



**Saturday, August 1, 2015
Breakout Session #26: 10:45 - 11:45am
Schaumburg A-B**

**Promoting Early Concept Development
through Adapted Books**

**Christine Spratling
Georgia PINES**

Presenter Information:

After graduating with a Bachelor of Science degree in Special Education for students who are deaf/hard-of-hearing, Christine started teaching in the public school systems in 1986. Since then, I have taught in a variety of settings from self-contained to itinerant and added a Master's Degree in interrelated special education and an EDS in educational leadership.

About 6 years ago, I also began to work for Georgia PINES as a parent advisor in an early intervention setting. Georgia PINES serves families of young children with vision and/or hearing loss including those with CHARGE Syndrome.

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

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

Presentation Abstract:

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

Promoting Early Concept Development

{ Through adapted books

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



“For people who can see and hear, the world extends outward as far as his or her eyes and ears can reach. For the young child who is deaf-blind, the world is initially much smaller.”



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

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



Barbara Miles
Overview on Deaf-Blindness DB Link October 2008

Promoting Early Concept Development



to our world.

“Concepts are
the ideas that
give meaning

We develop concepts based on
our particular experiences.”

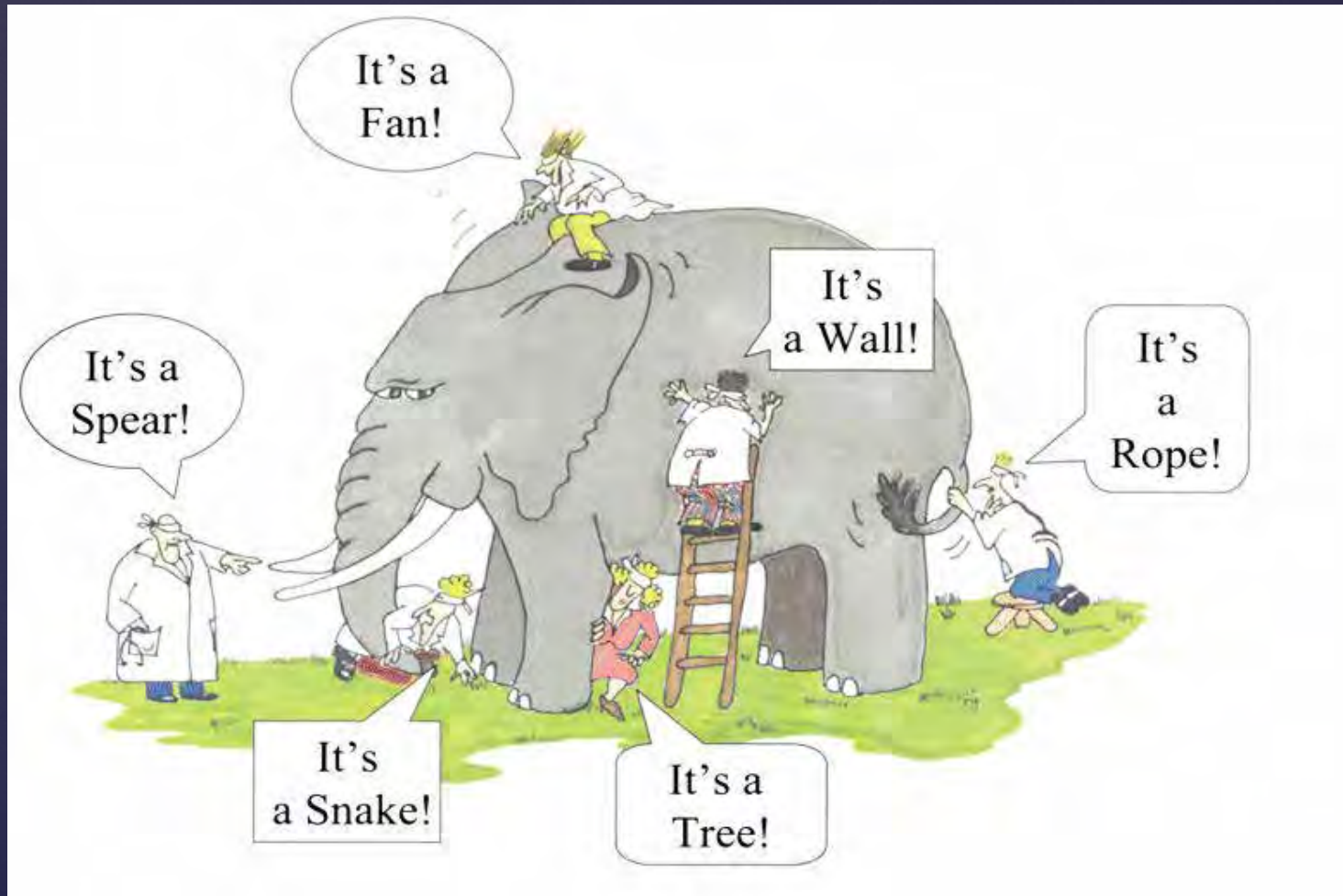
Barbara Miles

Developing Concepts with Children Who Are Deaf-Blind

DB Link 2008

Promoting Early Concept Development

What happens to concept development when we have limited experiences:



What do you see?



6

Promoting Early Concept Development

What does the literature say about concepts:

Three groups of concepts:

1. Concrete concepts (tangible things)
2. Semi-concrete concepts (something that can be demonstrated but is not tangible)
3. Abstract concepts

Types of concepts:

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

6 Areas of concept development:

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

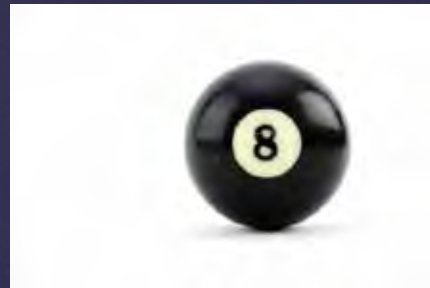
1. Objects exist



2. Objects have permanence



3. Objects differ



4. Objects have names or labels

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

5. Objects have characteristics:

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



6. Objects have functions or use



O&M Concepts

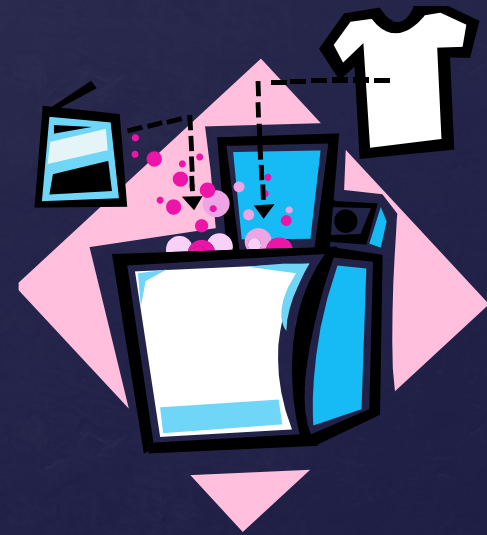
- **Body Image Concepts**
- **Spatial Concepts**
- **Environmental Concepts**

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

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

Difference between concepts and skills:

- Skill - the ability to do something
- Concept – the ideas that help us understand “why”



How to choose a concept to work on:

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

Some assessments for concept development:

- 1. School Inventory of Problem Solving Skills (SIPSS)**
- 2. Home Inventory of Problem Solving Skills (HIPSS)**

<http://www.designtolearn.com/products>

3. Functional Scheme: Functions Skills Assessment

<http://www.lilliworks.com>

Strategies that help with concept development:

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

Planning instructional activities for concepts:

Weeks of: _____ through _____

CONCEPT: _____

O&M Skill Development / Real Life Applications

1. Whole body:

2. Object-to-body:

3. Objects-beyond-the-body:

Concept Category (check one):

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

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

Promoting Notes: _____

CONCEPT :Up/Down**O&M Skill Development / Real Life Applications**

1. Whole body: Using stairs/steps, jumping, going up steps/down slide, swinging up/down, stand up/sit down, arms up, legs up, head up, etc.
2. Object-to-body: Brushing teeth, Brushing, Pulling up/down sock, pants, pull-ups,
3. Objects-beyond-the-body: I can throw things up and they will come back down.

Concept Category (check one):

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

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

Notes:

Concept-based activity ideas for “Up and Down”

5 Little Monkeys Jumping on the Bed

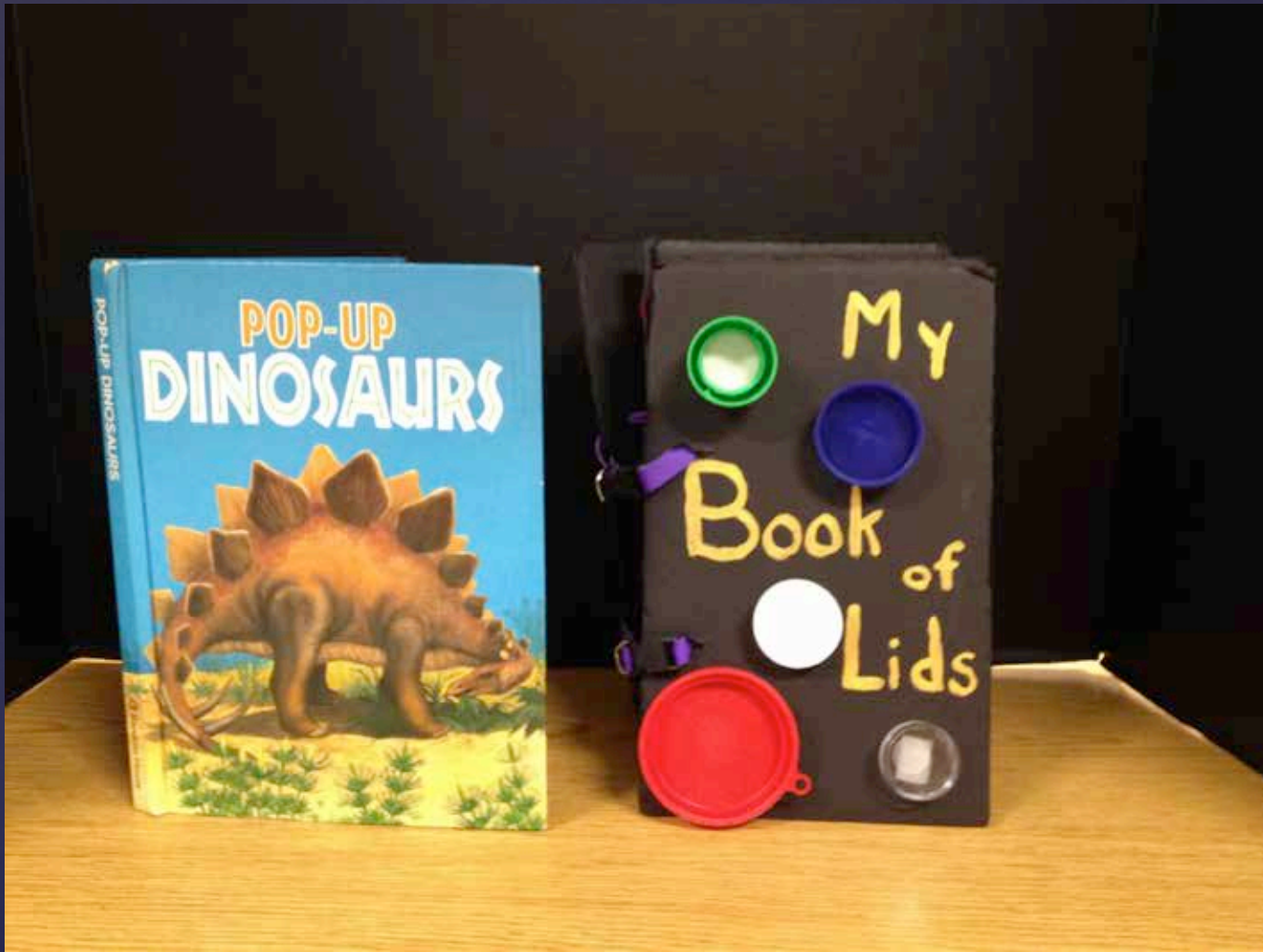
According to Barbara Miles, concepts are the ideas that give meaning to our world. People who can hear and see, experience the world as far as their eyes and ears reach. For our PINES babies who have a hearing or vision loss, the world becomes smaller depending on their sensory loss. For our babies who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

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

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

Concept Category	Activity Suggestions
1. Baby's own body (whole body up and down experiences)	<ul style="list-style-type: none"> * Bounce baby up and down on your lap. * Lift baby up and down. * Body parts can go up and down (arms, legs, hands, etc.) * Stand up and sit down. * Jump up and down, etc.
2. Objects touching the baby's body	<ul style="list-style-type: none"> * Clothes go up and down on the body, such as socks and pants. * Zippers go up and down. * Tickle up and down on baby's arms, legs, etc. * Move the monkeys up and down baby's arms, legs etc.
3. Objects beyond the baby's body Book extension ideas	<ul style="list-style-type: none"> * Use the monkeys to jump up on the sponge bed and fall down. * The monkeys on the clothespins can "jump" up and down on the book or fall down from the book. * Balloons, bubbles, balls, etc, go up and down.

Everybody has a special interest



Concept book for “on top”



Concept book for “up and down”



Also for “up and down”:



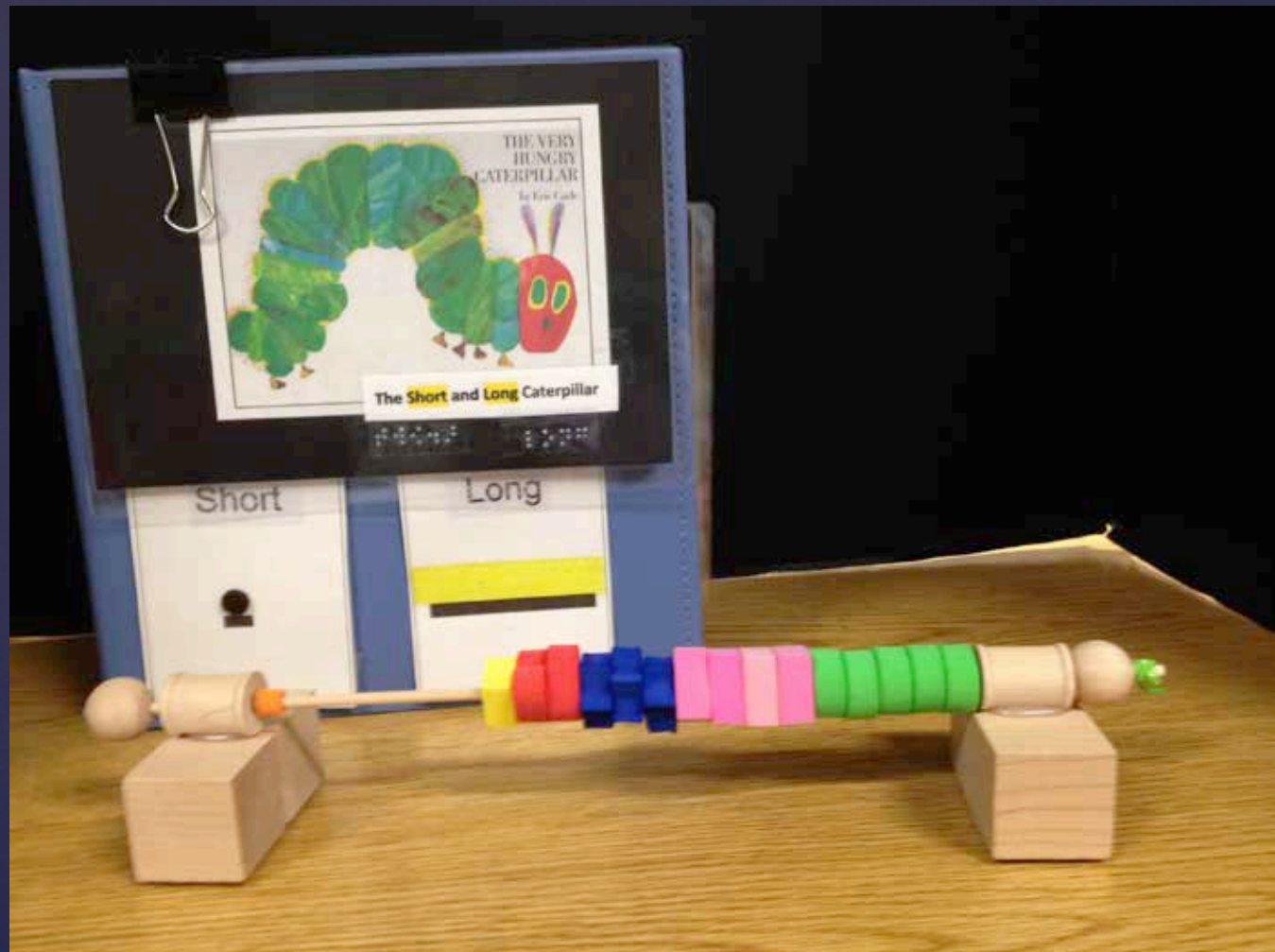
Concept book for “rolling”



Concept book for “wet/dry”



Concept book for “long/short”



Resources:

1. Concept Development

http://www.sparkle.usu.edu/Topics/concept_development/index.asp

2. **Developing Concepts with Children Who Are Deaf-Blind**

Barbara Miles, M.Ed. & Barbara McLetchie, Ph.D.
DB-Link February 1, 2008

3. Early Concept Development

by Holly Cooper, <http://www.tsbvi.edu/preschool/1117-early-concept-development>

More Resources:

4. Understanding Deaf blindness : Issues, Perspectives, and Strategies --Alsop, Linda, M.Ed. (Ed.) Logan, UT: SKI-HI Institute, Utah State University. (2002)
5. What a Concept!
Jim Durkel, CCC-SPL/A and Statewide Staff Development Coordinator (with help from Kate Moss, Stacy Shafer and Debra Sewell) Texas School for the Blind and Visually Impaired Outreach

More Resources:

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

by Barara Miles and Marianne Riggio, 1999



Saturday, August 1, 2015
Breakout Session #27: 10:45 - 11:45am
Schaumburg C-D

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

Linda Alsop, M.Ed.
**SKI-HI Institute, Center for Persons with
 Disabilities, Utah State University**
Beth Kennedy, M.Ed.

**DeafBlind Central: Michigan's Training &
 Resource Project**

**Parent Panel: Tim and Nancy Hartshorne,
 Paul and Jodie Beavers, Cheryl Kruger**

Presenter Information:

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

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

Presentation Abstract:

Interveners are individuals who work one-to-one with children and youth who are deafblind and who have training and specialized skills in deafblindness. The practice of using interveners as individualized supports for children with combined vision and hearing loss in educational settings is growing across the country, and this is creating an ongoing need to provide information and training to parents about the critical role that interveners play in the education of their children. You know that an intervener is what your child/student needs- but how do you make it happen? This presentation will provide parents and other members of the team with important information for making the case for intervener services. A parent panel will share personal stories of how interveners were added to their children's Individualized Education Plan (IEP), and what a difference having an intervener has made for their child.

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

Interveners are for people who are Deaf-Blind:

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

Interveners have training and a specific role:

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

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

Advocating for an intervener:

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

More information is available:

- To learn more, visit www.intervener.org.
- Find them on Facebook.



Saturday, August 1, 2015
Breakout Session #28: 10:45 - 11:45am
Schaumburg West

Sex Hormones and Teaching Safe Sexuality

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

Presenter Information:

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

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

Presentation Abstract:

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



Saturday, August 1, 2015
Breakout Session #29: 10:45 - 11:45am
Schaumburg East

Neurological Issues in CHARGE Syndrome

Dr. Jules Constantinou, MD FRACP

Presenter Information:

Dr. Jules Constantinou is Director of Pediatric Neurology at the Center for Autism and Developmental Delay and the Comprehensive Epilepsy Program in the Henry Ford Health System in Detroit. He completed Neurology Residency in Australia where he worked with Dr. George Williams, well known to the CHARGE community, and Neurology Fellowship at Boston Children's Hospital.

Presentation Abstract:

Dr. Constantinou will discuss a variety of issues relating to neurology and CHARGE syndrome with time for questions and answers.



Saturday, August 1, 2015
Breakout Session #30: 10:45 - 11:45am
Schaumburg E-G

**An interactive seminar in genetics: All
you want to know about the genetics of
CHARGE syndrome**

**Conny van Ravenswaaij-Arts, MD, PhD &
Nicole Corsten-Janssen, MD**
University Medical Center Groningen

Presenter Information:

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

Presentation Abstract:

After a short introduction, all questions of the participants regarding genetics, the CHD7 gene, mutations, recurrence risk, et cetera, will be answered using instructive illustrations. A handout that explains the main issues for a lay audience will be provided.

Based on the experience of previous years, we will offer parents the opportunity to discuss the specific genetic test results of their child with us in short sessions after the seminar.

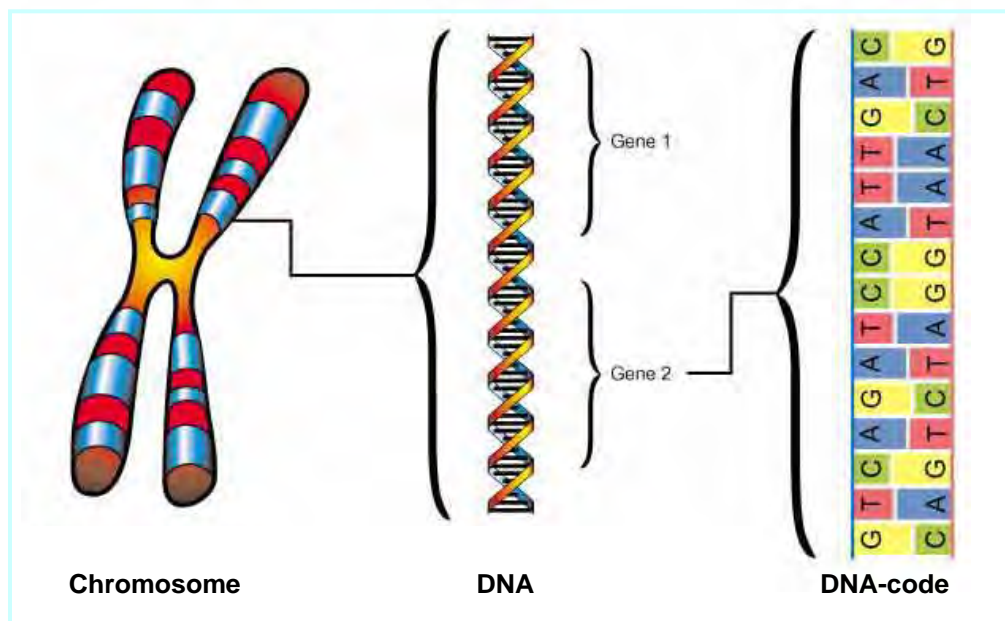
All you want to know about CHARGE and genetics

By Nicole Corsten-Janssen and Conny van Ravenswaaij-Arts
Department of Genetics, University Medical Center Groningen, The Netherlands
July 2013

What are chromosomes, genes, DNA and mutations?

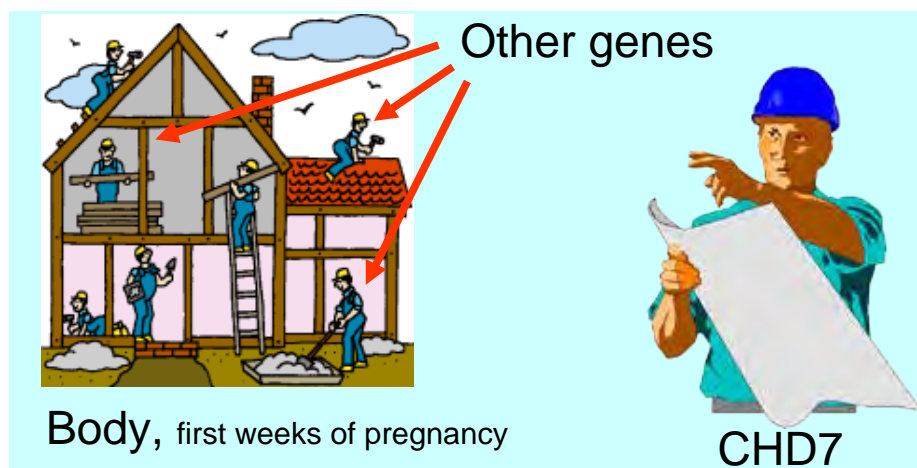
Our genetic information is tightly packed up on structures called chromosomes. Humans have 46 chromosomes grouped into 23 pairs. Everyone inherits 23 chromosomes from his father and 23 chromosomes from his mother. A chromosome consists of tightly packed up DNA.

DNA is our hereditary material and it is made up of four chemical bases (A,C,T,G). Genes are the pieces of DNA that hold the code for proteins, which are important for all kinds of functions in the body. A mutation is another word for a change in the DNA code, such a change in the DNA code can led to a change in the function of the proteins.



What is the *CHD7* gene and what does it do?

Since 2004 we know that CHARGE syndrome is caused by a change (mutation) in the *CHD7* gene. Every person has two *CHD7* genes, one inherited from their father and the other from their mother. *CHD7* is a regulatory gene. It regulates the work of developmental genes very early on, during the development of the fetus. If there is insufficient CHD7 protein being made, there is a higher risk of developmental defects occurring in specific organs like the heart, eye, ear, kidney, etc. A change in one of the two *CHD7* genes is enough to result in CHARGE syndrome. However, the syndrome is highly variable and it is not possible to predict the clinical consequences for the child from a specific change in *CHD7*.



How is CHD7 analysis done?

CHD7 analysis is performed on DNA. DNA is usually extracted from blood cells, but other tissues, e.g. skin or saliva, can also be used. Different types of mutations can be present in the *CHD7* gene. Most of these mutations will be detected by routine DNA analysis (called “sequencing”). Sometimes a part of the *CHD7* gene may be missing or even the whole gene (called a “deletion”). Deletions of *CHD7* are rare and occur in approximately 1% of CHARGE patients. They cannot be found by routine DNA analysis, but can be identified by other techniques (e.g. array, MLPA).

Type of mutations

Normal gene	Tim and his toy
Point mutation	Pim and his toy
Deletion	Tim and toy
Insertion	Tim and his big toy
Frameshift	Tim ndh ist oy

Why perform DNA analysis?

There are several reasons to perform DNA analysis:

- To provide certainty
- Confirm a tentative diagnosis in a child with atypical features (these two reasons have consequences for the clinical follow-up)
- Confirm the diagnosis, so that parents or siblings know their recurrence risk
- Prenatal options

What if no *CHD7* mutation is found in my child?

- A diagnosis of CHARGE syndrome can be made by identifying a *CHD7* mutation, or by looking at the clinical criteria for the syndrome. If your child fulfills the clinical criteria, he/she has CHARGE syndrome, irrespective of the results of *CHD7* analysis.
- Some other syndromes have clinical features that overlap with CHARGE syndrome and it is important to exclude these syndromes from the diagnosis.
- Current techniques are not good enough to identify all *CHD7* mutations.

- Other genes might also contribute to CHARGE syndrome (see below).

What is the risk that CHARGE syndrome will re-occur if we have another child?

Since familial CHARGE syndrome is extremely rare, the recurrence risk for parents who have a child with CHARGE syndrome is, in general, low. If parents want to learn more about their risk for future pregnancies, we recommend:

- A geneticist should see if the parents have any mild symptoms of CHARGE syndrome (hearing, balance, smell, shape of the ears).
- If a *CHD7* mutation has been found in a child, the parents can also be offered DNA analysis.

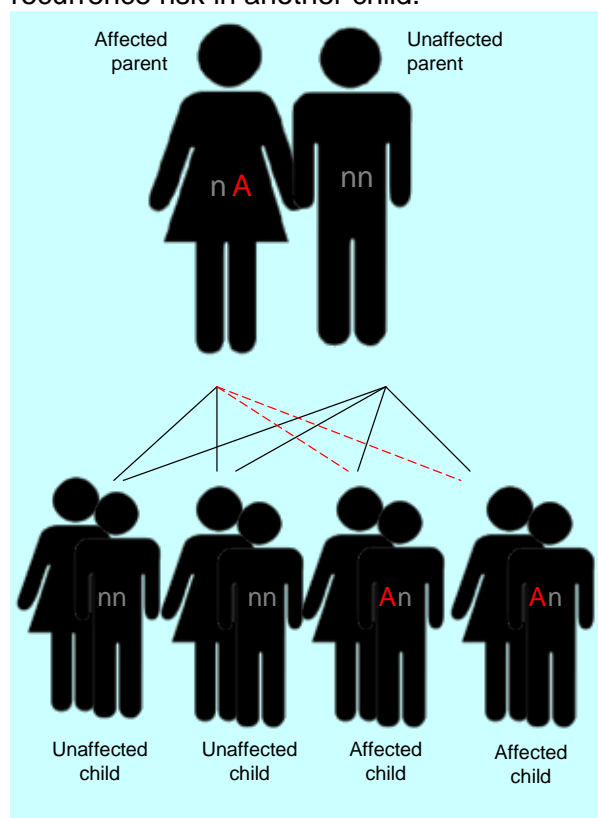
There are three possibilities:

1. Parent does not have CHARGE syndrome or a *CHD7* mutation

If the *CHD7* change is not found in one of the parents (the most common situation), there is still a small risk of carrying a *CHD7* mutation ("germline mosaicism"), so the recurrence risk is not zero, but 1% or 2%.

2. Parent has CHARGE syndrome

If one of the parents has CHARGE syndrome, the recurrence risk varies. Parents pass half of their genetic information on to their children. The parent with CHARGE syndrome can pass on either their normal *CHD7* gene or their gene with a mutation. This means that there is a 50% recurrence risk in another child.



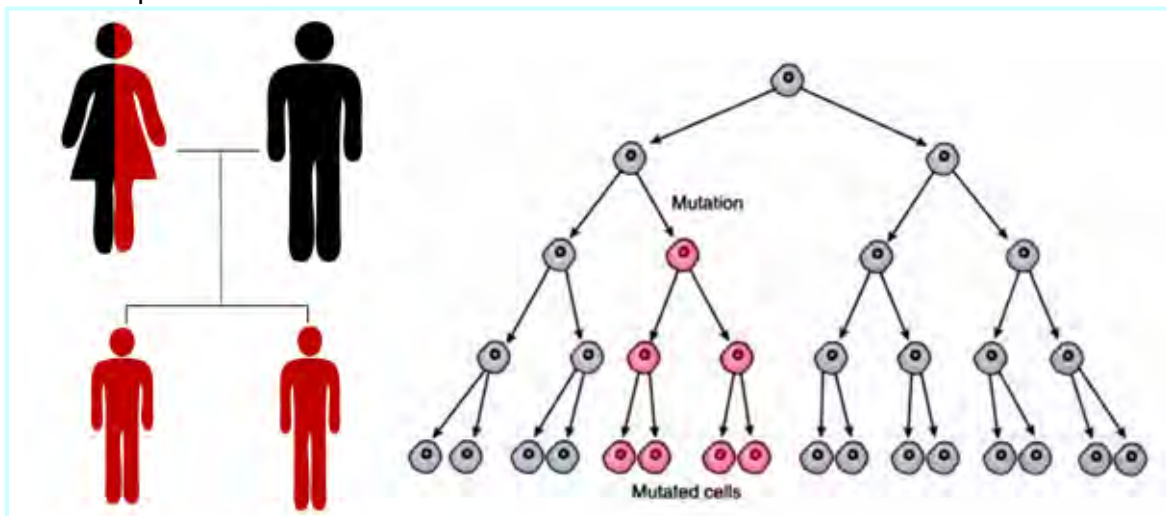
*The parent with mild CHARGE syndrome passes on the normal *CHD7* gene (n) or the abnormal *CHD7* gene (A). The recurrence risk is 50% for each pregnancy.*

3. Parent with a *CHD7* mutation in some of their cells ("mosaicism")

Very rarely, a family may have two affected children with the same *CHD7* change, while the parents do not have any features of CHARGE syndrome. How can this happen? We may find that one of the parents carries a change in the *CHD7* gene in only some of his/her body cells. This is called mosaicism and it can occur when the fertilized egg has a normal *CHD7* gene, but a spontaneous change occurs later in one cell as the cells divide and grow (see figure). Only the cells coming from that cell with the changed *CHD7* gene will have the mutation. If these

cells are also present in the parent's ovaries or testes, egg or sperm cells with the *CHD7* mutation can be formed and lead to the pregnancy of a child with CHARGE syndrome.

If a parent has a mosaic form of the *CHD7* mutation, the recurrence risk for further children can be up to a maximum of 50%.



*In this example of mosaicism in the mother, there is a *CHD7* change (mutation) in some of her body cells. The scheme on the right shows that this can occur when the change (red cells) arises spontaneously during a cell division after fertilization. The mother will not have CHARGE syndrome herself, but she does have an increased risk of having affected children because she can pass on the *CHD7* change via an egg cell. The subsequent child will have the *CHD7* change in all its cells and will therefore have CHARGE syndrome.*

Options in a subsequent pregnancy

Parents who have a child with CHARGE syndrome in whom a *CHD7* mutation has been identified, may choose to have prenatal diagnosis performed for a subsequent pregnancy. But please remember that DNA analysis cannot predict the severity of CHARGE syndrome. A fetal ultrasound can provide extra information, like whether the baby has a heart defect, but ultrasound cannot “see” everything – deafness, developmental delay and behavioral problems cannot be detected.

The choice to have prenatal diagnosis is a personal one, and the geneticist must inform the parents well, so that they can make a choice they feel good about.

Is *CHD7* the only gene causing CHARGE syndrome?

In 5% to 10% of patients with typical CHARGE syndrome, no mutation in the *CHD7* gene can be found. This percentage is higher in patients who do not fulfill the clinical criteria for CHARGE syndrome (atypical presentation). So there may well be other genes that are also responsible for CHARGE or CHARGE-like syndrome. Mutations in a gene called *SEMA3E*, had been identified in two patients. A few other genes, like *CHD8* (a “candidate gene”), have been studied, but no mutations were found in these genes in CHARGE syndrome patients. Recently, we started to search for other genes that might cause CHARGE syndrome using a new technique, called next-generation sequencing or whole exome sequencing. This technique makes it is possible to look for mutations in all the known genes in a single test.

Any questions? Please come and ask us during the CHARGE conference, or send an email to CHARGE@umcg.nl.

More information on our research projects on CHARGE syndrome is available at www.rug.nl/research/genetics/research/chargesyndrome/





**Saturday, August 1, 2015
Breakout Session #31: 1:00 - 2:00pm
Schaumburg A-B**

**Helping out at Home: Building
Independent Living Skills in the
Context of Family Routines**

**Sarah Bis, MS, OTR/L, C/NDT
& 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 with multi-sensory impairment. She has a Master of Science Degree in Occupational Therapy.

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:

Independent Living skills are a critical part of the Expanded Core Curriculum. Unlike typically developing children who may learn through observation, children with CHARGE syndrome will need to be directly and explicitly taught these skills. In this presentation, parents and caregivers will learn how to break down independent living tasks and embed teaching and practice into everyday routines. Participants will leave the session with a list of ideas that may be implemented at home immediately.

HELPING OUT AT HOME:

BUILDING INDEPENDENT LIVING
SKILLS IN THE CONTEXT OF FAMILY
ROUTINES

Presented by Sarah Bis, MS, OTR/L, C/NDT

Sharon Stelzer, M.Ed.

August 2015

WHAT ARE INDEPENDENT LIVING SKILLS?

☞ Activities of Daily Living (ADL)

☞ Bathing

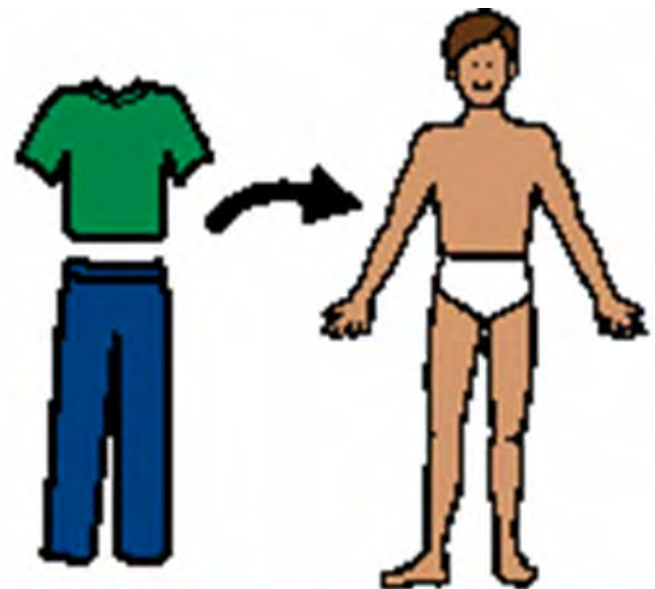
☞ Dressing

☞ Grooming

– Tooth & Hair Care

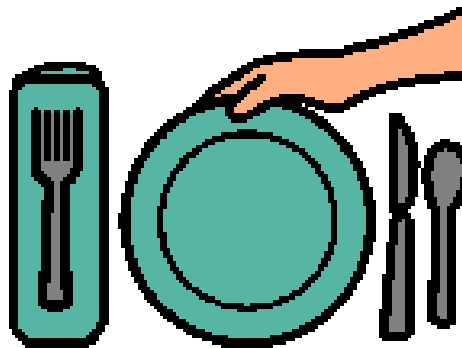
☞ Eating

☞ Toileting



WHAT ARE INDEPENDENT LIVING SKILLS?

- ☞ Instrumental Activities of Daily Living (IADL)
- ☞ Meal preparation
- ☞ Housekeeping
- ☞ Washing Dishes
- ☞ Laundry
- ☞ Shopping



WHY IS THIS TOPIC IMPORTANT ????

- ⌘ While many typical children are able to learn through observation, our children must be explicitly taught independent living skills.
- ⌘ Our end goal is for our children to function as independently as possible.
- ⌘ Lifelong skills, lots of practice, purposeful in how taught, they will not change / unlearn habits learned early
- ⌘ Daily routines provide an opportunity for **consistent** practice of skills in a familiar context and **predictable** environment, which leads to optimal learning.
- ⌘ Benefits across areas of development - Sequencing and Organization, Communication, Motor Skills, Sensory Processing, Reading, Math, Science



EXPANDED CORE CURRICULUM

- ¢ Compensatory Skills
- ¢ Functional Academics
- ¢ Orientation & Mobility
- ¢ Social Interaction
- ¢ Independent Living
- ¢ Career Education
- ¢ Recreation & Leisure
- ¢ Assistive Technology
- ¢ Sensory Efficiency
- ¢ Self-Determination



ROUTINES AND LEARNING

- ⌘ Routines are predictable and help us to organize our experiences. They help us understand and anticipate expectations.
- ⌘ When children can anticipate what will happen next, they are less stressed, and can use their energy on learning and communication.
- ⌘ Routines increase a child's sense of confidence and control over the environment.
- ⌘ Routines build a foundation of memories for additional learning.
- ⌘ For caregivers, it is easiest to embed teaching opportunities into activities that you already complete each day!



WHAT DO YOUR OTHER CHILDREN HELP WITH AT HOME?



CHORES *by age*

TODDLER

2-3 YEAR OLDS

Make bed
Take clothes to laundry
Put clean clothes away
Clean up toys

PRESCHOOLER

4-5 YEAR OLDS

Load dishwasher
Set table
Water plants
Help feed pets
Match socks
Clean room

ELEMENTARY

6-9 YEAR OLDS

Sweep
Sort laundry
Wipe counters
Help with meal prep
Empty dishwasher
Pull weeds

TWEEN

10-12 YEAR OLDS

Take out trash
Fold Laundry
Vacuum
Mop
Clean toilets
Make easy meals

TEEN

13+

Make meals/meal plan	Mow lawn
Clean fridge	
Babysit	



HAVE HIGH EXPECTATIONS FOR YOUR CHILD WITH CHARGE!!!



USING BEST PRACTICE TEACHING STRATEGIES

- ⌘ Task Analysis
- ⌘ Backward Chaining
- ⌘ Partial Participation
- ⌘ Routine/Consistency
- ⌘ Active vs. Passive
- ⌘ Beginning-Middle-End
- ⌘ Turn Taking
- ⌘ Negotiation



TEACHING STRATEGIES

⌘ Task Analysis

- Break task into smaller sequential components
- What components can the child do independently?
- For which components does the child require assistance?

⌘ Backward Chaining

- Break task into smaller sequential components
- Start by teaching the last step of the sequence
 - ⌘ Start to teach bed making by adding pillows, then teach pulling up comforter, then work on sheets
 - ⌘ Child feels successful at completion of task

⌘ Partial Participation

- Have the child participate in the pieces of the task that they can successfully complete.

⌘ Routine and Repetition

- Routines help build memory skill
- Task should be presented the same way each time
- Same sequence and cues
- Different caregivers working with the child need to use the same types of cues



TEACHING STRATEGIES (CONT.)

⌘ Active vs. Passive

- Alternate more passive seated activities with more active movement-oriented activities
 - ⌘ Remove clothes from dryer (Active)
 - ⌘ Sitting or Standing to fold laundry (Passive)
 - ⌘ Put away / hand up clothing (Active)

⌘ Beginning-Middle-End

- Establish clear cut set-up, doing of the activity, and clean-up of activity
- Builds anticipatory skills
- Facilitates organization and sequencing
- Fosters clear expectations

⌘ Turn Taking

- Break up task for reluctant chore participants
- Motivating
- Within a chore or for popular/unpopular chores

⌘ Negotiation

- Length, time
- Helps with motivation
- Helps build leadership skills
- Builds self-confidence and self-empowerment



WHAT **CAN** OUR CHILDREN DO?

PARTIAL PARTICIPATION

Child's Ability	Relevant Task
Move limbs	Push arms and legs in and out of clothes, lift body parts for bathing
Identify own body parts	Wash appropriate body parts, toothbrush in mouth, bring hairbrush to hair
Hold items with or without assistance	Use washcloth, toothbrush, hairbrush, cup, spoon
Put in and take out	Trash in bin, laundry in hamper, toys in finished bucket or toy chest, load and unload dishwasher
Carry	Bring laundry to washer/dryer, bring toys to toy box, set place at table, bring dishes to sink
Sort/ Match	Sort silverware, match socks

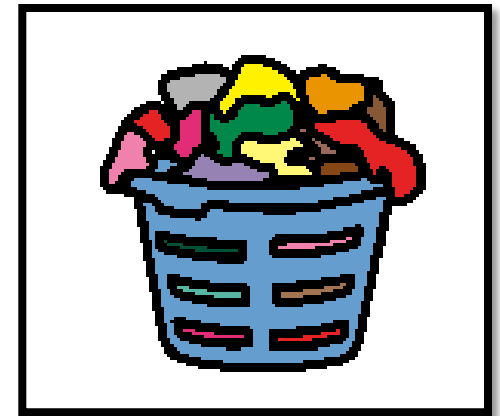


ADDITIONAL/ ADAPTIVE SUPPORTS

- ⌘ Talk to your child's therapists or providers for more ideas about what activities your child may be able to complete at this time.
- ⌘ Explore adaptive equipment
 - Bath mitt vs. loofa or washcloth
 - Built up handles
 - Scoop or suction bowl
 - Laundry basket on wheels



CHORES IN ACTION



MEET THE FINISHED BUCKET

(YOUR NEW BEST FRIEND)

- ⌘ Materials do not drop, get thrown, or just disappear – they go into the finished bucket when the task is complete.
- ⌘ Signifies a clear ending to the activity.
- ⌘ Hand over hand assistance can be provided if needed.
- ⌘ Great way to practice grasp and release.
- ⌘ Embed into daily routines: play time, mealtime, undressing



CLEANING UP AFTER LUNCH!



DUSTING



HANG UP AND PUT AWAY CLOTHES



MAKING THE BED – BACKWARD CHAINING APPROACH



MEAL PREPARATION – CHOPPING VEGETABLES



MEAL PREPARATION – MIXING INGREDIENTS, STUFFING SHELLS



COOKING ON THE STOVE



GARDENING



WHAT ARE THE BENEFITS OF TAKING TIME TO ADDRESS INDEPENDENT LIVING SKILLS??



SEQUENCING AND ORGANIZATION

- ¢ From gathering materials to clean up, children learn that activities have a clear beginning, middle, and end.
- ¢ Activities are scheduled at specific times of day.
- ¢ Participating in familiar self-help activities at home can form the basis for a daily schedule or calendar system.

First, Bathroom **Then, Snack**



COMMUNICATION

- ⌘ Routines help children develop a memory and reference point for additional learning
- ⌘ We can communicate with our child about what has happened and what we are about to do next
- ⌘ Use objects, pictures, or language that relate to the routine to talk about it.



MOTOR SKILLS

- ⌘ Meaningful routines provide a purpose and motivation for movement
- ⌘ Through household routines children can practice navigating the environment
- ⌘ Children have opportunities to use two hands together to complete functional tasks.
- ⌘ Opportunities to practice more complex fine and gross motor movements



SENSORY PROCESSING

- ⌘ Routines help children with overall organization and self-regulation as they learn to anticipate what comes next
- ⌘ Through predictable routines, children can better learn to expect and tolerate light touch sensations during tooth brushing, hair brushing, and nail care.
- ⌘ Opportunities to cross the midline of the body, helps student coordinate use of both hemispheres of the brain and both sides of the body.



SENSORY PROCESSING (CONT.)

- ⌘ Opportunities to push, pull, and manipulate objects provides input to muscles and joints (the proprioceptive sense), helping children to better perceive the position of their body in space and how it relates to the world around them.



Where is my body in space?



ACADEMICS

- ⌘ Life long learning
- ⌘ Math skills: counting, sequencing, 1:1 correspondence, sorting, matching
- ⌘ Literacy: vocabulary development, reading, pre-reading (left to right, top to bottom, etc.)
- ⌘ Science: Concepts-hot/cold, on/off, cooking



HOW WILL YOU EMBED INDEPENDENT LIVING SKILLS INTO YOUR FAMILY ROUTINES?

- ¢ Time
- ¢ Place
- ¢ Skill
- ¢ Materials
- ¢ Cues



HELPFUL HINTS & TIPS

- ⌘ Use a chore chart (avoid power struggles)
- ⌘ Rotate chores (avoid boredom, increase skills)
- ⌘ Use incentives (motivation!)
- ⌘ Creative names, creative presentation (framing in a positive light)
- ⌘ Choose activities that meet your child's sensory needs (e.g. movement, tactile defensiveness)



TEACHER & OCCUPATIONAL THERAPIST PERSPECTIVES

- ¢ Make a PB & J sandwich
- ¢ Unload dishwasher-put away silverware
- ¢ Make the bed



THINGS TO THINK ABOUT...

- ¢ Who does the chores in our house?
- ¢ Do my other children help out? How?
- ¢ What are my expectations?
- ¢ What might get in the way? (sensory impairment, attention span, skill level, communication, time, etc.)
- ¢ Where do I start?
- ¢ How do I communicate to others that helping out is an important skill?



HELPFUL ONLINE RESOURCES

⌘ Perkins Scout: Daily Living Skills in Young Children

<http://www.perkinselearning.org/scout/daily-living-skills-young-children-blind-multiple-disabilities-deafblind>

⌘ National Center on Deaf-Blindness

- Library
- Learning and Instruction
- Daily Living Skills

<https://nationaldb.org/library>



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**Saturday, August 1, 2015
Breakout Session #32: 1:00 - 2:00pm
Schaumburg C-D**

**Demonstrating the Communication
Matrix Online Assessment &
Community of Practice: Applications
for Children with CHARGE Syndrome,
Their Families, and Educators**

**Alexandria Cook & Charity Rowland, Ph.D.
Oregon Health & Science University
Amy Parker, Ph.D.
National Center on Deaf-Blindness**

Presenter Information:

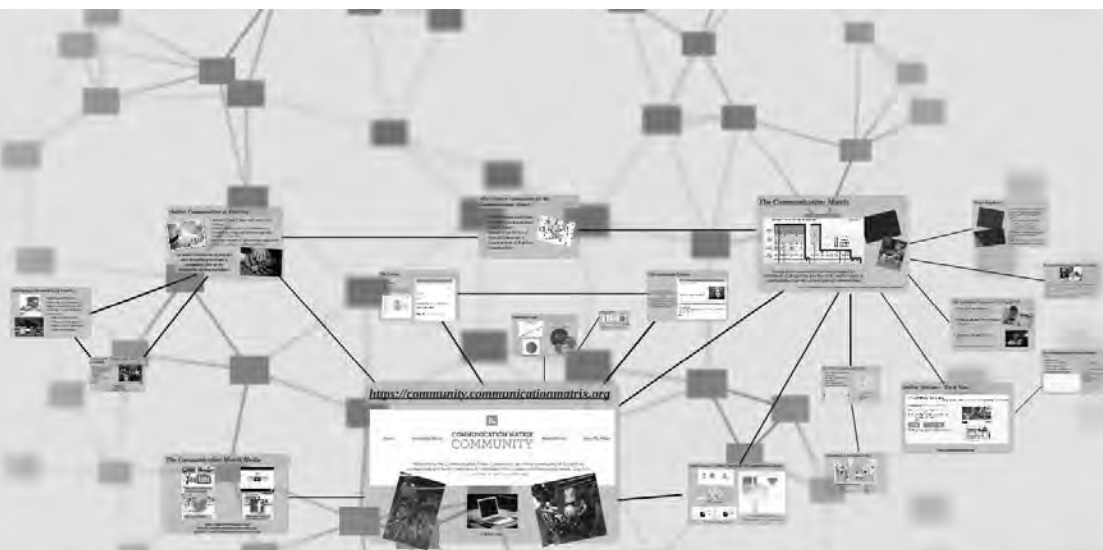
Alexandria Cook graduated from Reed College with a B.A. in psychology and is currently a research assistant in the Design to Learn Lab at Oregon Health & Science University (OHSU). Her research interests include motivational and neurological development of children and adolescents of all developmental capabilities, including risk taking, communication, 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.

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 everyday. She lives with her husband and two children in Monmouth, Oregon and loves to travel.

Presentation Abstract:

The Communication Matrix is an assessment tool for individuals with complex communication needs that is used world-wide to show how individuals at the earliest stages of communication express themselves. Data entered into the website (www.communicationmatrix.org) are mined to provide detailed information about behaviors used to communicate and messages expressed. We will demonstrate the Communication Matrix and the new virtual community of practice, as well as present data on individuals with CHARGE from our database.



Demonstrating the Communication Matrix Online Assessment & Community of Practice: Applications for Children with CHARGE Syndrome, Their Families, and Educators



Alexandria Cook, Oregon Health & Science University
Dr. Charity Rowland, Oregon Health & Science University
Dr. Amy Parker, National Center on Deaf-Blindness







The Communication Matrix

Communication Matrix Profile for Parents and Professionals

Standard View

I Pre-Intentional Behavior	A1 Sustained Attention	A2 Sustained Contact					A3 Experiments Interest in Other People		
	B1 Pretends	B2 Communicates an Action	B3 Obtains More of Something			B4 Attracts Attention			
II Intentional Behavior	C1 Refuses or Rejects something	C2 Requests More of an Action	C3 Requests New Action	C4 Requests More of an Object	C5 Misses Chances	C6 Requests a New Object	C8 Requests Attention	C9 Shows Affection	
III Unconventional Communication									
IV Conventional Communication									
V Perhaps or									

Key:

-  = Surpassed
-  = Not Used
-  = Emerging
-  = Mastered



*An easy to use assessment instrument designed for
individuals of all ages who function at the earliest stages of
communication and who use any form of communication.*

See Dr. Charity Rowland's work: *Communication Matrix: Description, Research Basis and Data* (2012)

Target Population



- The earliest stages of communication (0-24 mo. in typical development).
- Use any type of communicative behavior
 - Augmentative and alternative communication (AAC)
 - Pre-symbolic and more
- Any type or degree of disability, including profound intellectual and multiple disabilities (PIMD).
- All ages.
- Do NOT already use some form of language meaningfully and fluently.

The Need: Weaknesses of Many Assessments



- Accommodate speech only
- Don't address earliest stages of communication in sufficient detail to show progress
- May not probe for meaningful behaviors, as opposed to mere production of behavior
- Do not accommodate sensory or physical impairments
- Emphasize what the child CAN'T do

The Solution: Focus on CAN and DO

- What CAN the child do?
Emphasis on the individual's strengths, not weaknesses.
- What can the child DO with these behaviors? Emphasis on the USES of communicative behaviors.
- What does the child WANT to do? What does s/he want to tell you?
- Parent perspective is crucial.



The Communication Matrix Online

- Free
- Ease of use
- Increase availability
- Encourage parents
- Encourage collaboration
- Generate scientific data

Privacy

- No identifying information collected
- Information entered is stored in database
- IRB-approved privacy mechanisms



Online Version-- Try it Now!


COMMUNICATION MATRIX

[About](#) | [Data Entry](#) | [Results](#) | [FAQ](#) | [Custom Report](#) | [Who's using this?](#) | [New Translations](#)

Communication Assessment for Parents & Professionals

An easy to use assessment instrument designed for individuals of all ages who function at the earliest stages of communication and who use any form of communication.

Try It Now

Create Your Account


[Download the entire handbook.](#)
 Need Adobe Reader?, get it [here](#).



FREE

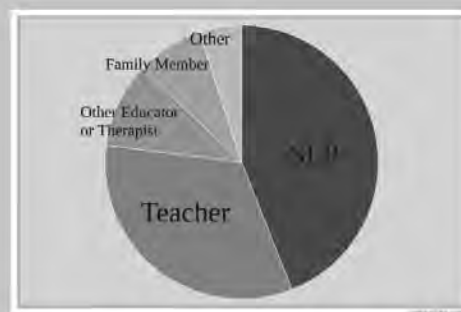


<https://communicationmatrix.org>

Use of The Communication Matrix

- Over 100,000 assessments completed
- On over 49,000 different individuals
- From 144 countries
- Approximately 600/week

Rare Disorder	Estimated Incidence
Aicardi (n = 39)	1: 100,000
Angelman (n = 714)	1: 15,000
★ CHARGE (n = 437)	1: 11,000 ★
Cornelia de Lange (n = 165)	1: 20,000
Rett (n = 516)	1: 16,000
Deafblindness (n = 4,176)	9,387
in U.S. (0-21 yr.)	in U.S. (0-21 yr.)

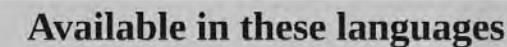


Most Common Disabilities/Etiologies

Autism (Primary Diagnosis)
 Cerebral Palsy
 Developmental Disability/Delay
 Deafblindness (Primary Diagnosis)
 Down Syndrome
 Multiple/Severe Disabilities

-

!



- English
- Korean
- Chinese (traditional)
- Spanish
- Vietnamese
- Czech
- Russian

Why create a Community for the Communication Matrix?

- Demonstrated need from previous Communication Matrix grants
- Interest from Office of Special Education in Communities of Practice
- Sustainability



Online Communities of Practice



- Allows for "Just in Time" and "Just for Me" learning
- Crowd sourcing: sharing of resources and expertise by a large and diverse group with multiple perspectives
- Especially needed for low incidence populations
- Great for isolated or rural communities

An online community of practice uses technology to create a community free of the constraints of time and place.



Developing Communities of Practice



Working with a team including communication experts, practitioners, web developers, and designers we answer:

- Who are the users?
- What are their goals?
- What is their experience with similar tools?

Developing The Communication Matrix Community

Participants will use the Communication Matrix to assess one or more children and beta test the online Community. Participants include:

- Teachers and Speech-Language Pathologists
- Family Members
- Students: 0-21 years old who use less than 3 words meaningfully together

We are recruiting more sites in the coming years



-Clackamas ESD, OR
-High Desert, OR

-Corpus Christi, TX
-Kirk School, IL

-Wing Lake
Developmental
Center, MI

Potential outcomes include:

- better IEP goals
- high parent goal satisfaction
- high student goal attainment
- professional satisfaction with online community

<https://community.communicationmatrix.org>



Forum

Community Library

COMMUNICATION MATRIX COMMUNITY

Shared Science

About The Project

Welcome to the Communication Matrix Community: an online community of support for professionals and family members of individuals with complex communication needs. Log in or join now to start contributing!

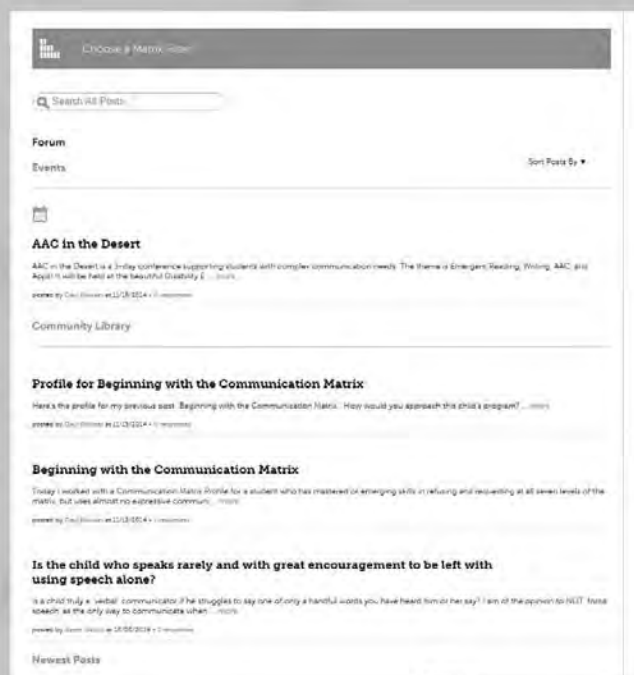
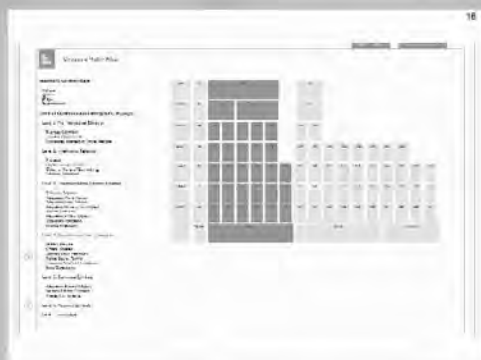


Explore now!



The Forum

- Events
- Newest Posts
- Following
- Share videos, photos, and more
- Communication Matrix Filter



The Community Library

Invited guest Moderators promote the most interesting posts
Moderators include parents and experts in the field of Speech-Language Pathology, Special Education, and more!

Home > Moderators > Amy Parker

Amy Parker, National Center on Deaf-Blindness
<https://www.natcenter.org>

I have 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. I have a 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.

The Communication Matrix is one of the best assessments I have ever used to empower the teams and families of those that serve students with significant disabilities.

Chris Klein TED Talks
 I am always finding myself watching TED talks for inspiration and to learn about new topics. Chris Klein uses augmentative and alternative communication to elaborate on discrimination and ignorance. [more](#)

posted by Amy Parker on 6/18/2014 • 2 responses

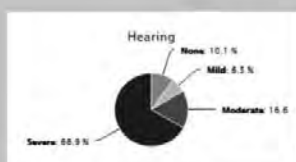
Polite Social Forms
 This little girl signs "hamburger please" to make a polite request. [more](#)

posted by Cherry Goodland on 6/9/2014 • 2 responses

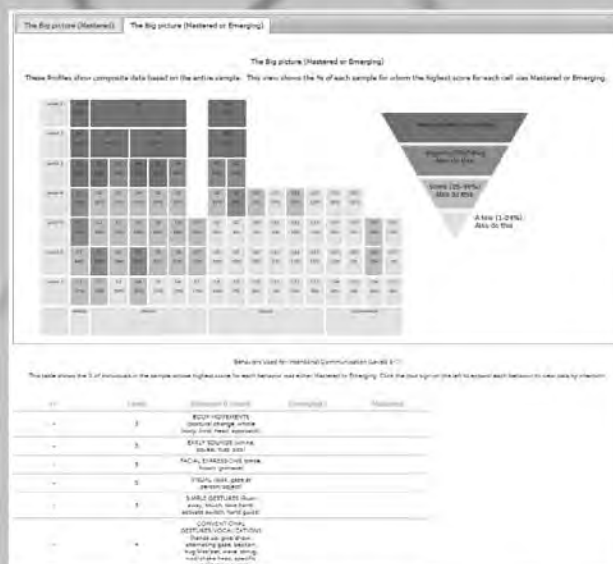
Shared Science: CHARGE Data from the Communication Matrix



• Demographics



• Functional Impairments



• Average Matrix Profile
 • Behaviors used for Communication

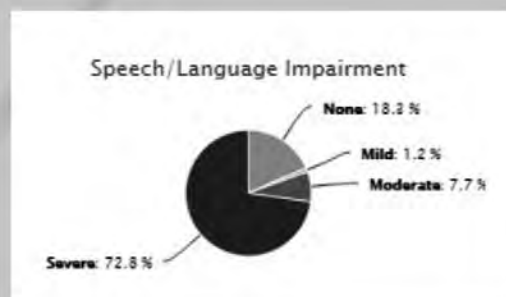
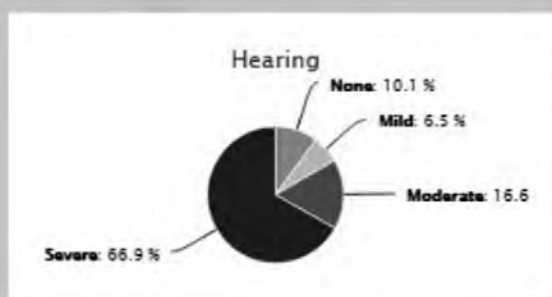
Shared Science: CHARGE Data from the



• Demographics



• Demographics



• Functional Impairments

The Big picture (Mastered or Emerging)

These Profiles show composite data based on the entire sample. This view shows the % of each sample for whom the highest score for each cell was

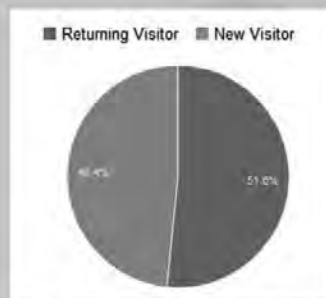
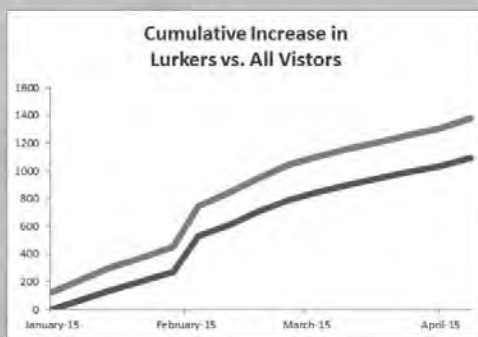


Behaviors Used for Intentional Communication (Levels 3-7)

This table shows the % of individuals in the sample whose highest score for each behavior was either Mastered or Emerging. Click the plus sign on the left to expand each behavior

Table showing the percentage of individuals in the sample whose highest score for each behavior was either Mastered or Emerging. The table is partially obscured by a large plus sign on the left, indicating that the data is expandable.

Preliminary Data



January 12- April 27, 2015

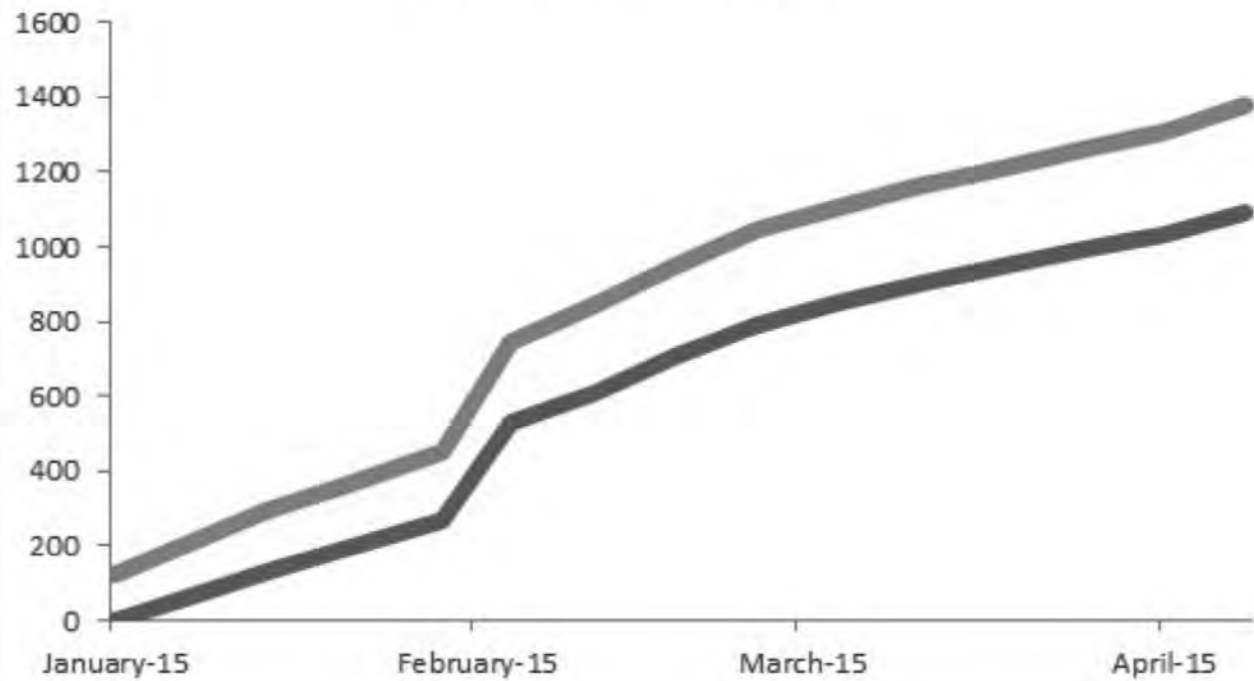
Average Session Duration (all visitors):
4.5 min

~Over 300 members~

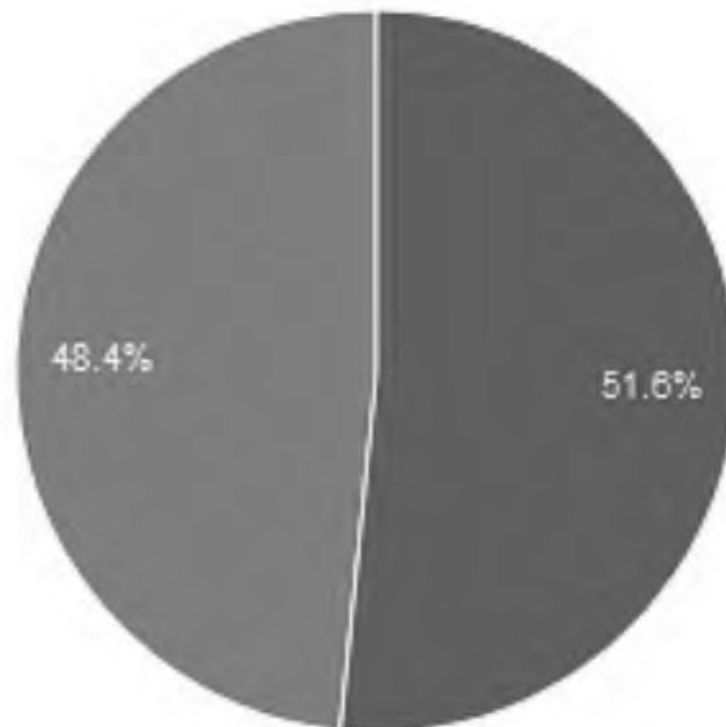


As of April 27, 2015

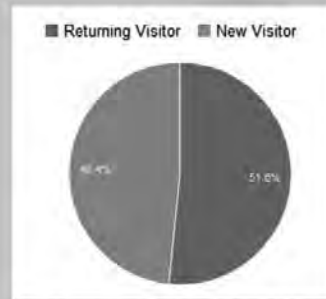
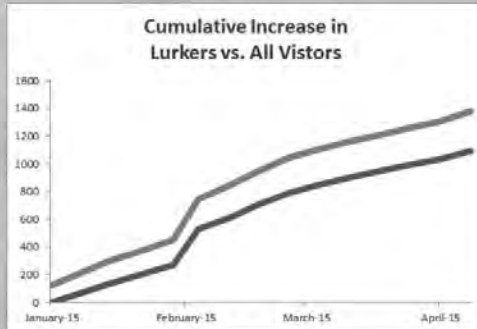
Cumulative Increase in Lurkers vs. All Vistors



■ Returning Visitor ■ New Visitor



Preliminary Data



January 12- April 27, 2015

Average Session Duration (all visitors):
4.5 min

~Over 300 members~



As of April 27, 2015

The Communication Matrix Media



<https://communicationmatrix.org>
[https://community.communicationmatrix.org/
questions? email info@communicationmatrix.org](https://community.communicationmatrix.org/questions?email=info@communicationmatrix.org)



**Saturday, August 1, 2015
Breakout Session #33: 1:00 - 2:00pm
Schaumburg West**

**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.

Keeping it Simple: Strategies for Managing Behavior

Kasee Stratton, Ph.D., NCSP

Daniel Gadke, Ph.D., NCSP, BCBA

Assistant Professors, School Psychology

Licensed Psychologists

Mississippi State University



Prepared for the 12th International CHARGE Syndrome Conference

**What behaviors are you
dealing with?**



Advanced Agenda

- Introduction to behavioral principles
 - Better understanding of the functions of behavior
- ABA Basics
- Building Your Tool Kit
- Case Examples
- Q & A



How We Deal With It

- **All behavior is communication**
- Ongoing Assessment and Intervention
- Techniques Rooted in Applied Behavioral Analysis
- Understanding CHARGE--Behavioral Phenotype



ABA 101...It's not a thing.

- A theoretical orientation that evolved out of traditional behaviorism
- A **lens** that allows us to view the relationship between behaviors and the environment
- Behavior is learned
- Behavior occurs for a reason



Key Terms

- Antecedents – precursors, things that happen before the behavior
- Behavior – specifically what the behavior looks like
- Consequence – what happens immediately after the behavior
- Reinforcement – **Addition or removal of a stimuli to increase behavior**
- Punishment – Addition or Removal of a stimuli to decrease behavior



Functions

- Gain access to or obtain...
- Escape or avoid...
- Commonly Overlooked à Pain
- Ideally, we seek functional equivalency through replacement behaviors
- Is possible for multiple functions... or a universal function...



Building Your Tool Kit

Prevention & Intervention



Tools

- Praise
- Directions
- Ignoring
- Schedules
- Timers
- Signs/PECS
- Visual Stimuli
- Token Charts
- Rewards





CATCH THEM BEING GOOD!

Caption

Praise

- Give praise the child enjoys
- Praise incompatible behaviors
- Remember small improvements!



Giving Directions

DO

- Be specific
- Keep it short
- Avoid “May, Please, Can we, Let’s...”
- Follow with preferred activities



DON'T

- Be vague
- “Be careful”
- “Calm down”
- “Would you put the toys away?”



Directions & Compliance

1. Present Command

- Wait 5-20 seconds for action
- If child complies, **PRAISE**

2. Model Command & Restate

- If child complies with 5-20 seconds, **PRAISE**

3. Guide

- Physically guide completion
- **No talking**



Ignoring

Not looking, not scolding, not noticing at all!

- Do not ignore harmful behaviors
- Grab your favorite distracter
- Prepare for the BURST






First This



-----'s Earns New



!!!

If ----- is able to follow the rules for 5 days in a week, he earns a  piece. If he earns 4  pieces, he earns a new  at the store!

Monday	Tuesday	Wednesday
<input type="text"/>	<input type="text"/>	<input type="text"/>
Thursday	Friday	Saturday
<input type="text"/>	<input type="text"/>	<input type="text"/>
Sunday	<input type="text"/>	



Choiceworkstm

WII NOT AVAILABLE



-----'s Fun!

If ----- follows the rules, he is able to do fun things. If ----- breaks his rules he gets an "X". Every time ----- gets an "X", he loses something fun for the day.

=



=



=



Behavior Case Examples





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**Saturday, August 1, 2015
Breakout Session #34: 1:00 - 2:00pm
Schaumburg East**

**Why Does My Child Do That?
Explanation of and Strategies for
Dealing with Compulsive Behaviors
and OCD in CHARGE Syndrome**

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

Presenter Information:

Nancy Salem-Hartshorne, Ph.D. is Assistant Professor of Psychology at Delta College, and a School Psychologist. She has authored articles and book chapters about developmental outcomes for individuals with CHARGE syndrome. Her son, Jacob, 25, has CHARGE syndrome, and lives in his own home. Nancy is an advocate for individuals with disabilities, teamwork, thorough planning, and forward thinking for quality life outcomes for all individuals.

Presentation Abstract:

All of us have repetitive or compulsive behaviors. All of us have hobbies. But when we see these obsessions and compulsions in our children with CHARGE, they can look odd or disturbing, or may prevent them from accomplishing all that they can, and we worry. Anxiety is the key! Dr. Hartshorne will talk about strategies to address anxiety and OCD-like behaviors in children with CHARGE syndrome.



Why Does My Child Do That?

Compulsive Behaviors and OCD in CHARGE Syndrome

- **DSM-5 Diagnostic Criteria for Obsessive-Compulsive Disorder (300.3)**

Obsessions

Recurrent and persistent thoughts, urges, or impulses that are experienced, at some time during the disturbance, as intrusive and unwanted, and that in most individuals cause marked anxiety or distress. (Typically irrational fears.)

The individual attempts to ignore or suppress such thoughts, urges, or images, or to neutralize them with some other thought or action (i.e., by performing a compulsion).

Compulsions

Repetitive behaviors (e.g., hand washing, ordering, checking) or mental acts (e.g., praying, counting, repeating words silently) that the individual feels driven to perform in response to an obsession or according to rules that must be applied rigidly.

The behaviors or mental acts are aimed at preventing or reducing anxiety or distress, or preventing some dreaded event or situation; however, these behaviors or mental acts are not connected in a realistic way with what they are designed to neutralize or prevent, or are clearly excessive.

Obsessions: Disturbing, Intrusive Thoughts or Fears (Irrational)

Typical Obsessive Fears/Thoughts

Harm to property: burglars, fire, electrical fire, losing something important

Contamination: deadly disease, germs crawling into skin

Harm to a loved one or fear you will harm someone

Magical thoughts

A number or day is good or bad

Step on a crack...

Hearing or saying bad words will produce bad results

Making a religious mistake/sinching will result in going to hell

Fear of not having everything “just right”

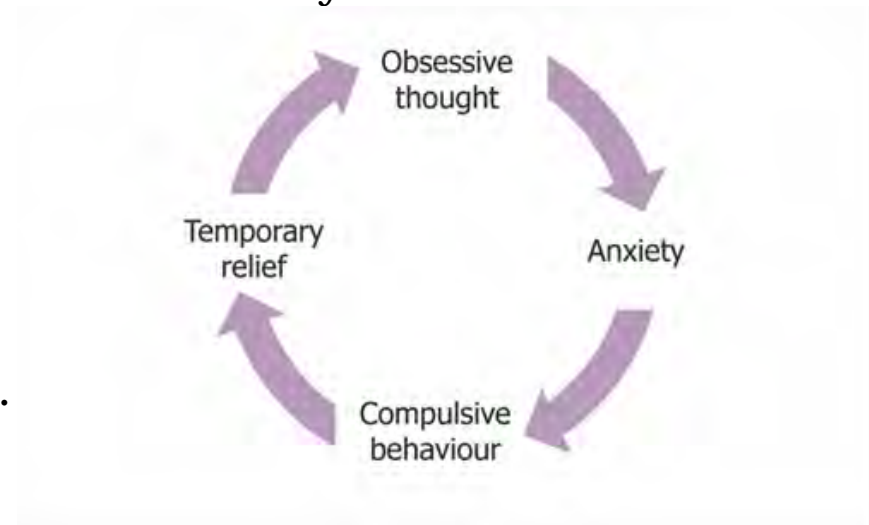
Compulsions: Behaviors/Actions to alleviate worry over the irrational thoughts

Obsession	Typical Compulsions
Harm to property	Check locks, oven, hoard objects
Contamination	Wash hands
Harm to loved one	Text or call repeatedly to check
Magical thoughts	Avoid cracks
Numbers	Avoid using or ending on that number
Bad words	Fix the bad word by saying something else
Religious mistake	Repetitive and excessive prayer
Everything just right	Neatness, orderly, line up, make symmetrical

OCD is an ANXIETY disorder

- You feel stress or anxiety
- You perform compulsive acts to relieve it
- You can try to stop your behavior, but it will only increase anxiety

- A pattern that has developed through reinforcement.
- Relief after compulsion felt as a temporary reward.
- You want that reward again, so you'll do the compulsive act again.
- If it's in the way of your life, it's considered a disorder.



Example: Nancy at age 16

- Obsession over sin/fear of hell: Excessive, repetitive prayer
- Fear of not waking up: Staying up all night
- Rigidly applying rules: counting: 7, 15, 17, 25, 37; Saying/touching things 7 times; don't step on cracks; unwind when you turn around
- Fear of darkness/night: Sit outside and watch sunset until gone, panicking
- Fear of sin/hell: repeating religious swear words heard under breath with alternatives (gosh, heck, darn)
- This was utterly debilitating. It took ALL of my time to attend to these things.

I suggest to you.....

- Most individuals with CHARGE syndrome don't have *true* OCD.
 - Someone with true OCD has irrational thoughts leading to irrational anxiety.
 - The treatment involves exposure to the irrational thought, and prevention of the compulsive response.
-
- Individuals with CHARGE have TRUE circumstances that lead to UNDERSTANDABLE anxiety!

OCD-like behaviors seen in CHARGE

- **My son Jacob uses “Everything must be just right.”**
 - Ordering/Lining up/Making symmetrical
 - Placing/moving to correct place
 - Light switches
 - Doors closed/open
 - Handing cup to caregivers when he sees it
- I’ve seen many individuals with CHARGE use this method.
- **Other things reported or seen:**
 - Repetitive question-asking: “What color is your car?” “What color is your house?”
 - Stuffing things into slots—especially into places from which they are difficult to retrieve.
 - Repetitive, idiosyncratic behaviors: Hand movements, tics
 - Rigid inability to switch activities
 - All-consuming focus on one idea, activity, or item, to the point that it’s not just a hobby/intense interest—Keeping in mind that we ALL have interests/hobbies!
 - What else?

Prevalence of OCD-Like Behaviors in CHARGE

Salem-Hartshorne, N., Blake, K., (in process): 53
Participants. 33 male. Ages 13+

- 49.1% OCD-Like Behaviors
- 45.3% Anxiety

“A lot of what looks like OCD in CHARGE is really just a reaction to having multi-sensory impairments” – D. Brown 2015

These are actually very creative responses to abnormal, anxiety-provoking circumstances.

Caveat: Someone with CHARGE syndrome could possibly also develop typical OCD, but this is not the norm.



What could
our kids
possibly have
to be anxious
about?

The following things may produce real and understandable anxiety for individuals with CHARGE syndrome. These triggers may cause the fear and anxiety, which then produce compulsive behavior.

1. PAIN

It doesn't often
come with a big
sign



2. Sensory overload or underload issues

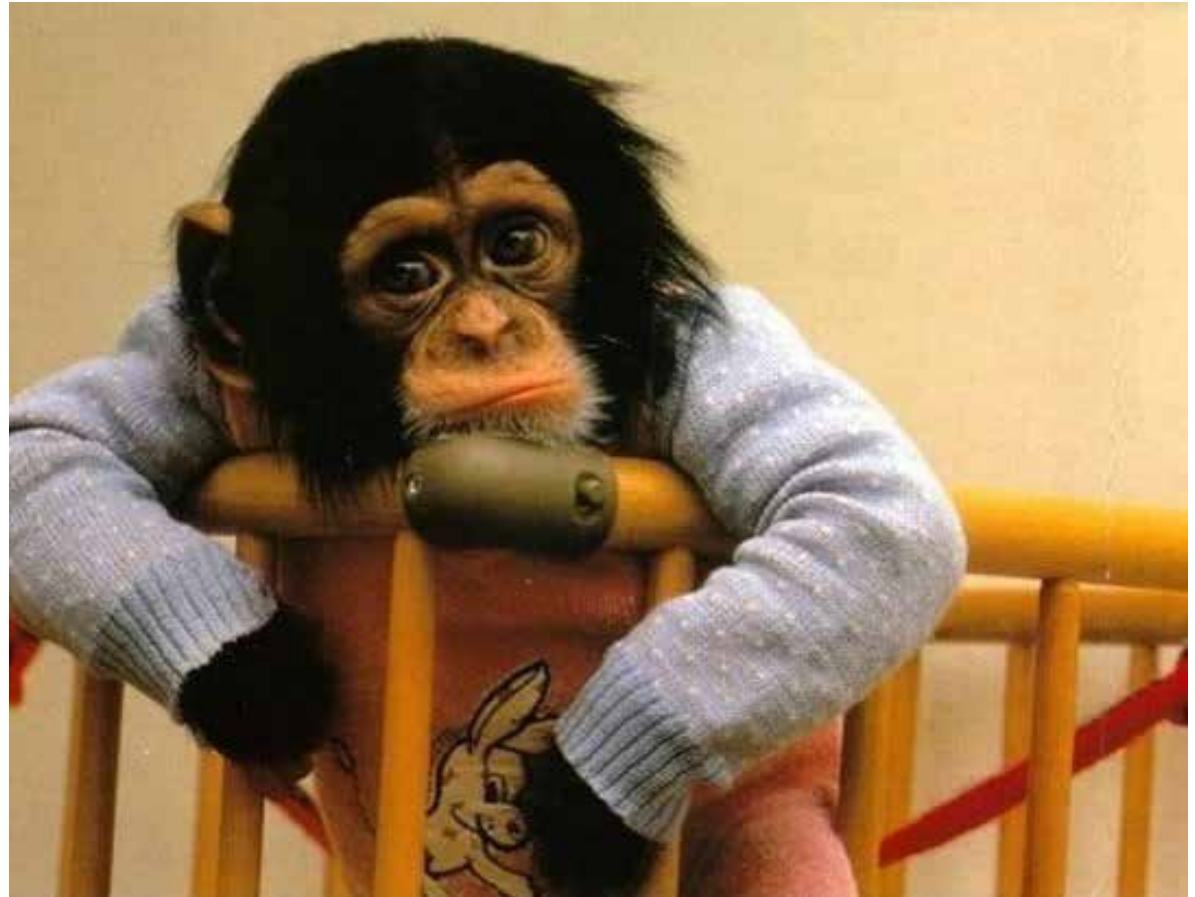


How about just plain boredom?

- When you're waiting/bored, do you:
 - Tap a pencil
 - Wag your legs when sitting
 - Play with your hair
 - Bite your nails
 - (Now that we have smartphones, we do other things as well).

These are all ways of keeping ourselves occupied, or of keeping ourselves aroused and alert.

What do your kids do that look like these, but may not seem as “normal”?



How about checking?

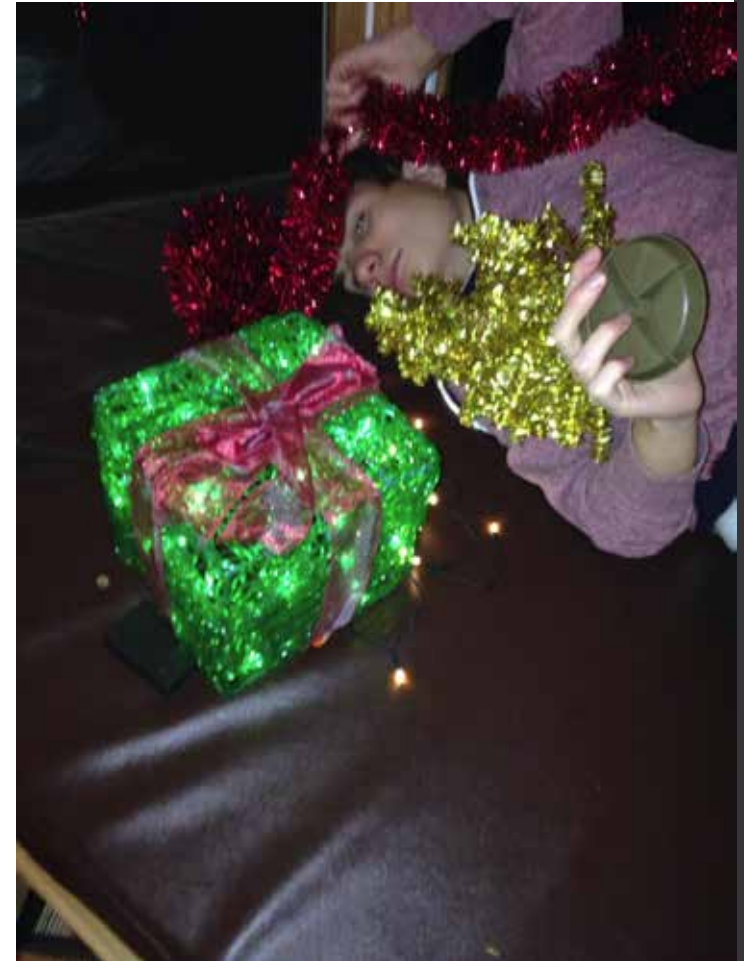
“Checking” is an OCD compulsion. For example, checking to see if the oven has been turned off....seven times....

David Brown's Thoughts:

If you don't have all of the sensory information you need to be reassured, if you never get complete information about your surroundings, if your environment is chaotic and constantly changing, these behaviors make a sort of sense:

- ∅ Tapping things with your hand
- ∅ Running your hand along the edge of a table
- ∅ Arranging items in regular rows or stacks
- ∅ Confirming where things are
- ∅ Constantly checking if things have changed (has the chair moved, etc?)

If small things can change, then it's scary to think that bigger things in the world can change as well. If I can control the small things, it helps with the anxiety over the bigger things.



Sleep

- Sleep and anxiety are related in the general population
 - Fuller, Waters, Binks, & Anderson (1997) found a strong association between high anxiety/worry and clinically significant sleep disturbance
- Salem-Hartshorne & Blake (in process) found almost 60% had sleep problems in adolescent/adult CHARGE sample.
- If there's any way to increase sleep quality, anxiety may decrease.
- Also, reducing anxiety by increasing predictability, helping with sensory issues, and alleviating pain may help sleep quality.

3. Stress

- Life is chaotic.
- I cannot hear/see/balance/predict what's going to happen next.
- I want to escape the chaos.



Steps to Help

To do

or

not to do?

Step One:
Figure out the purpose of the behavior

- What is its goal? What function does it serve for the individual?

What looks like an inappropriate goal may be masking something else.

Repetitive questions: “What color is your car, what color is your house?”

- Ø Need for social interaction but not having social skills to initiate true conversations—they know they are supposed to do something. This is what they know how to do.
- Ø Need to do something to organize a chaotic situation with many people around—they are overwhelmed and this is one way to make the situation understandable.
- Ø Need to know more about people to feel safe around them—they are among strangers.

“I want to be a professor...”



Step Two: How urgent is it?

Urgency of Intervention Questions (D. Brown, 2015)

1. Is this a behavior that just bugs you personally, so that it can be accepted and ignored?
2. Is this a behavior that seems to help the child to function in a positive way, so that it can be accepted and ignored?
3. Is this a behavior that seems to help the child to function in a positive way, but could be reduced, or replaced by another more appropriate behavior?
4. Is this a behavior that is undesirable and really needs to be reduced or replaced over time?
5. Is this a behavior that needs to be prevented immediately?
6. Finally, how much can we improve things by changing our behavior and the environment that the child is in, rather than directly trying to change them?

Step Three: Intervention



Pain

Communication of pain may devolve until you get someone's attention, especially if your communication skills are limited or you have difficulty understanding what's happening to you when you are in pain.

Level 1. Avoiding work, putting my head down, or lying on the floor

Level 2. Crying, whining, complaining, acting out

Level 3. Hitting, biting, pulling hair

- This person is communicating, but we aren't understanding. So they change their communication mode until we notice!
- There is a need to help them find a way to more appropriately identify and communicate pain.
- For some, pain behaviors, although worrisome, can be quite helpful.
 - When Jacob blows raspberries repeatedly, we know it's gas pain or cramping
 - When Jacob digs things into his neck, we know it's his ears bothering him
 - He doesn't have other ways (yet) to tell us these things.

CHARGE Non-Vocal Pain Assessment

(Stratton, 2012) (Excerpt)

- **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

Sensory Issues

- Sensory Integration (deep pressure, weighted garments, etc., to bring arousal level down)
- Sensory breaks (to bring arousal level up)
- Allow to withdraw when overwhelmed
- Set up a better sensory situation for environment

Boredom

Example: Waiting in a doctor's waiting room or at a meeting or restaurant can bring on all kinds of strange-looking behaviors in Jacob.

- Rocking
- Blowing
- Hand-flapping

Why?

He can't see or hear the TV everyone else is watching

He doesn't have a smart phone. (What did we do without those?)

He needs something to stimulate his brain.

**For Jake, sensory toys really help.
What would work for you?**



Checking

These behaviors may be necessary to alleviate the anxiety brought on by not having full availability of sensory information.

Interventions:

- Ø Allow the behavior, as it serves a clear purpose for the individual
- Ø Make the environment as stable and predictable as possible

Sleep

If sleep and anxiety are related, there are two things that might help:

1. **Improve quality of sleep**

1. Sleep Hygiene
2. Pay attention to sensory overload
3. Melatonin has been used by some to help bring on sleepiness

2. **Reduce anxiety**

Stress/Anxiety: The Key is *Predictability*

(adapted from Tim Hartshorne, 2014)



We like to know what is going on.

- What are we doing right now?
- What are we going to do next?
- What did we just do?

Sun	Mon	Tue	Wed	Thur	Fri	Sat
July						
2015						
			1	2	3	4
5	6	7	8	9	10	11
12	13	14	15	16	17	18
19	20	21	22	23	24	25
26	27	28	29	30	31	

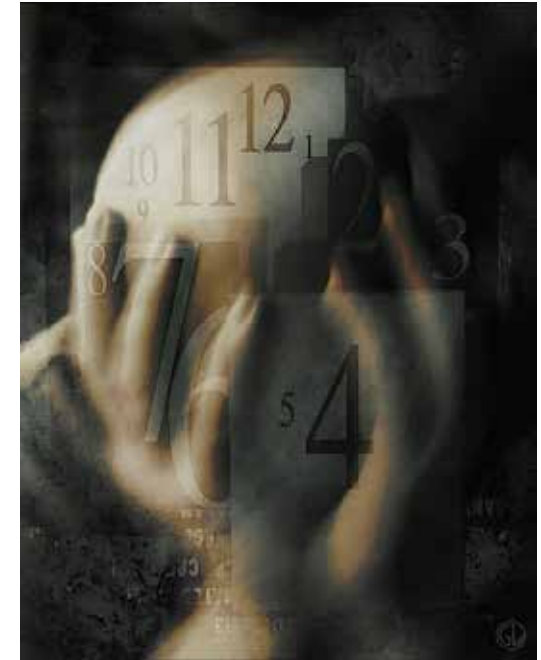
www.whoneedsacalendar.com

Why use a calendar system?

- Security of knowing what comes next
- Being able to anticipate things – looking forward
- Alert to unexpected changes in routine
- Allows for participation in decisions about events
- Allows for conversation about what has happened
- Provides for a mutually understood topic for dialogues
- Clearly represents the passage of time
- Reduces anxiety about what has, is, and will happen

Imagine waking up and...

- No idea what time it is
- No idea of what will happen today
- No idea how soon something might happen
- No idea of the expectations for what will happen
- Problems detecting the true passage of time
- Will I like it?
- Jacob in the car
- This can cause lots of anxiety!

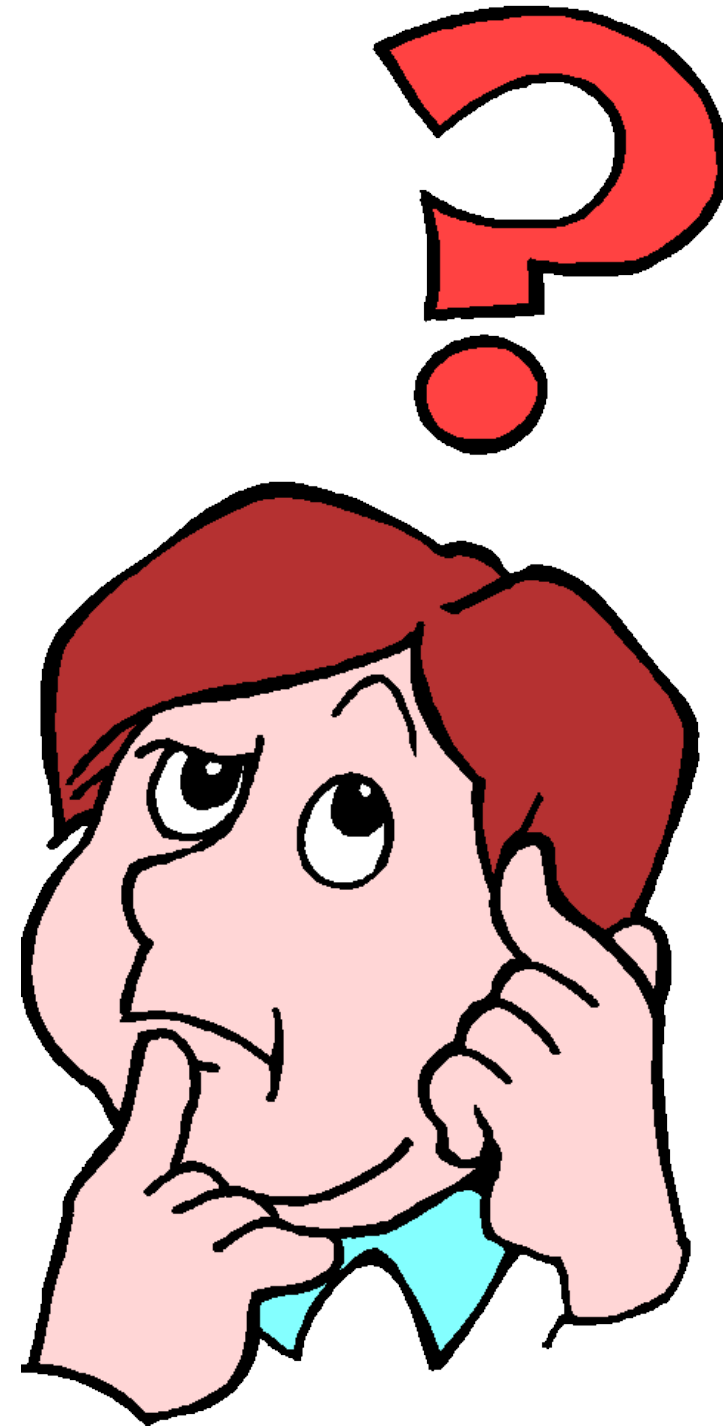


Communication

- Calendars provide an opportunity for conversation around the schedule.
 - What we are doing now
 - What we are doing later
 - What we just did
- Calendar systems also teach what time is!
- Once the individual understands the concept of the calendar, they can make choices within it, and it can be used for all kinds of routines.
- Technology for these types of things is exploding (If-Then Visual Schedule on iPad)



Questions,
Comments,
Concerns,
Bellyaches?





Saturday, August 1, 2015
Breakout Session #35: 1:00 - 2:00pm
Schaumburg E-G

**An interactive seminar in genetics: All
you want to know about the genetics of
CHARGE syndrome**

**Conny van Ravenswaaij-Arts, MD, PhD &
Nicole Corsten-Janssen, MD**
University Medical Center Groningen

Presenter Information:

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

Presentation Abstract:

After a short introduction, all questions of the participants regarding genetics, the CHD7 gene, mutations, recurrence risk, et cetera, will be answered using instructive illustrations. A handout that explains the main issues for a lay audience will be provided.

Based on the experience of previous years, we will offer parents the opportunity to discuss the specific genetic test results of their child with us in short sessions after the seminar.

All you want to know about CHARGE and genetics

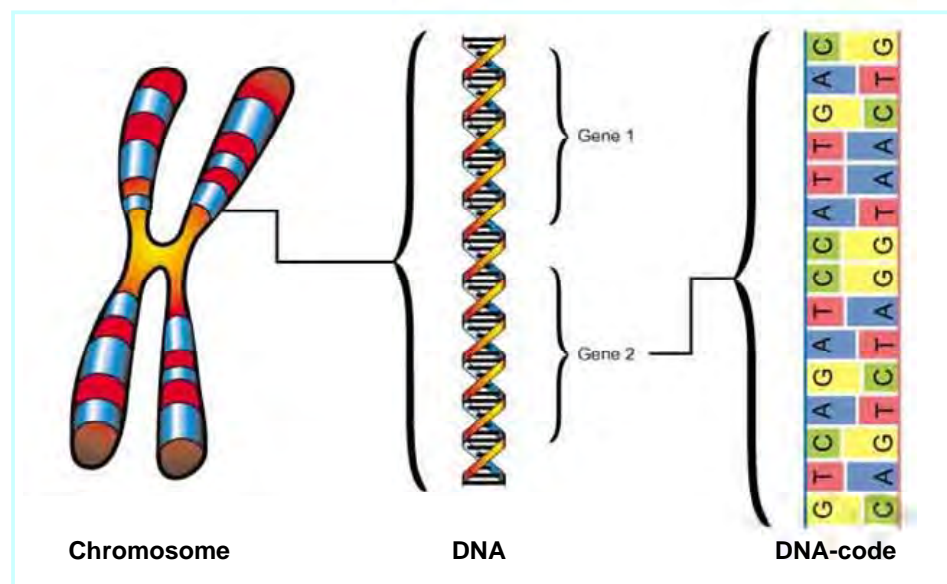
By Nicole Corsten-Janssen and Conny van Ravenswaaij-Arts
Department of Genetics, University Medical Center Groningen, The Netherlands



What are chromosomes, genes, DNA and mutations?

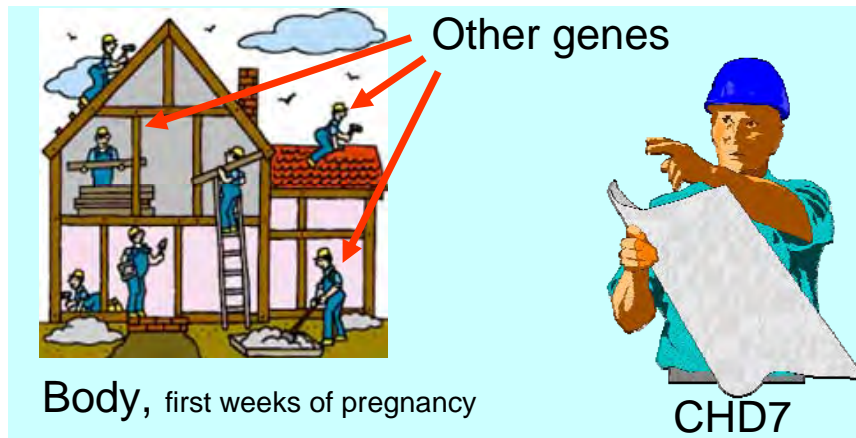
Our genetic information is tightly packed up on structures called chromosomes. Humans have 46 chromosomes grouped into 23 pairs. Everyone inherits 23 chromosomes from his father and 23 chromosomes from his mother. A chromosome consists of tightly packed up DNA.

DNA is our hereditary material and it is made up of four chemical bases (A,C,T,G). Genes are the pieces of DNA that hold the code for proteins, which are important for all kinds of functions in the body. A mutation is another word for a change in the DNA code, such a change in the DNA code can lead to a change in the function of the proteins.



What is the *CHD7* gene and what does it do?

Since 2004 we know that CHARGE syndrome is caused by a change (mutation) in the *CHD7* gene. Every person has two *CHD7* genes, one inherited from their father and the other from their mother. *CHD7* is a regulatory gene. It regulates the work of developmental genes very early on, during the development of the fetus. If there is insufficient *CHD7* protein being made, there is a higher risk of developmental defects occurring in specific organs like the heart, eye, ear, kidney, etc. A change in one of the two *CHD7* genes is enough to result in CHARGE syndrome. However, the syndrome is highly variable and it is not possible to predict the clinical consequences for the child from a specific change in *CHD7*.



CHD7 regulates the function of genes influencing fetal development during early pregnancy.

How is CHD7 analysis done?

CHD7 analysis is performed on DNA. DNA is usually extracted from blood cells, but other tissues, e.g. skin or saliva, can also be used. Different types of mutations can be present in the *CHD7* gene. Most of these mutations will be detected by routine DNA analysis (called “sequencing”). Sometimes a part of the *CHD7* gene may be missing or even the whole gene (called a “deletion”). Deletions of *CHD7* are rare and occur in approximately 1% of CHARGE patients. They cannot be found by routine DNA analysis, but can be identified by other techniques (e.g. array, MLPA).

Type of mutations

Normal gene	Tim and his toy
Point mutation	Pim and his toy
Deletion	Tim and toy
Insertion	Tim and his big toy
Frameshift	Tim ndh ist oy

Why perform DNA analysis?

There are several reasons to perform DNA analysis:

- To provide certainty
- Confirm a tentative diagnosis in a child with atypical features (these two reasons have consequences for the clinical follow-up)
- Confirm the diagnosis, so that parents or siblings know their recurrence risk
- Prenatal options

What if no *CHD7* mutation is found in my child?

- A diagnosis of CHARGE syndrome can be made by identifying a *CHD7* mutation, or by looking at the clinical criteria for the syndrome. If your child

fulfills the clinical criteria, he/she has CHARGE syndrome, irrespective of the results of *CHD7* analysis.

- Some other syndromes have clinical features that overlap with CHARGE syndrome and it is important to exclude these syndromes from the diagnosis.
- Current techniques are not good enough to identify all *CHD7* mutations.
- Other genes might also contribute to CHARGE syndrome (see below).

What is the risk that CHARGE syndrome will re-occur if we have another child?

Since familial CHARGE syndrome is extremely rare, the recurrence risk for parents who have a child with CHARGE syndrome is, in general, low. If parents want to learn more about their risk for future pregnancies, we recommend:

- A geneticist should see if the parents have any mild symptoms of CHARGE syndrome (hearing, balance, smell, shape of the ears).
- If a *CHD7* mutation has been found in a child, the parents can also be offered DNA analysis.

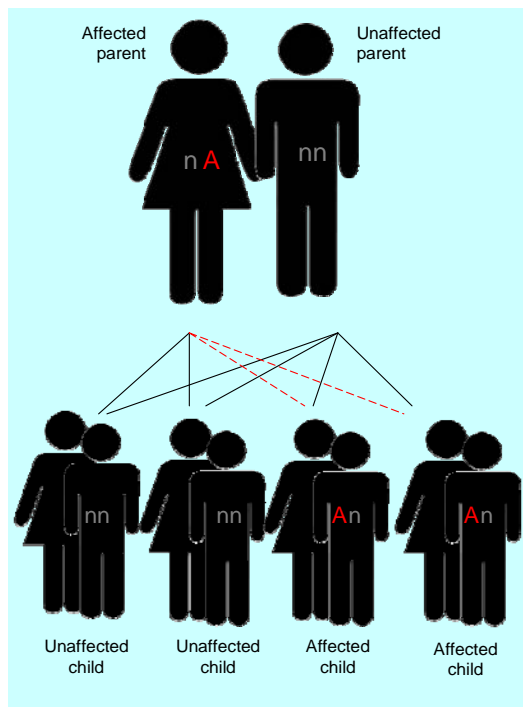
There are three possibilities:

1. Parent does not have CHARGE syndrome or a *CHD7* mutation

If the *CHD7* change is not found in one of the parents (the most common situation), there is still a small risk of carrying a *CHD7* mutation ("germline mosaicism"), so the recurrence risk is not zero, but 1% or 2%.

2. Parent has CHARGE syndrome

If one of the parents has CHARGE syndrome, the recurrence risk varies. Parents pass half of their genetic information on to their children. The parent with CHARGE syndrome can pass on either their normal *CHD7* gene or their gene with a mutation. This means that there is a 50% recurrence risk in another child.

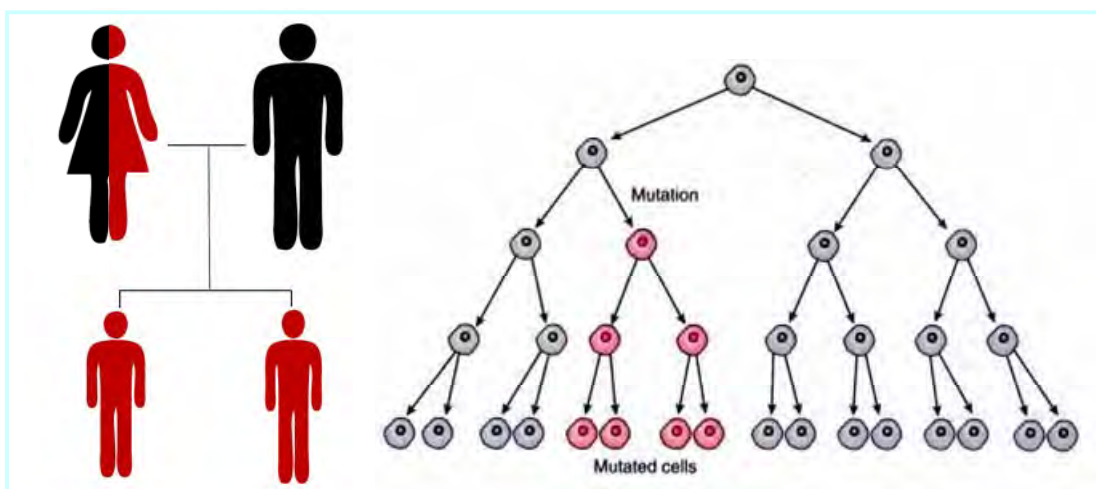


*The parent with mild CHARGE syndrome passes on the normal *CHD7* gene (n) or the abnormal *CHD7* gene (A). The recurrence risk is 50% for each pregnancy.*

3. Parent with a *CHD7* mutation in some of their cells (“mosaicism”)

Very rarely, a family may have two affected children with the same *CHD7* change, while the parents do not have any features of CHARGE syndrome. How can this happen? We may find that one of the parents carries a change in the *CHD7* gene in only some of his/her body cells. This is called mosaicism and it can occur when the fertilized egg has a normal *CHD7* gene, but a spontaneous change occurs later in one cell as the cells divide and grow (see figure). Only the cells coming from that cell with the changed *CHD7* gene will have the mutation. If these cells are also present in the parent's ovaries or testes, egg or sperm cells with the *CHD7* mutation can be formed and lead to the pregnancy of a child with CHARGE syndrome.

If a parent has a mosaic form of the *CHD7* mutation, the recurrence risk for further children can be up to a maximum of 50%.



*In this example of mosaicism in the mother, there is a *CHD7* change (mutation) in some of her body cells. The scheme on the right shows that this can occur when the change (red cells) arises spontaneously during a cell division after fertilization. The mother will not have CHARGE syndrome herself, but she does have an increased risk of having affected children because she can pass on the *CHD7* change via an egg cell. The subsequent child will have the *CHD7* change in all its cells and will therefore have CHARGE syndrome.*

Options in a subsequent pregnancy

Parents who have a child with CHARGE syndrome in whom a *CHD7* mutation has been identified, may choose to have prenatal diagnosis performed for a subsequent pregnancy. But please remember that DNA analysis cannot predict the severity of CHARGE syndrome. A fetal ultrasound can provide extra information, like whether the baby has a heart defect, but ultrasound cannot “see” everything – deafness, developmental delay and behavioral problems cannot be detected.

The choice to have prenatal diagnosis is a personal one, and the geneticist must inform the parents well, so that they can make a choice they feel good about.

Is *CHD7* the only gene causing CHARGE syndrome?

In 5% to 10% of patients with typical CHARGE syndrome, no mutation in the *CHD7* gene can be found. This percentage is higher in patients who do not fulfill the clinical criteria for CHARGE syndrome (atypical presentation). So there may well be other genes that are also responsible for CHARGE or CHARGE-like syndrome. Mutations in a gene called *SEMA3E*, had been identified in two patients. A few other genes, like

CHD8 (a “candidate gene”), have been studied, but no mutations were found in these genes in CHARGE syndrome patients. Recently, we started to search for other genes that might cause CHARGE syndrome using a new technique, called next-generation sequencing or whole exome sequencing. This technique makes it is possible to look for mutations in all the known genes in a single test.

Any questions? Please come and ask us during the CHARGE conference, or send an email to CHARGE@umcg.nl . More information on our research projects on CHARGE syndrome is available at www.rug.nl/research/genetics/research/chargesyndrome/





**Saturday, August 1, 2015
Breakout Session #36: 2:15 - 3:15pm
Schaumburg A-B**

**Which Way is Up? – How behavior
reveals sensory processing
differences in children with CHARGE
Syndrome**

**Kate Beals, OTR/L
Deafblind Program -
Perkins School for the Blind**

Presenter Information:

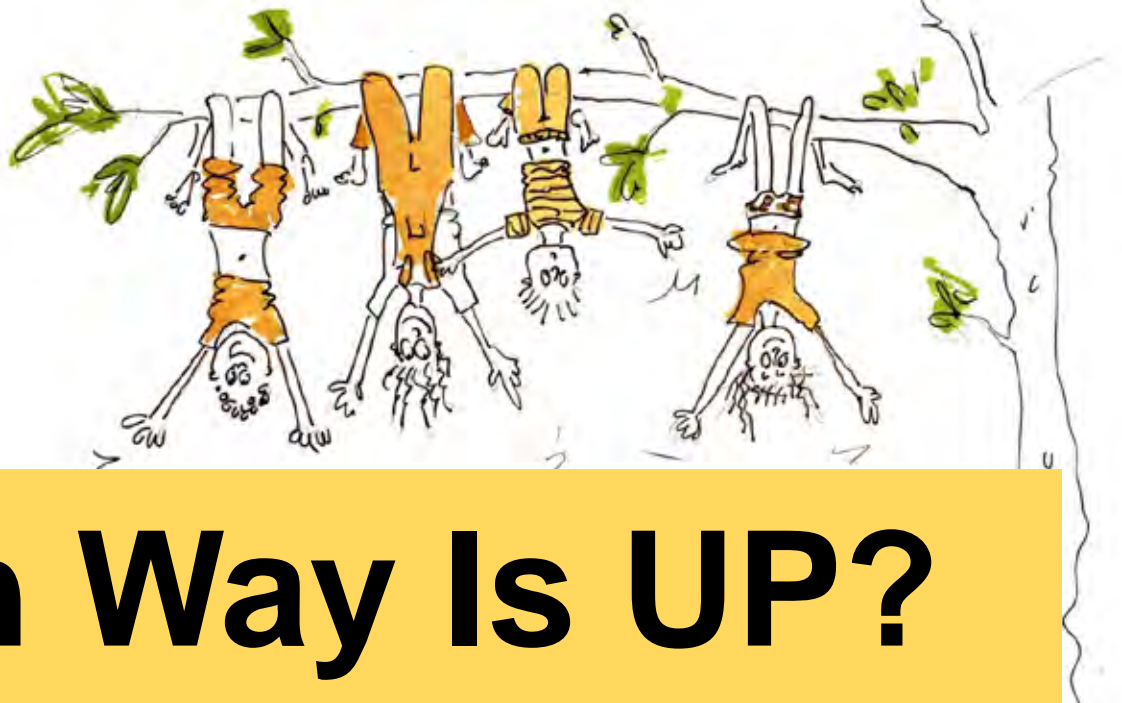
As a pediatric occupational therapist and single parent of a 25-year-old son with Autism, sensory processing/integration is a favorite topic of mine, both personally and professionally. I became interested in CHARGE Syndrome when I worked for the South Carolina Interagency Deaf-Blind Project from 2008 to 2013. At that time I was asked to “look into” sensory processing issues in CHARGE Syndrome – and hit the proverbial jackpot! Wow! What an amazingly complex set of sensory processing and integration issues children who have CHARGE Syndrome have to cope with! Thank goodness David Brown has written so many wonderful articles!

I attended my first CHARGE Conference in Chicago, then presented at the conferences in Florida and Arizona. People tell me I have a way of explaining complicated neurological processes (sensory-motor integration) in a way that “everyday people” can understand pretty easily. Now that I work in the Deafblind Program at Perkins School for the Blind, I provide OT services for many students with CHARGE Syndrome, and every day they teach me more about the unique ways they experience their own bodies and the world around them.

Presentation Abstract:

Children who have CHARGE Syndrome are known to have problems with sensory processing across multiple systems, which affects the way they experience themselves and the world around them. By understanding how the seven sensory systems operate differently in children with CHARGE Syndrome, families, caregivers, therapists, and teachers can learn to recognize behaviors that suggest specific sensory processing issues and needs.

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Which Way Is UP?

How behavior reveals
sensory processing differences
in children with CHARGE Syndrome



Basic Terms & Concepts

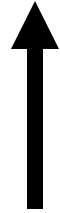
- Behavior = stuff we **DO** = **Motor function**
- Sensory Processing = how we **get** information about what is happening outside of, on, and inside of our bodies, and how we **understand** that information...
- “Sensory-Motor System”



The Brain

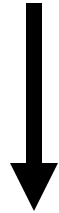


• Sensory in



- Messages (input) from all 7 sensory systems travel to the brain on “inbound” tracts of the nervous system.
- It is a one-way street.
- Sensory information only goes **IN**.

• Motor out



- Messages (output) travel to the muscles on motor pathways, or “outbound” tracts of the nervous system.
- It is a one-way street.
- Motor instructions only go **OUT**.

Seven Sensory Systems

- Vision
- Hearing
- Taste
- Smell
- Light touch



- Proprioception
(deep pressure)
- Vestibular
(head movement)

The Senses in CHARGE Syndrome

Individuals who have CHARGE Syndrome tend to have reduced (or altered) ability to receive input through some or all of their sensory systems.

For the most part, there are **STRUCTURAL DIFFERENCES** that make the accurate reception and processing of sensory information impossible.

Structural Differences

- **Vision** – Colobomas – absence of tissue
- **Hearing** – Malformation of the cochlea or the cochlear nerve (sometimes absent CN VIII)
- **Smell** – Absence or reduced number of olfactory bulbs and stems; nasal blockages
- **Vestibular** – Absence or hypoplasia of semicircular canals

Structural Differences

- **Taste** – Cranial nerve abnormalities; absence of smell reduces flavor
- **Proprioception** – structural difference? Don't know. But movement patterns resemble those of individual who lacks proprioceptive sense (see T. Jessell video)
- **Light Touch** – structural difference? Don't know. *Might* be an exception, but need more research...

The Brain and Experience

- Brain needs “experience” in order to grow, develop, make connections (“wrinkles”)
- Experience is sensory
- CHARGE Syndrome = “Multisensory impaired”
- “Hungry” for sensory input

“Sensory-Seekers”

“Sensory-Seeking” Behaviors

TO GET PROPRIOCEPTION

- **Walking** – feet turned out, extra slaps or taps with feet, exaggerated knee extension, walking on tip-toes (also compensatory))
- **Flapping** – hands, feet, arms, legs, fingers; repetitive and rapidly alternating flexion and extension
- **Posturing fingers** – habitually bending fingers into unusual positions, flicking fingers, moving fingers near eyes

“Sensory-Seeking” Behaviors

TO GET PROPRIOCEPTION

- **Hitting self** – Using fist, knuckles, or wrist to hit self on the face, head, chest, or stomach
- **Toe-walking** – Walking on tip-toes
- **Mouthing** – putting hands, fingers, or objects that are not food into the mouth
- **Lying on Back** – on floor, on bed, in beanbag, on sofa (also compensatory)
- **Chin propping** – elbow on table, chin propped in hand (also compensatory)

“Sensory-Seeking” Behaviors

TO GET VESTIBULAR + VISUAL

- **Spinning self** – turning own body in a circle, spinning on a swing or sit & spin toy
- **Spinning objects** – twirling things near the eyes
- **Upside Down** – hanging head back, over the edge of sofa or bed

Right side up,
I see the floor.

Upside down,
I see the ceiling.

“Look at the camera.”



Photo courtesy of California Deaf-Blind Services

Compensatory Strategies

- **Balance** comes from the interaction of the visual, vestibular, and proprioceptive systems, none of which functions optimally in an individual who has CHARGE Syndrome.
- **That makes staying upright against gravity very difficult, and very tiring.**
- Behaviors like chin propping, walking with feet turned out, and lying down on the back help compensate for decreased balance, and for the excess energy that has to be used in order to stay upright against gravity

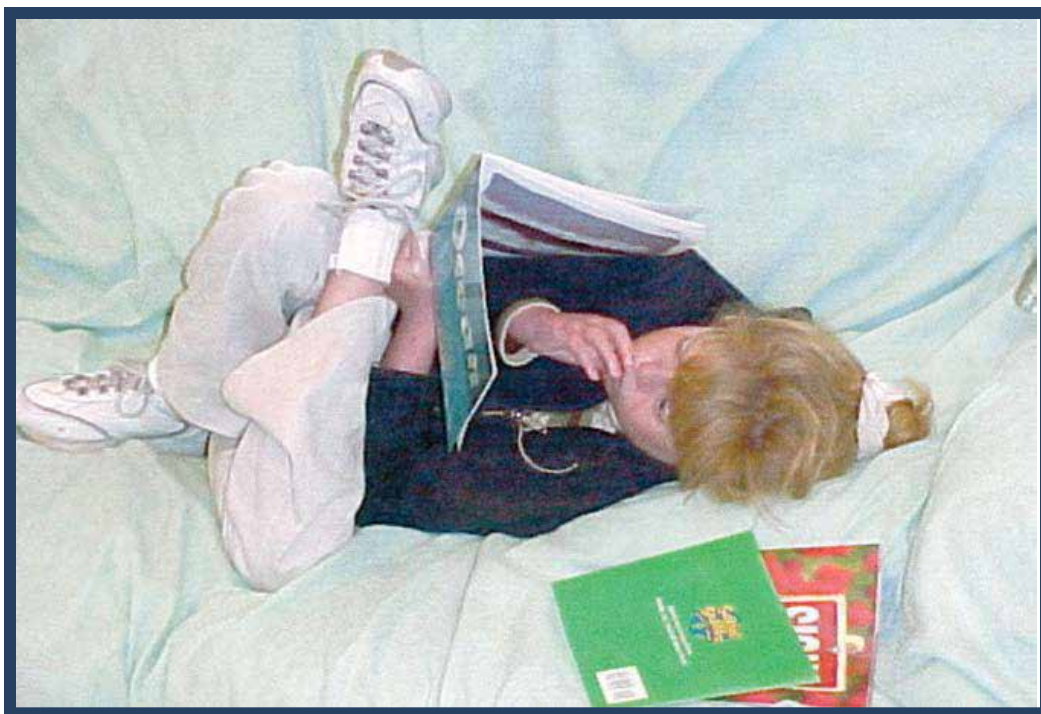


Photo and quote below
are from
*The forgotten sense:
Proprioception,*
By David Brown in
Deafblind
International Review,
July – December, 2006

"In order to use her residual vision to look at fine details in a book Amy needs to be horizontal with her entire body and head fully supported: having one ankle up on the other knee sends her brain a strong message, through the proprioceptive sense, that her lower body is fixed and stable and not moving."

- *David Brown*

What about – you know – *THAT*?

Let's talk about fecal smearing, okay?

- We hate it. They love it.
- What's the difference?

Sense of Smell

Is fecal smearing
sensory-seeking or attention-seeking?

Videos

Let's watch some videos and see if we can figure out what the child's behavior tells us about his or her sensory processing differences.

<http://media.hhmi.org/hl/08Lect3.html>



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Which Way is UP?

How behavior reveals
sensory processing differences
in children with CHARGE Syndrome

Kate Beals, OTR/L

Deafblind Program

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12th International
CHARGE Syndrome
Conference Chicago 2015





**Saturday, August 1, 2015
Breakout Session #37: 2:15 - 3:15pm
Schaumburg C-D**

Language Acquisition through Motor Planning (LAMP): AAC for Individuals with CHARGE Syndrome

**John Halloran, M.S.
The Center for AAC & Autism**

Presenter Information:

John Halloran, a speech-language pathologist, is the Senior Clinical Associate for The Center for AAC and Autism. John has worked in the field of AAC since 1994. He has a special interest in children who are challenged by severe physical or cognitive disabilities.

John Halloran received his masters in Communicative Disorders from the University of Arkansas for Medical Sciences in 1992. After graduation, he worked at Arkansas Easter Seals Rehabilitation Center, specializing in assistive technology. He has also owned a pediatric therapy clinic and after-school care for children with disabilities. He has taught augmentative communication at the University of Arkansas for Medical Sciences.

He was the primary developer of Language Acquisition through Motor Planning and presents nationally. He is a past presenter at the International CHARGE Syndrome Conference and has supported families of nonverbal individuals in addressing their unique communication and language needs.

Presentation Abstract:

Language Acquisition through Motor Planning (LAMP) is a therapeutic approach using motor learning principles and a voice output communication aid to give non-verbal individuals with CHARGE syndrome and other developmental disabilities a method to develop independent and spontaneous communication. Strategies and tools to teach language/communication skills within this framework will be discussed.



**Saturday, August 1, 2015
Breakout Session #38: 2:15 - 3:15pm
Schaumburg West**

**Family Leadership and Empowerment:
The Lived Experience of Parents of
Children with Charge Syndrome in
Advocating and Navigating Systems**

**Seth Harkins, EdD
Philip J. Rock Center & School
Panel: Janay Mohamed (Parent), Maria & Dan
Roeters (Parents), Scott Boroff (Parent),
Karen Windy (Deaf-blind specialist)**

Presenter Information:

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

Presentation Abstract:

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

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

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

Abstract

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

Presentation Outline

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

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



**Saturday, August 1, 2015
Breakout Session #39: 2:15 - 3:15pm
Schaumburg East**

Anticipation and Behavior

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

Presenter Information:

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

Presentation Abstract:

Anxiety about what is occurring or going to happen is one source of challenging behavior for children with CHARGE. This presentation reviews calendar systems and activity routines as ways to increase the predictability of life for those who have CHARGE, and to reduce the experience of anxiety and the incidence of challenging behavior.

Anticipation and Behavior

Tim Hartshorne, Central Michigan
University

Anxiety

- “A feeling of worry, nervousness, or unease, typically about an imminent event or something with an uncertain outcome.”
- What does anxiety look like in children?

- *Pessimism and negative thinking patterns**
- *Anger, aggression, restlessness, irritability, tantrums, defiance**
- *Constant worry about things that might happen or have happened**
- *Crying**
- *Physical complaints such as stomachaches, headaches, fatigue**
- *Avoidance behaviors**
- *Sleeping difficulties**
- *Perfectionism**
- *Excessive clinginess and separation anxiety**
- *Procrastination**
- *Poor memory and concentration**
- *Withdrawal from activities and family interactions**
- *Eating disturbances**

One difficulty is that the same behavior can reflect different experiences

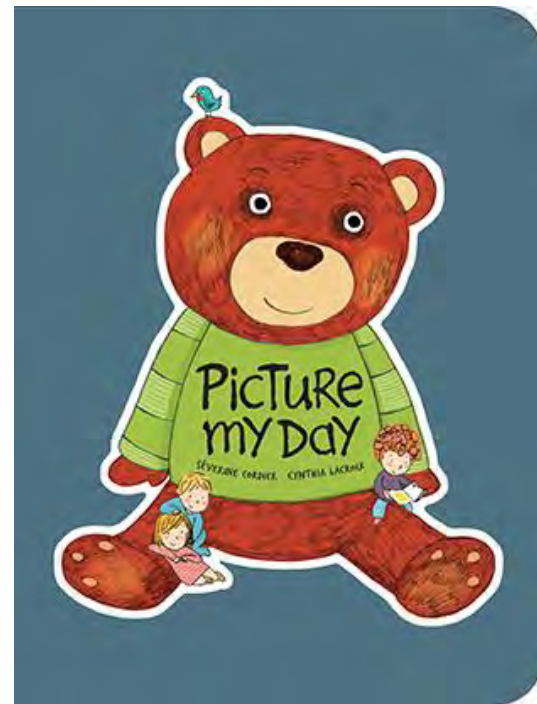
- Sleep problems
 - Pain
 - Circadian rhythm and melatonin problems
 - Self-regulation issues
 - Anxiety
- Tantrums
 - General complaints
 - Frustration and Anger
 - Pain
 - Anxiety

Things to be anxious about

- **Novelty**
- **Predictability**
- **Event uncertainty**
- **Imminence**
- **Duration**
- **Temporal uncertainty**
- **Ambiguity**

We like to know what is going on.

- What are we doing right now?
- What are we going to do next?
- What did we just do?



When we are not sure...

- We become anxious
- We engage in behavior which expresses our feelings
- We engage in behavior or activities that help us feel more secure

Anxiety is related to uncertainty about what is going to happen next

- What will happen today?
- What do they want me to do?
- Where are we going?
- What will happen when we get there?
- Will I feel safe?
- The world does not always make sense
- It can be unpredictable

One way to increase predictability

July 2015 Calendarpedia
Your source for calendars

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
28	29	30	1	2	3	4
5	6	7	8	9	10	11
12	13	14	15	16	17	18
19	20	21	22	23	24	25
26	27	28	29	30	31	1

© www.calendarpedia.com ML: Independence Day observed, 4th Independence Day www.calendarpedia.com

August 2015 Calendarpedia
Your source for calendars

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
26	27	28	29	30	31	1
2	3	4	5	6	7	8
9	10	11	12	13	14	15
16	17	18	19	20	21	22
23	24	25	26	27	28	29
30	31	1	2	3	4	5

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Why use a calendar system?

- Security of knowing what comes next
- Being able to anticipate things – looking forward
- Alert to unexpected changes in routine
- Allows for participation in decisions about events
- Allows for conversation about what has happened
- Provides a mutually understood topic for dialogues
- Clearly represents the passage of time
- Reduces anxiety about what has, is, and will happen

Passage of time

- From when you got up this morning
 - Your plan for the day
- To when you get home this evening
 - Future plans
- Reflecting back on the day
 - “Wow, the presentations were great!”

This is so much of what we talk about.

Imagine waking up and...

- No idea what time it is
- No idea of what will happen today
- No idea how soon something might happen
- No idea of the expectations for what will happen
- Problems detecting the true passage of time
 - “Today the minutes seem like hours, the hours go so slowly, and still the sky is light.”
- Will I like it?

To start

- The child needs to have a communication system of some kind and a plan for its development
 - Objects
 - Pictures
 - Signs
 - Words
- Activity routines must be in place
 - A calendar makes things more predictable
 - So you have to have a predictable schedule



This is an anticipation calendar

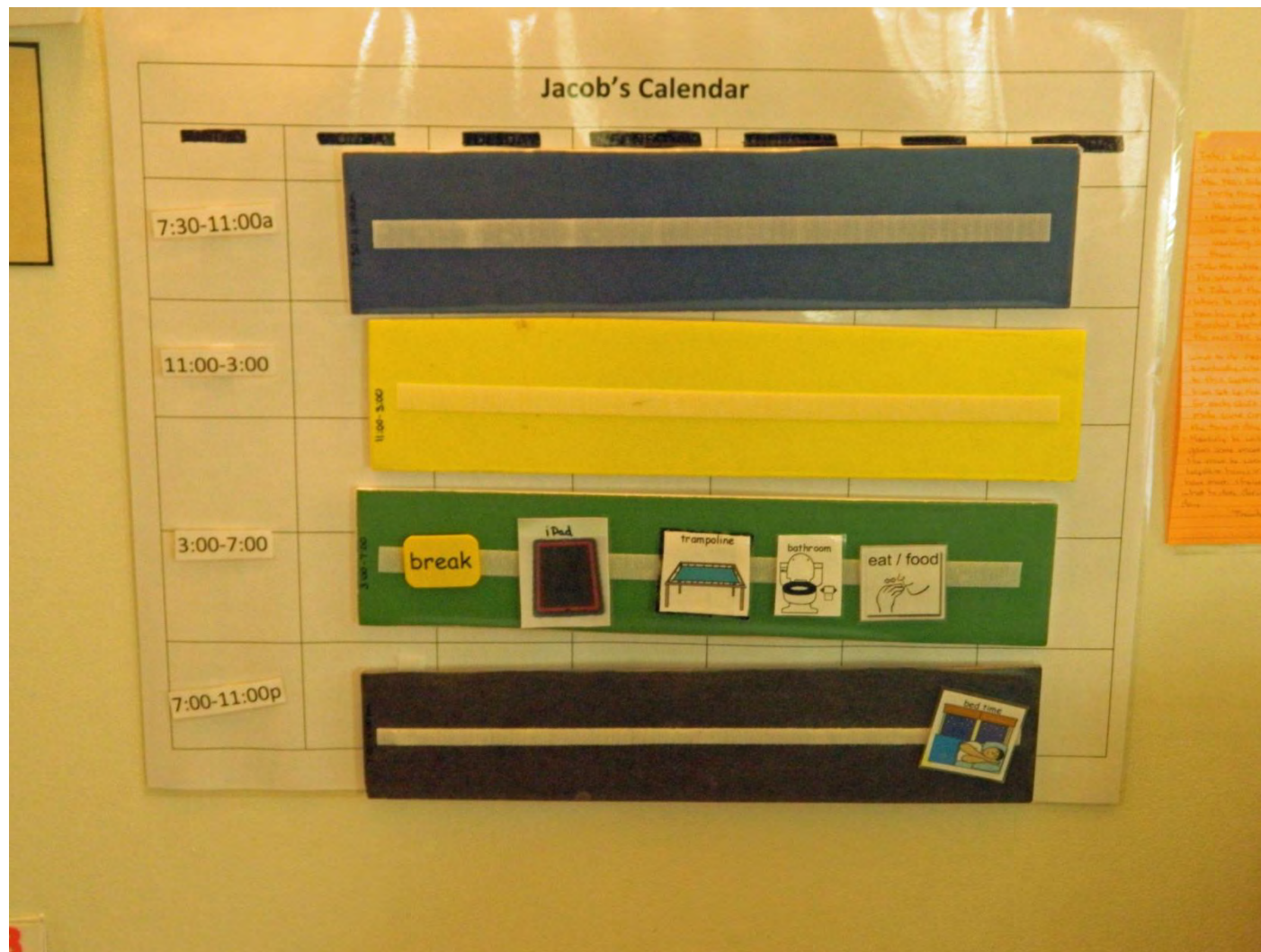
- Two distinctive baskets
- One has an object that represents an activity
- The other is the finish basket where the object will be placed once the activity is done.
- The object may represent “free play time.”
- When the child feels the object, the child knows it is time to go into the play room and do whatever he or she wishes.
- When play time is over, the object will be put in the finish basket.

A bit more sophisticated



This is a simple daily calendar. It shows a sequence of events.

Shift planning



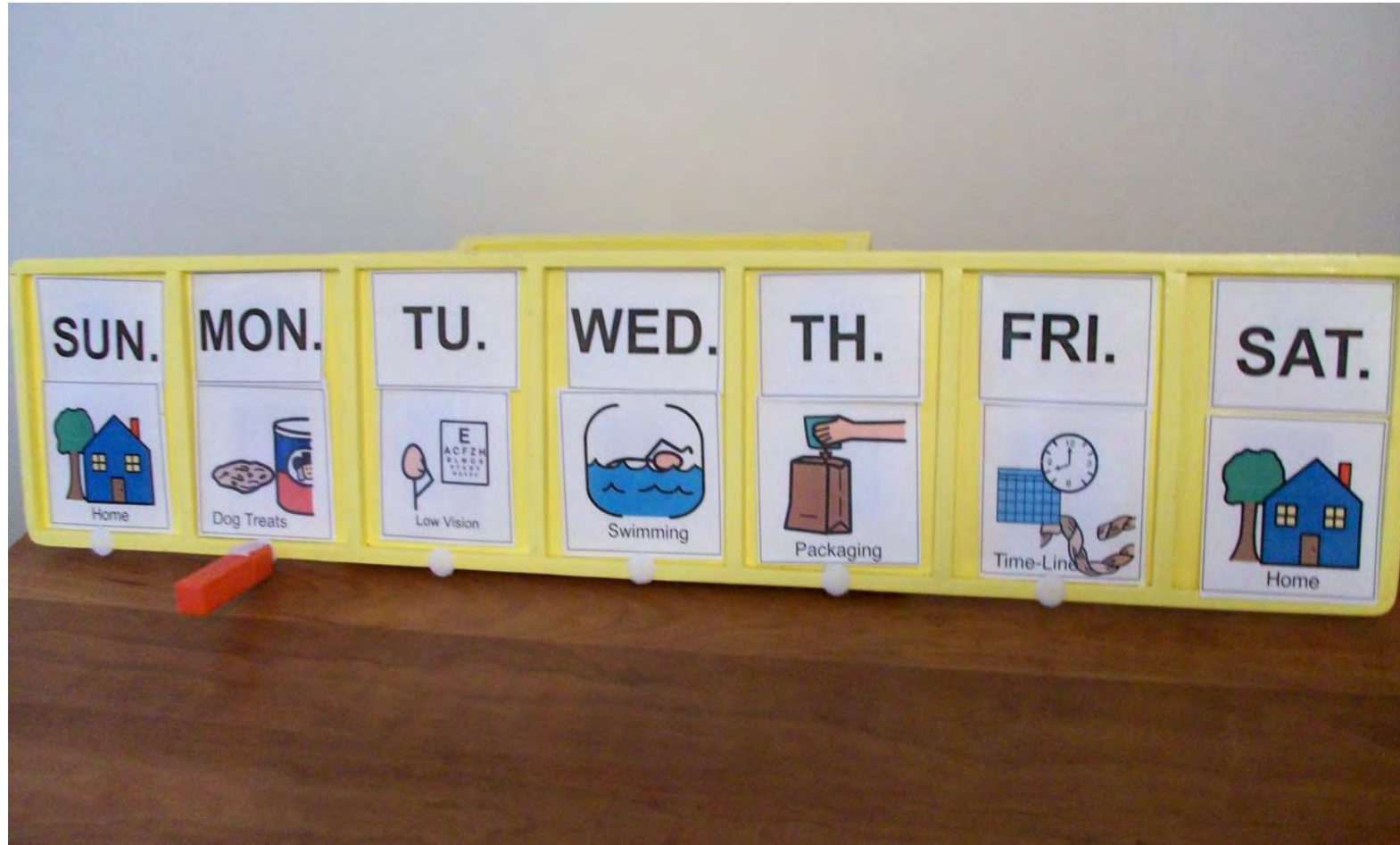
Finished



Predicting the week



If each day is distinct or highlighted



Activity routines



More



Communication

- Calendars provide an opportunity for conversation around the schedule.
 - What we are doing now
 - What we are doing later
 - What we just did
- At the end of each shift ideally go through the finish box and talk about what happened
- Life is good with a bit of predictability and review

“A feeling of worry, nervousness, or unease, typically about an imminent event or something with an uncertain outcome.”

- You still should consider pain, self-regulation, and changes in routine.
- But all of these can also lead to anxiety.
- Calendar systems and activity routines can reduce anxiety by making life more predictable.



Saturday, August 1, 2015
Breakout Session #40: 2:15 - 3:15pm
Schaumburg E-G

Feeding problems and how they can be related to the heart

**Nicole Corsten-Janssen, MD &
Conny van Ravenswaaij, MD PhD**
**University of Groningen, University Medical
Center Groningen**

Presenter Information:

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

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

Presentation Abstract:

Most individuals with CHARGE syndrome have feeding problems. Feeding problems can have multiple causes. We focus on the relation between feeding problems and heart defects, especially on the less well known arch vessel anomalies. Arch vessel anomalies occur in CHARGE syndrome and may cause problems with feeding due to compression of the esophagus. It can be difficult to recognize this often treatable cause of feeding problems, so one should be aware of it.

Handout international CHARGE conference 2015

Feeding problems and its relation with the heart

Nicole Corsten-Janssen

CHARGE syndrome and feeding problems

Difficulties with feeding and/or swallowing are a frequent problem in children with CHARGE syndrome. We know that feeding and swallowing problems can have multiple causes in CHARGE syndrome including cranial nerve dysfunction, anosmia (=no smell), choanal atresia, cleft lip palate, gastro-esophageal reflux disease and constipation. We will focus on the relation between feeding problems and the heart.

Feeding and swallowing in general

Feeding is important for a child's physical and mental growth. Feeding and swallowing can be divided into several phases:

The pre-oral phase starts before food or liquids gets into the mouth. It starts by looking at the food, producing saliva and putting the food into the mouth etc.

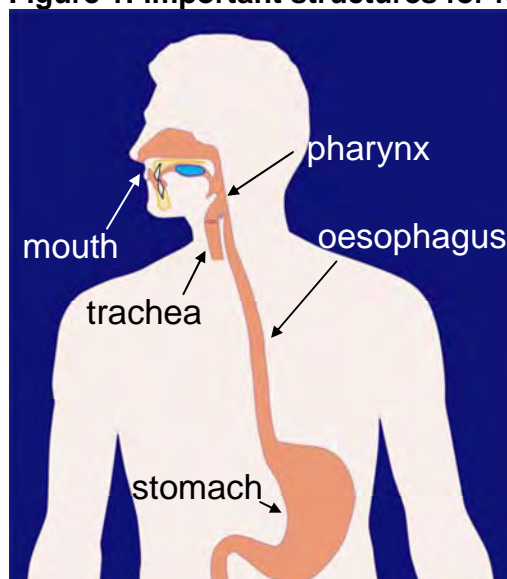
The oral phase consists of sucking, chewing, and moving food or liquid into the throat.

The pharyngeal phase is important for squeezing food down the throat, simultaneously closing off the airway.

The esophageal phase consists of moving the food down the esophagus and into the stomach.

All this illustrates, that feeding and swallowing are complex processes in which lots of things can go wrong.

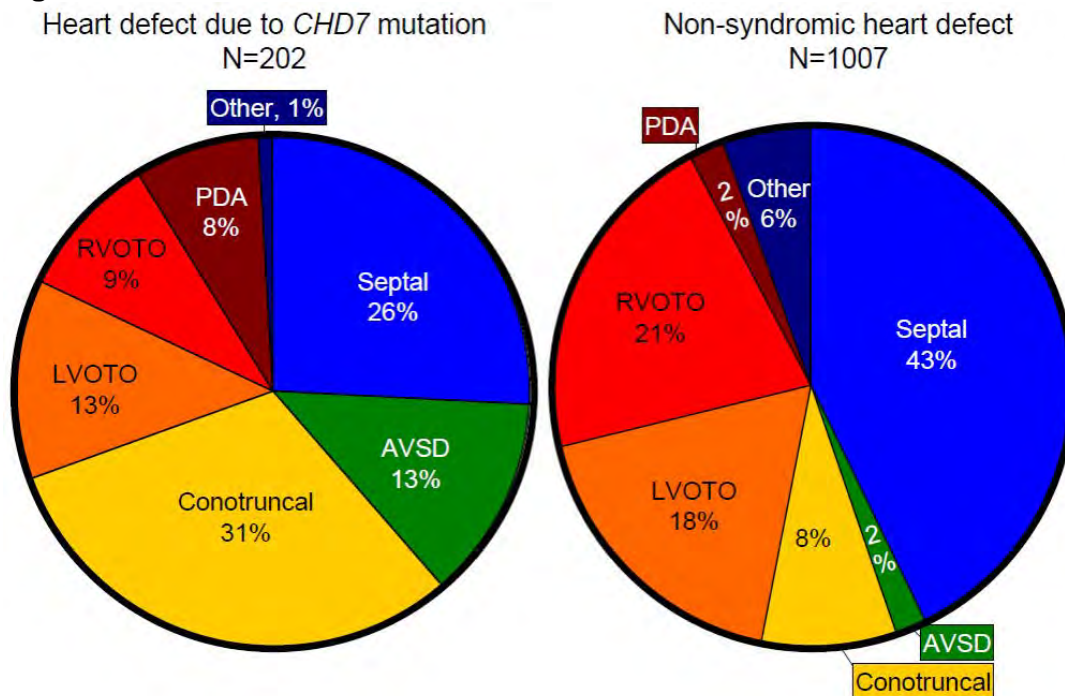
Figure 1: important structures for feeding and swallowing



Congenital heart defects in CHARGE syndrome

Most individuals with CHARGE syndrome have a defect (=mutation) in a hereditary factor, the *CHD7* gene. We previously studied heart defects in 299 individuals with CHARGE syndrome and a proven *CHD7* mutation¹. Congenital heart defects were present in 220 (74%) of the individuals with CHARGE syndrome. The type of heart defects were variable, but remarkably two types of heart defects occurred more often in individuals with CHARGE syndrome than in individuals with non-syndromic heart defects (figure 2). The first type is an atrioventricular septal defect (AVSD) or atrioventricular canal defect, in which an opening in the middle of the heart exists. The second type is a conotruncal or outflow tract abnormality. Examples of this type are a tetralogy of Fallot and abnormalities of the large vessels of the heart that we call arch vessel anomalies (figure 3).

Figure 2



In these pie charts heart defects due to CHD7 mutations are compared to non syndromic heart defects. Conotruncal heart defects (yellow) and atrioventricular septal defects (AVSD, green) are overrepresented in CHARGE patients.¹ Conotruncal heart defects include: tetralogy of Fallot, double outflow right ventricle and arch vessels anomalies.

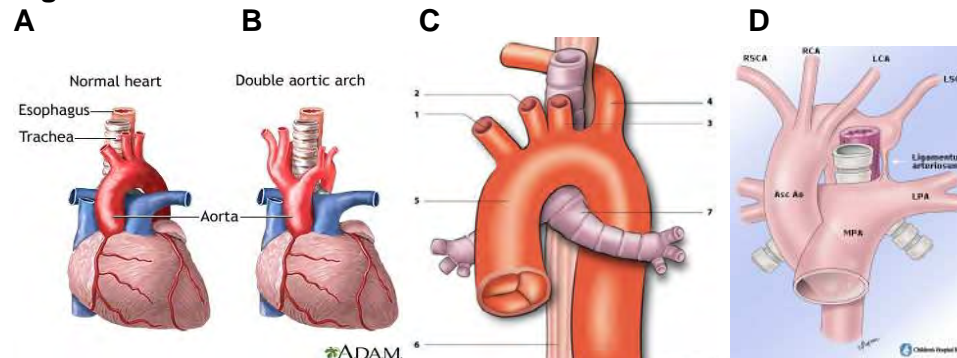
Feeding or swallowing problems and its relation with the heart

Feeding a baby with a heart defect can be challenging. A child with a heart defect may grow more slowly during infancy and childhood. Growth often varies according to the type and severity of heart defect. A heart defect alone can be a cause of feeding difficulty because the effort required to feed is greater for a child with a heart defect.

We will focus on a specific type of heart defects: defects of the large vessel leaving the heart called the aorta. We call these type of defects "arch vessel anomalies (figure 3)". As is shown in figure 3A the aorta lies close to the trachea (airway) and esophagus. An arch vessel anomaly may cause compression of these structures and in this way cause problems with breathing or feeding. This is a less known, but often treatable cause of feeding problems in CHARGE syndrome.

We studied arch vessel anomalies in CHARGE syndrome, because we had heard the story of a child with CHARGE syndrome who experienced periods of choking due to an aberrant subclavian artery (figure 3C).

Figure 3 arch vessel anomalies



A. A normal heart with a normal aortic arch and vessels in red on the left picture.

B. An example of a vascular ring caused by a double aortic arch

C. An aberrant subclavian artery

D. Right sided aortic arch

In our cohort 42 (14%) of the 299 individuals with CHARGE syndrome definitely had an aortic arch anomaly, mostly aberrant subclavian artery or right aortic arch (figure 3C and 3D). The individuals with aortic arch anomalies usually had other congenital heart defects as well (81%). Most patients had feeding difficulty, but we don't know if this was related to their arch vessel anomaly.

Based on our study, arch vessel anomalies occur in a significant proportion of individuals with CHARGE syndrome. We think the prevalence of arch vessel anomalies might be even higher than we found because arch vessel anomalies are easily missed on ultrasound and our data are not complete. Further studies are needed to exactly establish the prevalence of arch vessel anomalies in CHARGE syndrome and their contribution to feeding problems.

Take home message

Feeding and swallowing problems can have multiple causes in CHARGE syndrome. A rare but treatable cause is compression of the esophagus or airway by an arch vessel anomaly. If your child has problems with feeding, breathing or swallowing please discuss with your doctor if an arch vessel anomaly has been excluded.

Reference

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



Saturday, August 1, 2015
Breakout Session #41: 3:30 - 4:30pm
Schaumburg East

The Power of Jacob

Timothy S. Hartshorne, Ph.D.
Central Michigan University, Department of
Psychology
Panel of Jacob's caregivers:
Toni Hayward, Trevor Karr, Mandy Odren &
Amber Schwennessen

Presenter Information:

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

Presentation Abstract:

Disability is often associated with a great deal of discouragement. How can a person with severe limitations possibly contribute to society? Jacob is a 26 year old young man who functions on the severe end of the CHARGE continuum. In this presentation, some of Jacob's caregivers will talk about his impact on their lives.

The Power of Jacob

Tim Hartshorne

Toni Hayward, Trevor Karr, Mandy Odren, Amber Schwennesen

Abstract: Disability is often associated with a great deal of discouragement. How can a person with severe limitations possibly contribute to society? Jacob is a 26 year old young man who functions on the severe end of the CHARGE continuum. In this presentation, some of Jacob's caregivers will talk about his impact on their lives.

Presentation description: Disability is often associated with a great deal of discouragement. How can a person with severe limitations possibly contribute to society? What power do they have to contribute? Aren't they just a drain on society's resources? Think about all the extra medical and educational supports they require, and consider all of what their parents and siblings have to sacrifice in order to provide for them. On the other hand, what does it really mean to contribute? There are people in the world with great gifts and opportunity who accomplish virtually nothing. It is not the gifts you have that make a difference in the world; it is what you do with those gifts. Jacob is 26 years old and has CHARGE syndrome, is deafblind, communicates through push/pull and pictures, and has many physical problems. Nevertheless, Jacob has changed many lives for the better, making a big impact on kids he went to school with and often their families, certainly his family, and his care givers. There are rich and powerful people in the world who have done nothing but ill with their gifts, and here is this kid with huge disabilities changing lives for the better. CHARGE syndrome does not define who our children are. We live in a society that does not get that disability is just different ability and that our kids have much to offer the world. Society needs to recognize that the investments we make in all children pay off. Jacob lives in his own home with continual support from caregivers. This presentation will feature four of his support staff discussing their experience of Jacob and his abilities, and how his use of those abilities has impacted their lives.

Two useful perspectives:

(1) Perfect courage is the courage to be imperfect – Not one of us is perfect, and we must learn to live courageously knowing that we are imperfect.

(2) A psychology of use and not possession – It is not what gifts we have been given that matter; what matters are the contributions we make with the gifts we possess.



**Saturday, August 1, 2015
Breakout Session #42: 3:30 - 4:30pm
Schaumburg E-G**

**Ataxia in CHARGE patients: a novel
feature?**

**Christa (C.M.) de Geus, MD
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**Collaborators: Dr. D.A. Sival, Dr. L.C. Meiners,
Prof. Dr. C.M.A. van Ravenswaaij-Arts,
University of Groningen, University Medical
Center Groningen**

Presenter Information:

Christa de Geus is a medical doctor in training to become a clinical geneticist. Within clinical genetics she has a particular interest in neurology and neuroradiology.

In 2014 she joined the research group of prof. Conny van Ravenswaaij as a PhD student. Her PhD focuses on neurological symptoms in CHARGE syndrome.

Presentation Abstract:

One of the problems that children with CHARGE face is a lack of balance due to absence of the semicircular canals. Recent research has shown that cerebellar anomalies occur in 55% of CHARGE patients. The cerebellum plays an important role in balance and coordination. We wondered whether cerebellar dysfunction may contribute to balance and coordination issues (ataxia) in CHARGE syndrome and have started a study investigating this.



Ataxia in CHARGE syndrome, a novel feature?

About the presenter



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

Introduction

Children with CHARGE syndrome often have neurological problems, such as low muscle tone, delayed motor milestones and poor balance.

Maintaining balance is a complicated skill to develop. First of all, we can use lots of different types of signals to help determine the position of our limbs, body and head:

- The vestibular system in the inner ear gives the brain information on the position of the head. This helps you know which way your head is tilted and helps detect turning and speed changes.
- The eyes give the brain visual information, telling you which way is up.
- Specialised spindle cells in the muscles tell the brain where different parts of the body are in relation to each other. This helps you know where your body parts are, whether you can see them or not.
- Sensory nerves give the brain information about pressure on the skin: for instance the pressure on the bottom of your feet, when you are standing.

The brain integrates all these different types of signals and then decides which muscles to activate to maintain balance.

Sensory input for balance in CHARGE

Typical CHARGE problems can affect many of these types of sensory input:

- most CHARGE patients have dysplasia of the semicircular canals. The semicircular canals are a part of the vestibular system in the inner ears.
- colobomas may affect eye sight
- low muscle tone can interfere with the ability of the muscles to correct for balance disturbances

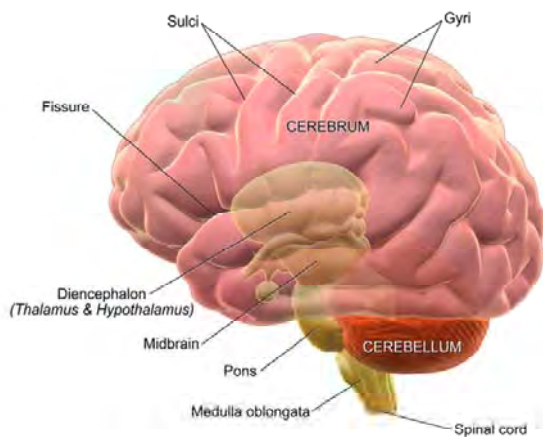
All in all, it is not surprising that CHARGE patients often have trouble balancing!

The role of the cerebellum in balance

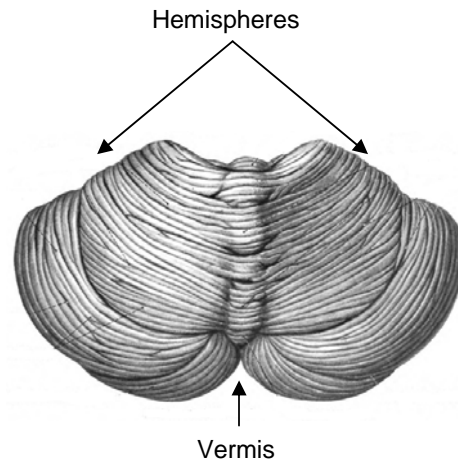
The part of the brain that plays the most important role in balance is the cerebellum. It integrates all the incoming sensory information, controls balance and fine-tunes our movements.

The cerebellum ("small brain") is located at the back and the base of the brain (see figure A). The cerebellum consists of a central part (the vermis) which connects two

hemispheres. Postural balance and gait are mainly controlled from the vermis, while coordination of the arms and legs is controlled by the hemispheres (see figure B). When a person has cerebellar problems, they will often have difficulties with their balance and coordination. A person may walk with a staggering gait, slur their speech or be imprecise in their movements. This is called ataxia.



A: Brain. (Image from: Blausen.com)



B: cerebellum. (Image from Sobotta anatomical atlas)

The cerebellum in CHARGE syndrome

Recently we found that in a small group of patients, cerebellar anomalies occurred in approximately half of them. We mainly saw congenital anomalies of the cerebellar vermis. Because of the function of the vermis in regulating balance, we wondered whether vermis anomalies could be an extra cause of balance problems in CHARGE syndrome.

To determine this, we are conducting a study in which we are evaluating the presence of neurological functioning and ataxia in CHARGE patients.

Methods

We hope to include 40 patients with a proven *CHD7* mutation. We are collecting the following data:

- systematic (re)evaluation of magnetic resonance imaging (MR imaging, MRI).
- neurological evaluation by a pediatric neurologist
- evaluation of (possible) ataxia with two validated ataxia scales: the SARA and PBS. These scales consist of a number of different tasks, such as walking, standing on one leg or pointing. The tests are videotaped.
- Ultrasound of muscles in a subset of patients

Preliminary results

As of May 2015, we have included 17 patients in the neurological evaluation. Approximately a third of these were able to complete the SARA/PBS scores. For 12 patients we have MRI data available. In addition, 22 patients were included for whom only MRI data are available so far.

Neuroradiology

We have found that approximately half (52%) of patients have a cerebellar abnormality, with vermis abnormalities (42%) more common than abnormalities of the cerebellar hemispheres (14%). Abnormalities range from hypoplasia, abnormal foliation of the vermis or the hemispheres, rotation of the vermis or a combination of these.

General neurological evaluation

General neurological evaluation has so far mainly shown that there is a strikingly common occurrence of sloping shoulders. In a small subset of patients this leads to functional problems. We are currently assessing this further by performing muscle ultrasounds in a concurrent project.

Ataxia scores

Preliminary evaluation shows that CHARGE patients may score higher on ataxia scores than healthy children. This is not unexpected, as balance problems are part of the score. Further analysis to determine whether patients with cerebellar anomalies have more problems than patients with a normal cerebellum, will have to wait until our study population is larger. In fact, we will be including CHARGE patients at the conference!

Future

In the nearby future we hope to be able to provide more insight into the occurrence of ataxia in CHARGE syndrome and the role of the cerebellum in CHARGE. In a related project, we are investigating the cause of the 'sloping shoulders' we see in many of our patients. With these projects we hope to provide more insight into neurological functioning in CHARGE syndrome.

Acknowledgements

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- Dr. D.A. Sival, pediatric neurologist, UMCG
- Dr. Linda Meiners, pediatric neuroradiologist UMCG

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Contact information

For further information or questions, please do contact me at: charge@umcg.nl

References

1. Yu T, Meiners LC, Danielsen K, et al. Deregulated FGF and homeotic gene expression underlies cerebellar vermis hypoplasia in CHARGE syndrome. *Elife* 2013;2.