeding difficulties in children with CHARGE syndrome: prevalence, risk factors, and prognosis. *Dysphagia, 23*(2), 127–35. doi:10.1007/s00455-007-9111-6

Goldberg, E., Barton, S., Xanthopoulos, M. S., Stettler, N., & Liacouras, C. A. (2010). A descriptive study of complications of gastrostomy tubes in children. *Journal of Pediatric Nursing.* doi:10.1016/j.pedn.2008.07.008

Guerriere, D. N., McKeever, P., Llewellyn-Thomas, H., & Berall, G. (2003). Mothers' decisions about gastrostomy tube insertion in children: factors contributing to uncertainty. *Developmental Medicine and Child Neurology, 45*(7), 470–6. Retrieved from http://www.ncbi.nlm.nih.gov/pubmed/12828401

Jackson, C., Cheater, F.M., & Reid, I. (2008). A systematic review of decision support needs of parents making child health decisions. *Health expectations: an international journal of public participation in health care and health policy, 11*(3), 232–51. doi:10.1111/j.1369-7625.2008.00496.x

Lipstein, E.A, Brinkman, W.B., & Britto, M.T. (2012). What is known about parents' treatment decisions? a narrative review of pediatric decision making. *Medical decision making: an international journal of the Society for Medical Decision Making, 32*(2), 246–58. doi:10.1177/0272989X11421528

National Institutes of Health (2012). *Feeding tube insertion - gastrostomy.* Retrieved from http://www.nlm.nih.gov/medlineplus/ency/article/002937.htm

Nelson, P. A., Caress, A.L., Glenny, A.M., & Kirk, S.A. (2012). "Doing the 'right' thing": how parents experience and manage decision-making for children's "normalising" surgeries. *Social Science & Medicine (1982), 74*(5), 796–804. doi:10.1016/j.socscimed.2011.11.024

Nemours. (2013). *Surgeries and procedures: gastrostomy tube.* Retrieved from http://kidshealth.org/parent/system/med_procedures/g_tube.html

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UNIVERSITY

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

CMU
CENTRAL MICHIGAN UNIVERSITY
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
  - Lissencephaly
  - 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
  - Agenesis of the septum pellucidum
- Posterior fossa
  - Cerebellar (vermis) hypoplasia
  - Dandy-Walker malformation
  - Cerebellar heterotopias
- Brainstem
  - Agenesis of the 7th nucleus
  - Brainstem hypotrophy

Problems with the Literature

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

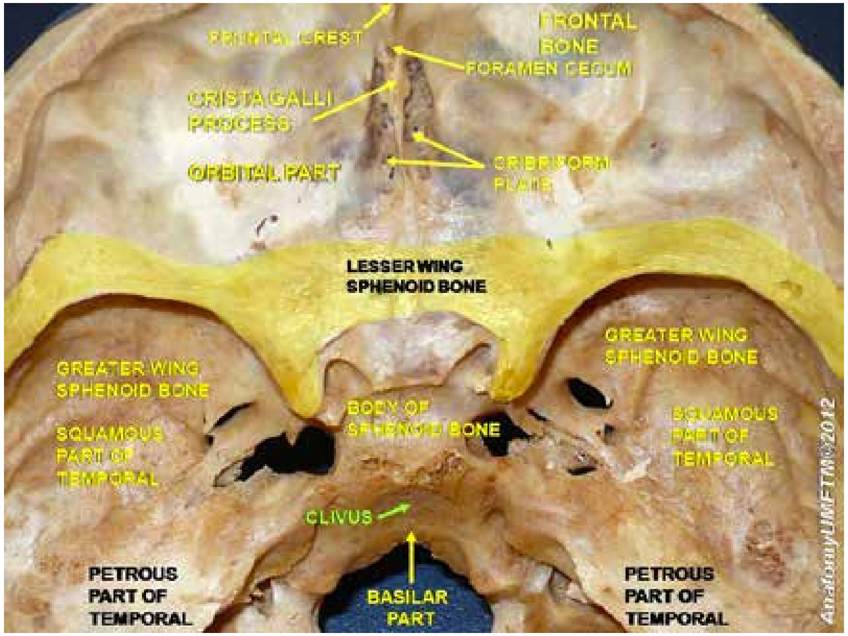
From the Netherlands

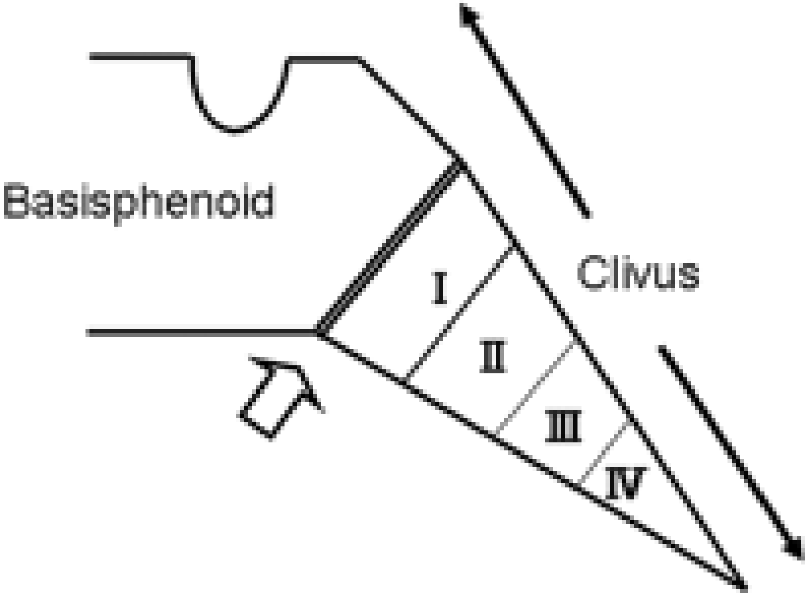
- Cortex
  - Cerebral/cortical atrophy
  - Dysgenesis of the frontal lobes
  - Gyral abnormalities
  - ~~Lissencephaly~~
  - ~~Nodular heterotopias~~
- Ventricles
  - Ventriculomegaly
  - Hydrocephalus
  - Ventricular dilation
- Anterior pituitary hypoplasia
- Hippocampal hypoplasia
- Olfactory system
  - Arhinencephaly
  - Hypoplasia of olfactory bulbs
- ~~Craniosynostosis~~
- Hemorrhagic/ischemic lesions
- Basioccipital hypoplasia and basilar invagination
- Midline defects
  - ~~Holoprosencephaly~~
  - **Partial** corpus callosum agenesis
  - ~~Meningoencephalocele~~
  - ~~Agenesis of the septum pellucidum~~
- Posterior fossa
  - Cerebellar (vermis) hypoplasia
  - Dandy-Walker malformation
  - **Foliation defects**
  - ~~Cerebellar heterotopias~~
- Brainstem
  - ~~Agenesis of the 7th nucleus~~
  - Brainstem hypotrophy

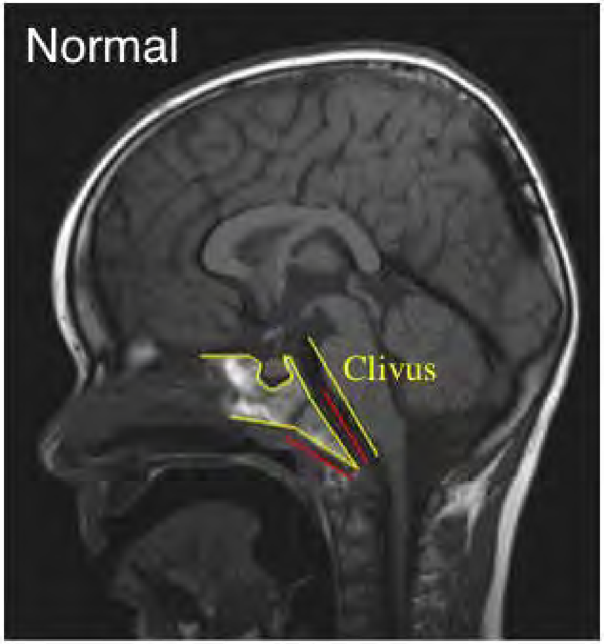
De Geus, C. Neurology in CHARGE syndrome. 2015 [powerpoint slides].

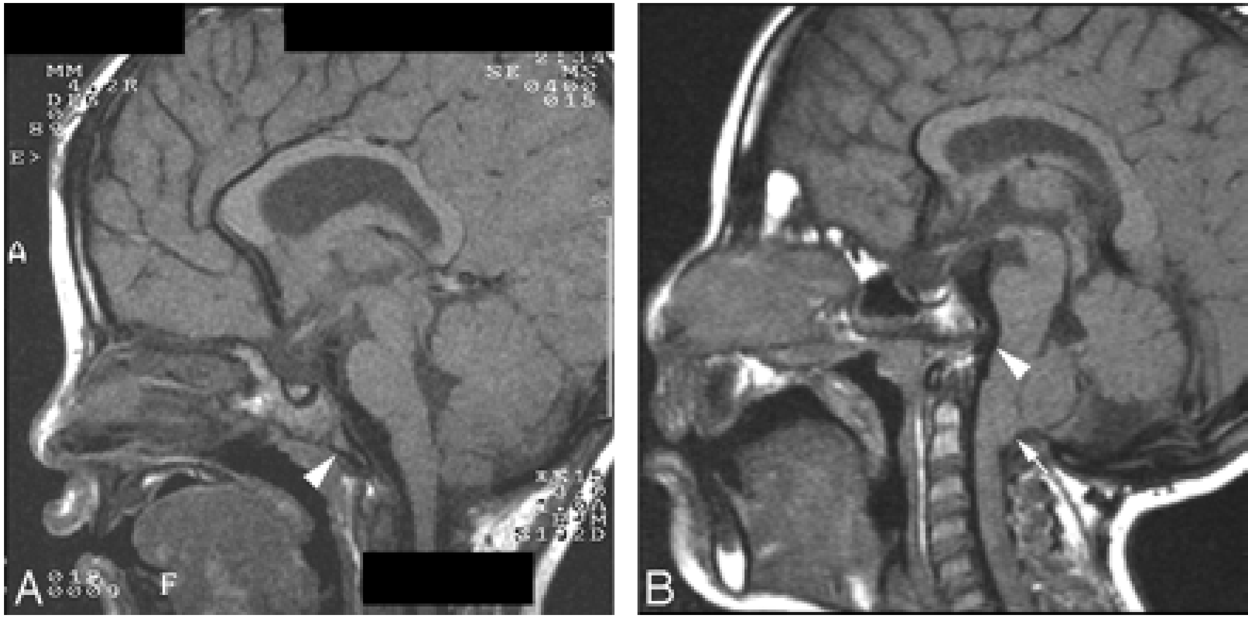
The Clivus in CHARGE

- 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
    - Basilar invagination
    - 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
    - “Sloping shoulders”
    - Goal
    - Challenges
    - 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
- De Geus, C. Neurology in CHARGE syndrome. 2015 [powerpoint slides].
- Fujita et al., Abnormal basiocciput development in CHARGE syndrome. AJNR Am J Neurorad 2009;30(3):629-34
- Yu et al., Deregulated FGF and homeotic gene expression underlies cerebellar vermis hypoplasia in CHARGE syndrome. Elife, Vol. 2, e01305, 24.12.2013.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

CHARGE Families in the Know Read and Listen to …

Lori Swanson, Sheri Stanger, and Megan Cote

Presenter Information:
Lori is an Associate Professor, Communication Sciences and Disorders at the University of Wisconsin-River Falls.
Sheri is the Director of Outreach with the CHARGE Syndrome Foundation.
Megan is Project Specialist for EI/R and Family Engagement at the National Center on Deaf-Blindness

Presentation Abstract:

This poster session will strive to showcase a range of current, pertinent written materials that families will want to know about and be able to access. Some of these resources will be CHARGE specific while others will come from the latest research and evidence-based practices on deaf-blindness but they will also be applicable to the CHARGE population. The session will offer a listing and sample of some of the “must reads” and “must listen to presentations” for families. NCDB/DB-LINK will be asked to assist in carefully selecting these “education” related resources. It is anticipated that the selection of materials on display will cover a wide range of areas such as early intervention, transition, recreation or socialization.

Family Leadership Project

The purpose of FamiliesLead.org is to bring a network of people and organizations together to share ideas, resources and support for leadership training for parents and other family members of individuals with disabilities.

Parent involvement in a child's education is key to success. When parents actively engage in their child's education at home, their children do better in school. Parents who become advocates and decision-makers at schools and in their community have the power to influence educational policies and practices that shape the future of their child's education.

The website features the following:

1) What is a Family Leader?

Learn more about why family leaders are important and how you can take action to shape the services your child receives.

2) Meet Family Leaders

Meet family leaders from across the country and learn their stories about how they chose to make a difference.

3) Power of Families

Learn more about how to get started making a difference in the life of your child and community.

4) Learning Materials

Find learning materials that will help you develop the fundamental knowledge and skills needed to create change.

5) Planning Tools

Discover tools to assist trainers and organizations to plan and deliver family leadership training.

www.familieslead.org

Literacy for Children with Combined Vision and Hearing Loss

ALL children CAN read…let us show you how!

This site is for individuals interested in beginning or enhancing literacy instruction for children with combined vision and hearing loss. Its content is also designed to improve literacy instruction for children with multiple disabilities and other complex learning challenges. Our contributors include State Deaf Blind Project staff as well as teachers who want to give back to the field and help more families play a role in educating children with complex learning challenges. The instructional techniques and tips provided on this site are evidence based practices for increasing literacy skills.




http://literacy.nationaldb.org

The Open Hands Open Access (OHOA) Intervener Learning Modules are a national resource designed to increase awareness, knowledge, and skills related to intervention for students who are deaf-blind and are being served in educational settings (ages 3 through 21). The development of the modules is in response to Recommendation 3 of the Recommendations to Improve Intervener Services (NCDB, 2012). Recommendation 3 is one of a set of recommendations intended to establish a strong national foundation for intervener training and workplace supports.

The module content was created by a diverse group of experts in the field of deaf-blindness including state and national deaf-blind project staff, parents of children who are deaf-blind, higher education faculty, teachers, educational interpreters, and interveners. Each includes a variety of accessible videos, photographs, slide presentations, and learning activities. The modules have been guided by an advisory committee, and reviewed by a variety of experts in deaf-blindness and the process of intervention, experts in module design, and field-test participants.

Register To Review OHOA for Free:
https://nationaldb.org/ohoaregister

http://moodle.nationaldb.org/

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

Michael Braga BS, M.Ed., Ruth Braga MSN, RN and Mindy Braga

Presenter Information:
Ruth and Michael are parents to Brandon (CHARGE)-17, Mindy-14, and Nicholas-7. Brandon has led them through multiple adventures, but Mindy inspired them to create a sibling support group for kids in their area who have a sibling with disabilities.

Presentation Abstract:
A survey was conducted about children with CHARGE and their siblings. The results will be shared. We are from many places and backgrounds, but we are all here because of an individual with CHARGE! We look forward to learning from everyone!

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Roles for Chd7 in inner ear development and function

Ethan D. Sperry, B.S., B.A., Diana F. Syam, M.Sc., and Donna M. Martin, M.D., 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).

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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 multi-disciplinary 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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

Hayley Hoesch & Timothy Hartshorne
Central Michigan University

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 because of who they 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.

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! :)**

Contact the Authors

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tim.hartshorne@cmich.edu

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Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

CHD7 controls cerebellar growth via Reelin

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

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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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Keeping it Simple: Strategies for Managing Behavior

Kasee Stratton, Ph.D., NCSP & Dan Gadke, Ph.D., NCSP, BCBA, Mississippi State University

Presenter Information:
Dr. Kasee Stratton and Dr. Daniel Gadke are assistant professors of school psychology at Mississippi State University and licensed psychologists. Dr. Stratton currently runs the Bulldog CHARGE Syndrome Research Lab at MSU and Dr. Gadke is the director of the Autism and Developmental Disabilities Clinic at MSU. Both specialize in challenging behaviors among individuals with developmental delays and genetic conditions. 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.

Presentation Abstract:
Our goal is to provide some clarity of what to do when your child engages in challenging behavior. We will teach strategies to understand the basics of behavior in CHARGE and to also better understand behavior of all children (that's right…siblings, too!). We will cover why challenging behavior occurs, how to identify the function of the behavior, and what to do next to intervene.

The Sky's the Limit!

12th International CHARGE Syndrome Conference Chicago 2015

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

An educational model to reduce passivity and increase self-determination 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 self-determination 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 School[1]. 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.

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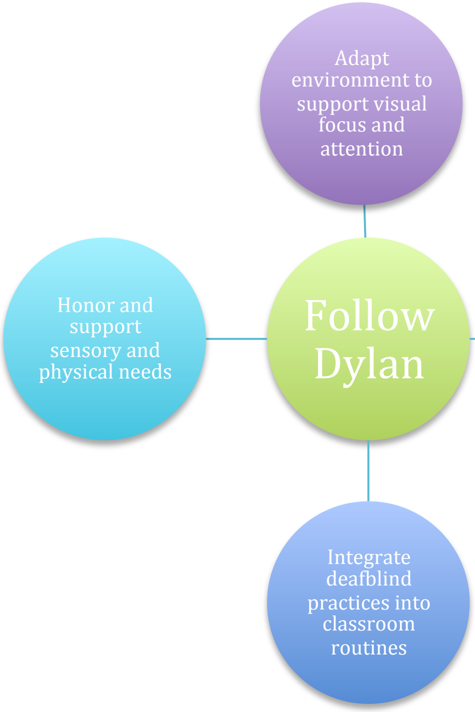
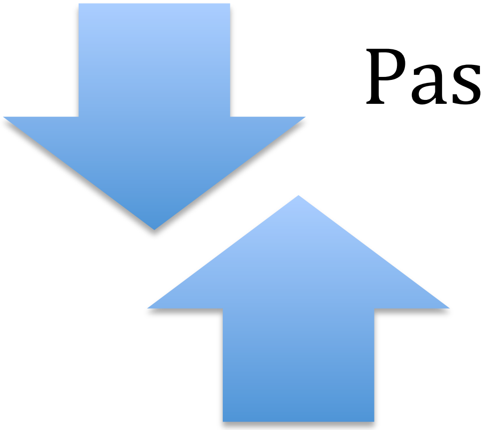
[1] Dylan's program is paid for by the State of Arizona through an Empowerment Scholarship Account.

Figure 1 Dylan's School-Home Educational Model



Create a Reactive and Responsive Environment for



Passivity

Self-Determination

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 Attention[2]

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.

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[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 Principles[4]

- **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.

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[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.

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[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.
Reactive or responsive environment
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.

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[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.

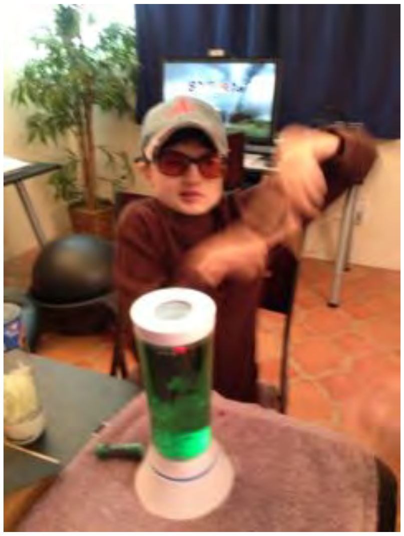
Bringing over the new science project—a tornado in a bottle—Dylan and Deb co-drew what it was and how to do it. Pouring water from a pre-filled pitcher into the bottle, the water spun in tornado fashion, but with clear water in a clear container could Dylan see it? Offering Dylan a box of food coloring, 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.



References

Alsop, L. e. (2012). *A Family's Guide to Interveners for Children with Combined Vision and Hearing Loss.* Logan, UT, USA: SKI-HI Intstitute.
Bluestone, J. (2004). *The Fabric of Autism: Weaving the Threads into a Cogent Theory.* Seattle, WA, USA: The HANDLE Institute.
Brown, D. (2014, Spring). What Does "Follow the Child" Mean? *ReSources , 19* (I).
Hartshorne, T. H. (2010). *CHARGE Syndrome.* (M. A. Hefner, Ed.) San Diego, CA, USA: Plural Publishing, Inc.
Lauger, K. (2013). The Box of Deafblindness. *DVI Quarterly , 58* (4), pp. 37-46.
Leotti, L. A. (2010). Born to Choose: The Origins and Value of the Need for Control. (R. Schwarzlose, Ed.) *Trends in Cognitive Science , 14* (10), 457-463.
McGinnis, J. M. (1999). *A Guide to Planning and Support for Individual who are Deafblind.* Toronto, Canada: University of Toronto Press.
Roman-Lantzy, C. (2007). *Cortical Visual Impairment: An approach to assessment and intervention.* New York, NY, USA: AFB Press.

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Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

Nancy Steele, 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

Consultation with DiCapta

Consultation with Described Captioned Media Program

Consultation with JKP Interpreting

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

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

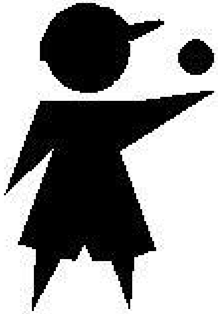
Self-Assessments- could serve as a rubric for grading

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.

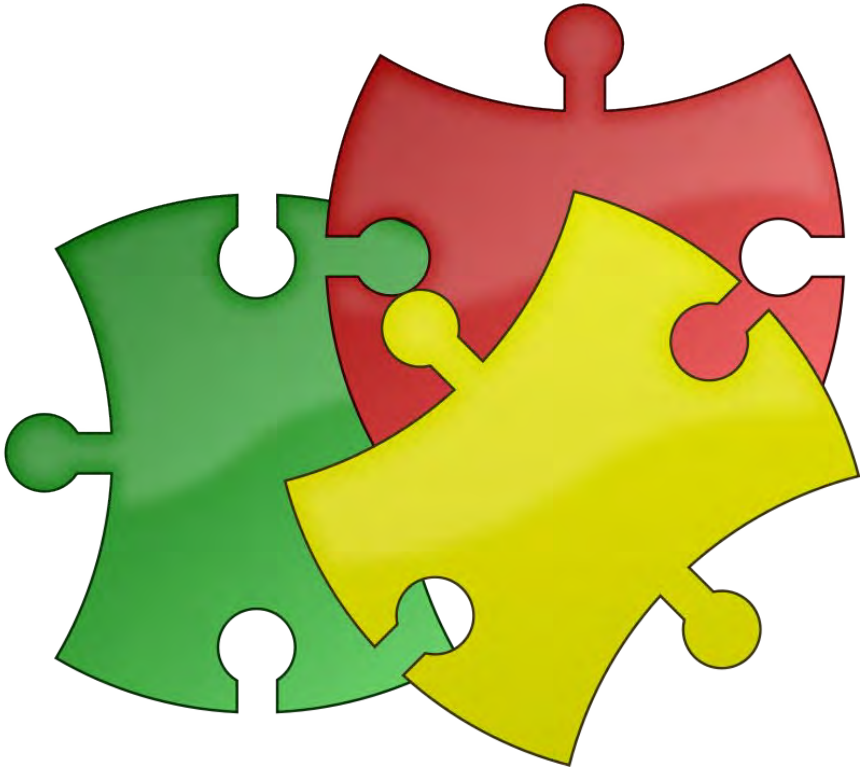


OHOA Online Modules
in Illinois
2013 - Spring/Summer 2015

PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES

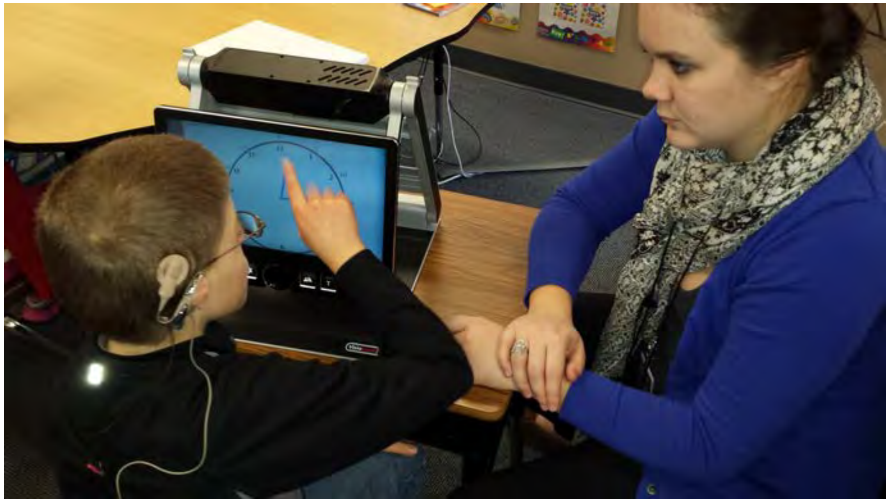
Prior to 2013

- 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 deaf-blind project



PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES

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

PROJECT REACH:
ILLINOIS DEAF-BLIND SERVICES

Who registered?

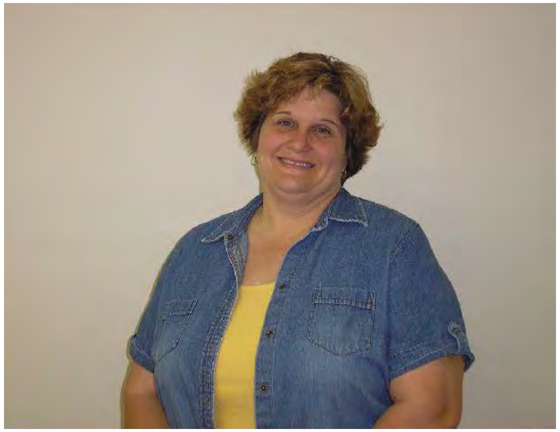
- Special Ed Teachers
- Parents
- Paraeducators
- Related Service Providers: O&M, SLP
- Administrators
- Teachers of the Deaf/Hard-of-Hearing (TDHH)
- Teachers of the Visually Impaired (TVI)

PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES

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).

PROJECT REACH:
ILLINOIS DEAF-BLIND SERVICES

What did participants think?

(5 point Likert scale- 5 = Strongly Agree)

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

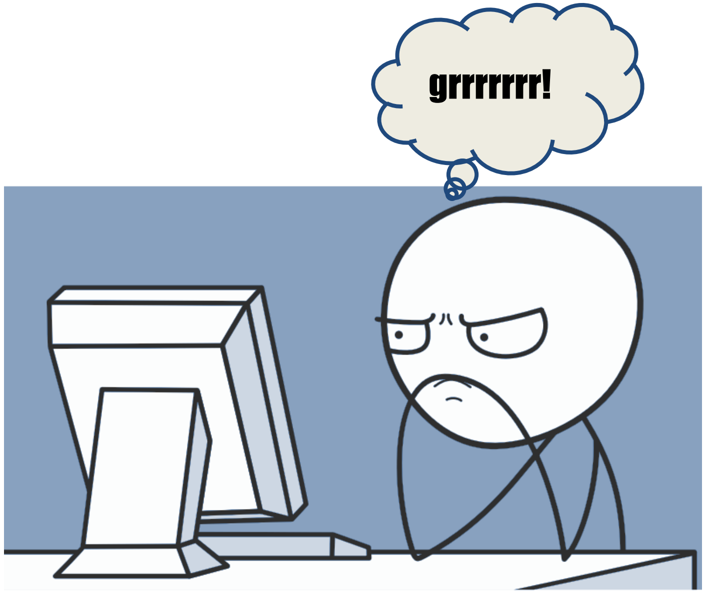
The ideas and activities were relevant to the field of deaf-blindness: 4.87

I found the information useful: 4.85

PROJECT REACH: ILLINOIS DEAF-BLIND SERVICES

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

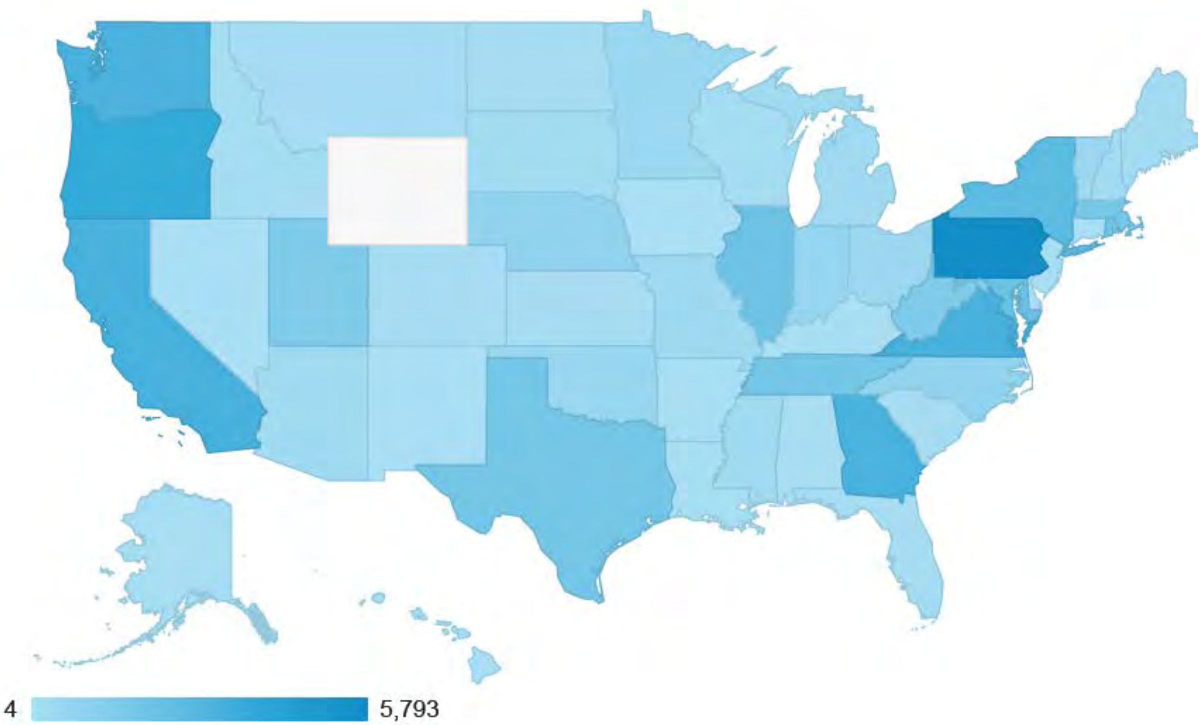
29 Host Training Sessions Completed

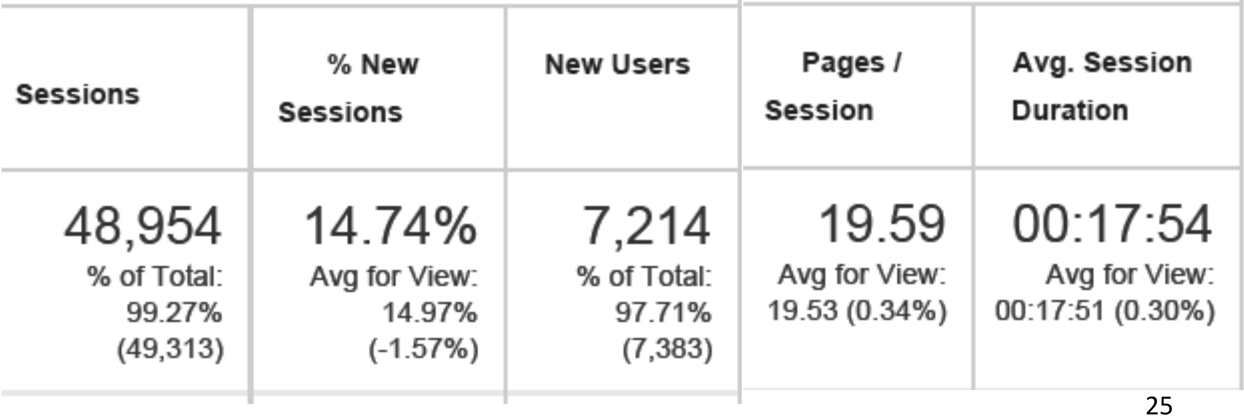
A Sample of Users- Demographics
N= 593

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)

Where have they logged on?

Jan 1, 2014 - May 11, 2015





Sessions	% New Sessions	New Users	Pages / Session	Avg. Session Duration
48,954 % of Total: 99.27% (49,313)	14.74% Avg for View: 14.97% (-1.57%)	7,214 % of Total: 97.71% (7,383)	19.59 Avg for View: 19.53 (0.34%)	00:17:54 Avg for View: 00:17:51 (0.30%)

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%

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/professionalstandards

Resources

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

Questions?

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Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Let the Games Begin: Choosing appropriate games for your child with CHARGE Syndrome

Sarah Bis, MS, OTR/L, C/NDT and Sharon Stelzer, M.Ed,
Perkins School For The Blind

Presenter Information:
Sarah Bis is a registered, licensed occupational therapist currently employed at Perkins School for the Blind's Deafblind Program in Watertown, MA. She has over 4 years of experience working with students with CHARGE Syndrome ages 3-22, in both school and residential settings. Sarah holds certification in Neurodevelopmental Treatment of children with cerebral palsy and other neurologically-based disorders. She also has extensive experience with evaluation and treatment of sensory processing in children.

Sharon Stelzer is a teacher at Perkins School for the Blind in the Deafblind Program in Watertown, MA. She has been teaching for thirty years. She has worked for over twenty years teaching students with CHARGE Syndrome from ages six to twenty-two. Sharon has presented both Nationally and Internationally on Teaching Strategies, Communication Strategies and Literacy Skills for students who are Deafblind and have CHARGE Syndrome. She has her Masters of Education Degree from Boston College with specialization in Deafblindness.

Presentation Abstract:
Parents and caregivers may often find it frustrating to choose appropriate games and leisure activities with their children with CHARGE Syndrome. In this presentation, parents will gain ideas for games that may be appropriate for their child. The presenters will review critical features of games and how to pre-teach skills to get children ready to participate in playing games.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Breathing and Posture: More Linked Than You Think!

Mary Massery, PT, DPT, DSc

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

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

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

How to talk to your child about having CHARGE

Kasee Stratton, Ph.D., NCSP, 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. Prior to joining faculty at Mississippi State University, Dr. Stratton completed her pre-doctoral and post-doctoral fellowship at Johns Hopkins School of Medicine and the Kennedy Krieger Institute. Dr. Stratton carried a heavy caseload of individuals with a variety of developmental concerns and helped many families start the discussion with their child about their syndrome. This work continued in 2013-2014 as Dr. Stratton was employed as a licensed psychologist at a center for children with disabilities.

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

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

David Brown and Julie Maier, Educational Specialist with California Deaf-Blind Services

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

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

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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 is the1st President of the German CHARGE Family Support Group and mom of a 10-year-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 Vice president of the German CHARGE Family Support Group and mom of a 15-year-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 self-awareness and self-confidence of the participants within their peer-group. Special guided outdoor-activities 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?!**”

We’ve got CHARGE – so what!

How young adults with CHARGE outshone themselves

In July 2013 a group of nine young adults with CHARGE met for the first time. Aged between 12 and 23 years, they took part in a weekend meet getaway - without their parents. The program for this first CHARGE Youth Weekend included a climbing challenge and a filmmaking project. The youth group was accompanied by 9 supervisors.

The young adults wanted to prove that, in spite of their limited seeing and hearing abilities and a lack of sense of balance, they could master the challenges posed by a climbing exercise. Even though the guide book describes the selected climbing route as short and easy, it was a formidable challenge for the young CHARGE people. The paths were narrow and uneven, and we had to climb over steps, stones and trees and even jump over a small stream. The young people managed to conquer all these hurdles by helping each other. They learnt how to clip onto the steel hawsers safely, and climbed a triumphant height of 175 m.

During the film project the young CHARGE people had so much, almost too much, to say! They felt it was very important to “tell the whole world” about CHARGE Syndrome and to make it quite clear that they “only” have CHARGE...
In one-to-one interviews and group interviews the young adults explained about themselves to the film team, describing living with CHARGE Syndrome from their point of view. They spoke about problems they have encountered in school, in work experience, and in their working lives. A common thread throughout the interviews was how much these young people enjoy life, even if it is a bit different.

A further highlight of the weekend scheduled for Sunday morning was a particularly unusual challenge – abseiling off the Youth Hostel balcony! The ropes hung all the way from the second floor right down to the cellar, just waiting for the young participants to abseil using their safety kit. Both participants and supervisors were thrilled and amazed at how confidently some mastered the art of abseiling.

CHARGE
CHARGE Syndrom e.V.

They outshone themselves. Everyone showed amazing courage and can proudly say

“We´ve CHARGE – so what!”

The weekend was a fantastic, unique experience for the supervisors as well. The Youth Weekend has since become an annual event.

The video from the filmmaking project 2013 is entitled „Wir haben CHARGE, na und!“
(“We’ve got CHARGE – so what!”)

and can be found on the **www.charge-syndrom.de** homepage.
There are subtitles in German and in English.

Claudia Junghans * Julia Benstz * CHARGE Syndrom e.V. * Germany * www.charge-syndrom.de

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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

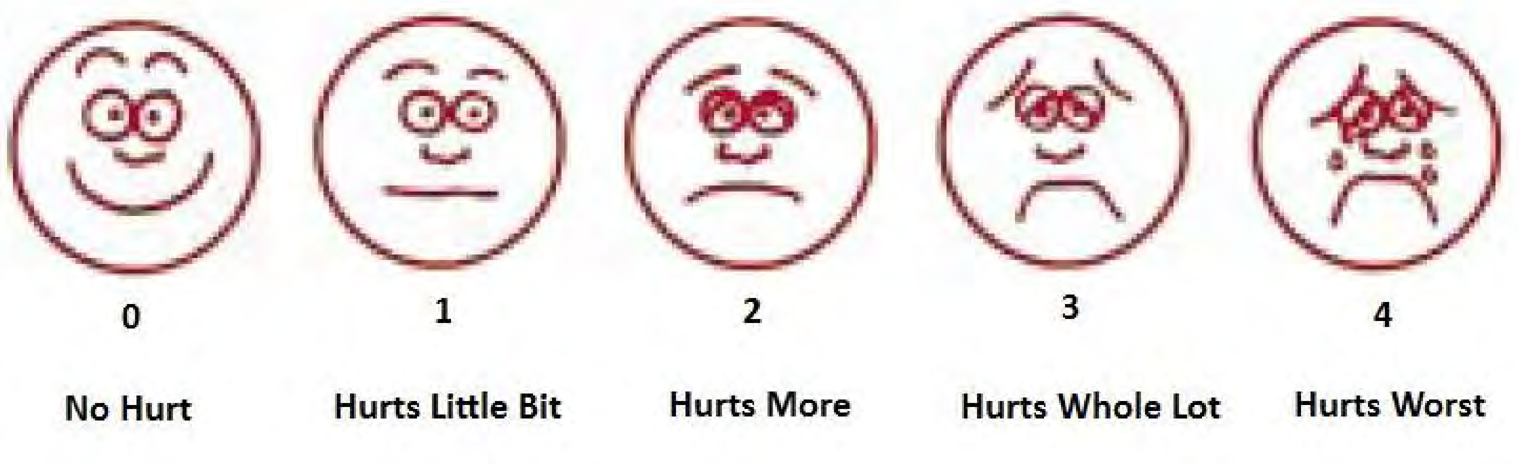
Pain Experience

- Acute
- Chronic
- Surgeries/Hospitalizations
- 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

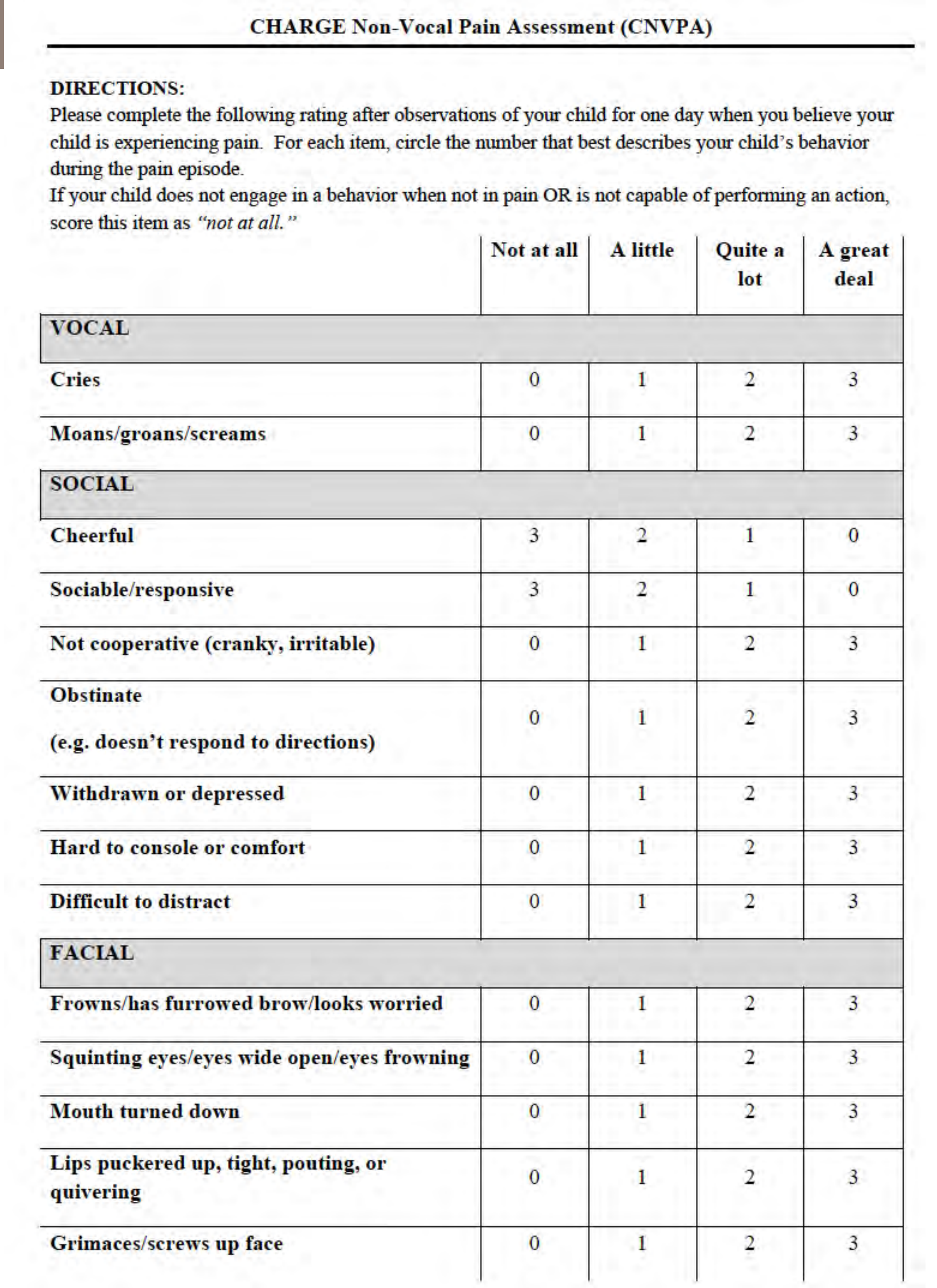
Study Conclusion

- 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



CHARGE Non-Vocal Pain Assessment (CNVPA)

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

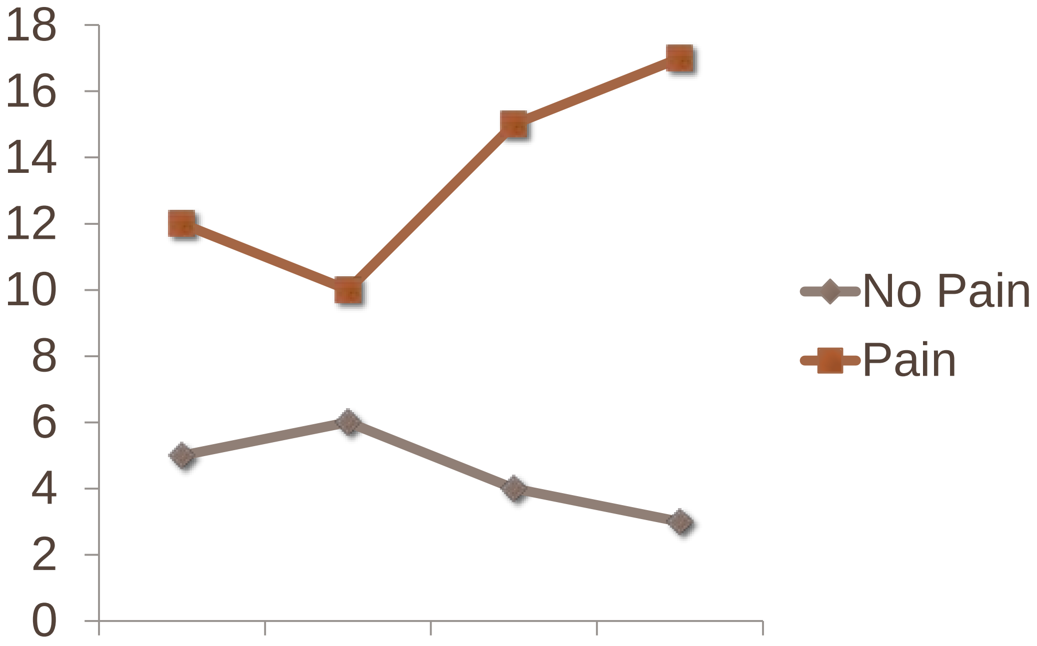
“But what about age?”

- 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



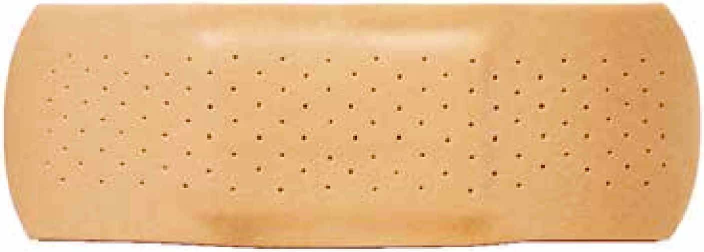
Pain Coping

“…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

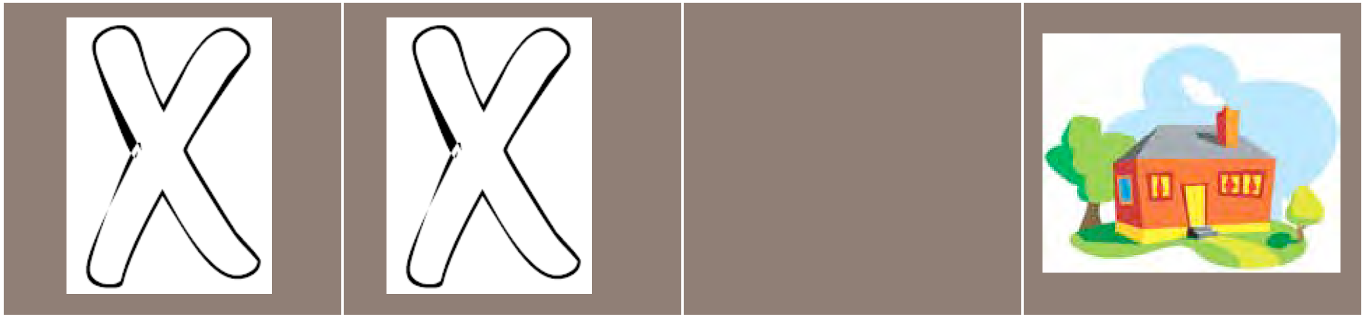


Preparing for Pain

- 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.

BULLDOG CHARGE SYNDROME RESEARCH LAB

Current projects:

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

facebook.com/BulldogCHARGELab

@BulldogCHARGE

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Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

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.

Friday, July 31, 2015
INFORMATION CENTRAL SESSION
2:15 - 4:30pm
Schaumburg East

Experiences with feeding and gastrointestinal motility in children with CHARGE syndrome

Meghan Macdonald, Faculty of Medicine, Dalhousie University, Halifax NS, Alexandra Hudson, Faculty of Medicine, Dalhousie University, Halifax NS, Angela Bladon, Feeding and Swallowing Team, McMaster Children's Hospital, Hamilton Ontario, Canada, Elyanne M. Ratcliffe, Division of Gastroenterology and Nutrition, Department of Pediatrics, McMaster University, Hamilton Ontario, Canada, Kim Blake, Department of Pediatrics, Dalhousie University, IWK Health Centre, Halifax NS

Presenter Information:
Meghan is a medical student at Dalhousie University, in her second year of study. Prior to attending medical school she completed undergraduate degrees in both biology and psychology, with research experience in training hypoglycemia detection dogs (i.e. diabetes alert dogs). This stimulated her research interests in clinical pediatrics. During her pre-med time she also worked on various medical education research projects with Dr. Blake. While in medical school she was given the opportunity to participate in clinically directed pediatric research, which led to the development of this CHARGE syndrome research project with Dr. Blake.

Presentation Abstract:
This research focuses on the eating and gastrointestinal motility issues that children with CHARGE syndrome experience. Data was collected via internet questionnaires completed by CHARGE families from around the world. Eating difficulties and gastrointestinal symptoms are prevalent for the majority of children and are completely tube fed. The results of this study may influence home and medical management of children with CHARGE syndrome.