10th International
CHARGE Syndrome Conference

July 29-31, 2011
Rosen Shingle Creek
Orlando, Florida

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CONFERENCE PROGRAM AND HANDOUTS

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The CHARGE Syndrome Foundation, Inc.
www.chargesyndrome.org

Wednesday, July 27th, 2011

9:00-6:00	**Conference Registration & Sales Center open at registration desk in Transportation Lobby** ***(between hotel lobby and conference center)***
9:00-6:00	**Professional Day Registration open at registration desk in Transportation Lobby** ***(between hotel lobby and conference center)***

Thursday, July 28th, 2011

6:00-6:00	**Conference Registration & Sales Center open at registration desk in Transportation Lobby** ***(between hotel lobby and conference center)***
6:00-12:00	**Professional Day Registration at registration desk in Transportation Lobby** ***(between hotel lobby and conference center)***
8:00-5:00	**Professional Day Conference in Wekiwa # 3-5**
12:00-6:00	**Camp Discovery & Camp Explorer registration open in Transportation Lobby** ***(between hotel lobby and conference center)***
7:00-9:00	**Reception in Conway Room – Light snacks included. Cash bar available**

Friday, July 29th, 2011

7:00-6:00	**Conference Registration open at registration desk in Panzacola Registration *(across from Panzacola F-1)***
7:00-6:00	**Sales Center open at Panzacola Registration *(across from Panzacola F-1)***
7:00-9:00	**Breakfast in Panzacola F&G**
7:00-9:00	**Camp Discovery & Camp Explorer registration at Sebastian Registration *(across from Sebastian I-1)***
8:00-6:00	**Exhibits Open in Pre-function area outside Panzacola F**
8:30-11:45	**Camp Discovery & Camp Explorer morning session in Panzacola H & Sebastian I**
9:00–9:30	**Welcome and opening remarks in Panzacola F&G**
9:30–9:45	**General Session – Tiina Urv: Navigating the NIH**
9:45-10:15	**General Session – John Halloran: Neuroplasticity and high expectations**
10:15-10:45	**Break with beverages**
10:45-11:45	**Breakout Session #1—See below**
11:45-1:00	**Lunch in Sebastian K**
12:30-5:15	**Camp Discovery & Camp Explorer afternoon session in Panzacola H & Sebastian I**
1:00-2:15	**Breakout Session #2—See below**
2:30-3:45	**Breakout Session #3—See below**
3:45-5:00	**Breakout Session #4—See below – snacks and drinks to be served during Information Central**
5:30-6:45	**Dinner in Sebastian K**
7:00-9:30	**2nd Annual CHARGE Syndrome "CONFERENCE IDOL" Talent Extravaganza in Panzacola F & G**

	Panzacola F3	**Panzacola F1 & F2**	**Panzacola F4**	**Panzacola G1**	**Panzacola G2**	**Wekiwa 3 & 4**	**Wekiwa 5**	**Wekiwa 6**
Breakout Session #1 10:45 – 11:45	**# 1** **Grandparents Session**	**# 2** **Perkins' Pal Program** **Stelzer & Gavin**	**# 3** **Fathers Forum** **Hartshorne & Gaudin**	**# 4** **CHARGE 101** **Hefner & Blake**	**# 5** **So many ways to have a conversation** **Majors**	**# 6**	**# 7** **Technology for learning and fun** **Cooper**	**SIB SHOP#1** **12:15 – 3:30 Advance Registration Required**
Breakout Session #2 1:00 – 2:15	**Information Central 2:00 – 5:00**		**# 8** **Personal Case Study of Post-secondary transition** **Sence & Sheriff**	**# 9** **CHARGE 102** **Davenport & Last**	**# 10** **Problems with self regulation** **Hartshorne**	**# 11** **A disturbance in the force** **Beals**	**# 12** **Understanding my team (English)** **Rodriguez-Gil**	
Breakout Session #3 2:30-3:45			**# 13** **Transition to independent living** **Hartshornes**	**# 14** **CHARGE 103** **Hefner, Blake, Davenport & Last**	**# 15** **What worked for you?** **Minkin**	**# 16** **Advances in CHD7 – Mice** **Martin, Hurd, Layman & Raphael**	**# 17** **Understanding my team (Spanish)** **Rodriguez-Gil**	
Breakout Session #4 3:45-5:00								

Saturday, July 30th, 2011

7:00–5:00	**Conference Registration & Sales Center open at Panzacola Registration *(across from Panzacola F-1)***
7:00-9:00	**Breakfast in Panzacola F&G**
8:30-11:45	**Camp Discovery & Camp Explorer morning session Panzacola H & Sebastian I**
8:00-2:30	**Exhibits Open in Pre-function area outside Panzacola F**
9:00–9:15	**Welcome and housekeeping in Panzacola F&G**
9:15-10:15	**Foundation General Board Meeting: Reports, Updates and Awards**
10:15-10:45	**Break with beverages**
10:45-11:45	**Breakout Session #5—See below**
11:45-1:00	**Lunch—In Sebastian K**
12:30-4:45	**Camp Discovery & Camp Explorer afternoon session in Panzacola H & Sebastian I**
1:00-2:00	**Breakout Session #6—See below**
2:15-3:15	**Breakout Session #7—See below**
3:15-3:45	**Break with snack**
3:45-4:45	**Breakout Session #8—See below**
6:30	**CHARGE-A-Palooza: Dinner, carnival and silent auction in Sebastian K & L**

	Panzacola F1 & F2	Panzacola F4	Panzacola G1	Panzacola G2	Wekiwa 3 & 4	Wekiwa 5	Wekiwa 6
Breakout Session #5 10:45 – 11:45	**# 18** **Taking back the IEP** **Steinbrick**	**# 19** **LAMP (part 1)** **Halloran**	**# 20** **Special needs planning program** **Ehlert**	**# 21** **Transition Strand Part 1: Social Skills** **Sacks**	**# 22** **Creating calm from CHARGE chaos** **Rose**	**# 23** **Molecular studies of CHD7** **Scacheri**	**SIB SHOP #2 8:30 – 11:30**
Breakout Session #6 1:00 – 2:00	**# 24** **Toilet Training** **Denno**	**# 25** **LAMP (part 2)** **Halloran**	**# 26** **Puberty & Growth Hormone (part 1)** **Van Ravenswaaij-Arts & Kirk**	**# 27** **Transition Strand Part 2: PCP** **Wiley**	**# 28** **Feeling good?** **Deuce**	**# 29** **Balance Group: Why does it work?** **Girardi**	
Breakout Session #7 2:15-3:15	**# 30** **Children…. A longitudinal study** **Last**	**# 31** **Person Centered Planning** **Salem-Hartshorne & Deuce**	**# 32** **Puberty & Growth Hormone (part 2)** **Kirk & Van Ravenswaaij-Arts**	**# 33** **Transition Strand Part 3: Adult Services** **Jordan**	**# 34** **Early Transitions** **Gerra**	**# 35** **My Intervener is deaf!** **Snyder & Morris**	**SIB SHOP #3 12:15 – 3:30**
Breakout Session #8 3:45-4:45	**# 36** **Ways and Means: Educating children with CHARGE in local schools.** **Brown**	**# 37** **Identifying the P in CHARGE** **Stratton**	**# 38** **ENT: Otolaryngologic considerations in CHARGE** **Cushing**	**# 39** **Transition Strand Part 4: Parent Panel** **McNulty & Parents**	**# 40** **The Heart and CHARGE syndrome** **Janssen**	**# 41** **National Cochlear Implant study** **Bashinski**	

Sunday, July 31st, 2011

7:00-9:00	**Breakfast in Panzacola F&G**
8:30-12:30	**Camp Discovery & Camp Explorer morning session Panzacola H & Sebastian I**
9:00–9:15	**Welcome and housekeeping in Panzacola F&G**
9:15–9:45	**Marianne Riggio**
9:45-10:45	**Young Adult Panel**
10:45-11:00	**Break**
11:00-11:45	**Deep Thoughts by David Brown**
11:45-12:00	**2011 Conference Farewell**

THE
CHARGE
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Navigating the NIH

Friday, 07/29/11
General Session: 9:30 – 9:45
Panzacola F & G

Tiina K. Urv, Ph.D.
***Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD)**
National Institutes of Health (NIH)

Presenter Information:

Tiina Urv, Ph.D., joined the Intellectual and Developmental Disabilities (IDD) Branch at the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), part of the National Institutes of Health (NIH) as a program director in October 2006. Dr. Urv is a developmental with a Ph.D. from Columbia University and over 25 years of experience working with individuals with intellectual disabilities in both clinical and research settings. Prior to joining the Branch, she was an assistant professor at University of Massachusetts Medical School's Eunice Kennedy Shriver Center and a research scientist at the New York State Institute for Basic Research in Developmental Disabilities. The focus of her work has been the behavioral aspect of aging and Alzheimer disease in adults with Down syndrome and developmental disabilities. Dr. Urv's work in the IDD Branch has focused on Newborn Screening of Rare Diseases and Fragile X syndrome (FXS).

Presentation Abstract:

Discussion of funding opportunities at the NIH for grants related to CHARGE syndrome.

THE
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FOUNDATION

Neuroplasticity and High Expectations

Friday 07/29/11
General Session: 9:45 – 10:15
Panzacola F & G

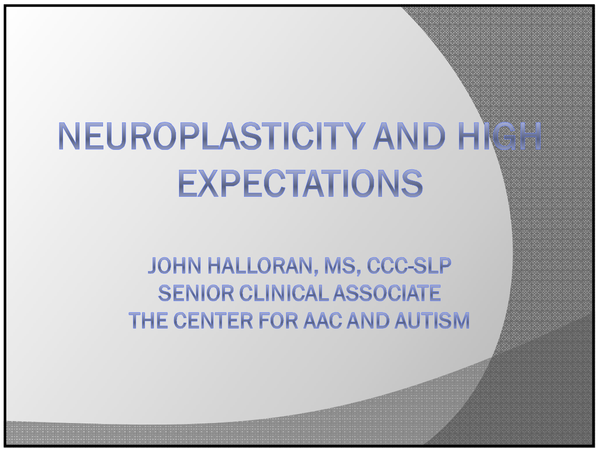
John Halloran M.S.CCC-SLP
Center for AAC and Autism

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. He also finds much reward in exploring ways to best implement assistive technology with children who have autism. John Halloran graduated from the University of Arkansas at Little Rock in 1990 with a bachelor's degree in Communication Disorders. He 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 is the primary developer of Language Acquisition through Motor Planning (LAMP) and presents internationally. John maintains membership in several professional organizations, including the American Speech Language Hearing Association and the Arkansas Speech Language Hearing Association.

Presentation Abstract:

Neuroplasticity refers to the relatively new understanding that our brains change throughout our lives based on experiences. We learn through our senses, and the ability to integrate sensations is critical for learning. There are no critical stages where learning must take place or the opportunity is lost, just windows where learning requires less effort. The benefits of multisensory integration on learning and the role that positive and negative reinforcers play in acquiring language through assistive technology will be discussed.



NEUROPLASTICITY AND HIGH EXPECTATIONS

JOHN HALLORAN, MS, CCC-SLP
SENIOR CLINICAL ASSOCIATE
THE CENTER FOR AAC AND AUTISM

THANK YOU!

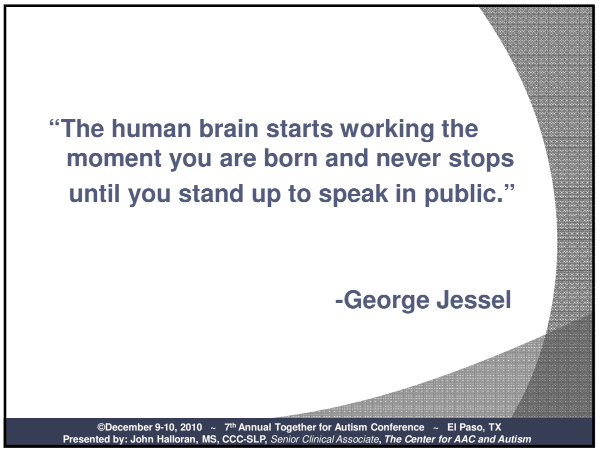
Children

Parents

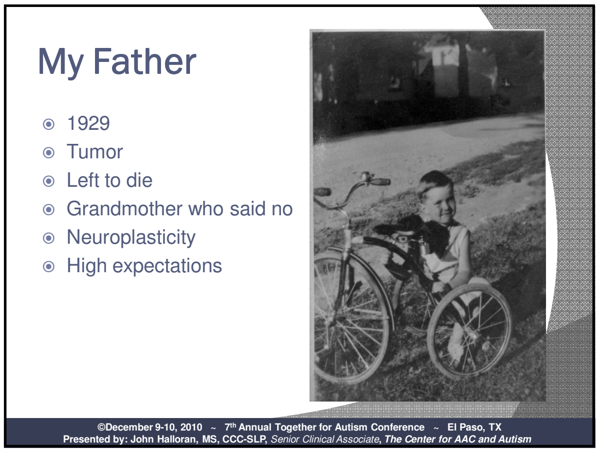
Teachers

Therapists

Committee of the CHARGE Conference

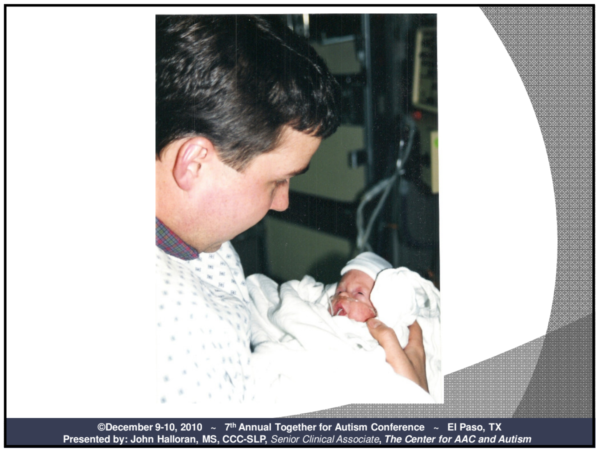


> **"The human brain starts working the moment you are born and never stops until you stand up to speak in public."**
>
> **-George Jessel**



My Father

- 1929
- Tumor
- Left to die
- Grandmother who said no
- Neuroplasticity
- High expectations



Rapid Neural Growth

250,000.00 neurons a minute for the two months prior to birth.

By 3 years of age: able to put complex sentences together

Frigid Mother Syndrome

- Evidence based
- Studies proved
- Children taken from their parents

What are we getting wrong today?

With what % graduating high school without functional speech, we cannot have the answers yet.

“The greatest obstacle to discovery is not ignorance, it is the illusion of knowledge.”

- Boorton

Neuroplasticity

The ability of the human brain to change as a result of one’s experiences. Learning may happen through change in the strength of the connections, by adding or removing connections, and by the formation of new cells.

Dispels concept that the brain does not change after critical stages.

Necessities for Plasticity

1. Novel (fun trip)
2. Repetitive (Golf Swing)
3. Person must give effort/try (crossword puzzle)
4. Trying to process sounds (new language)

Primary reasons we are not more successful in helping children with autism…

1. We reinforce what works instead of addressing the disorder.
    focus on visual strengths
    competitive nature of neurons (use it or lose it)
        example: Lazy eye
2. Not a homogenous disorder so we need many types of treatments based on the symptoms demonstrated.

Donald Hebb
The Organization of Behavior: A Neuropsychological Theory
1949

"Neurons that fire together wire together."

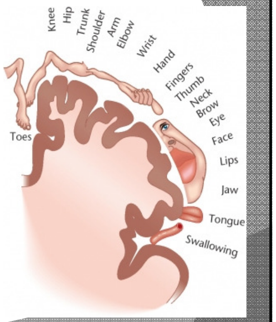
Critical Periods

A time in early development when there is a heightened sensitivity to certain stimuli, and learning is effortless but never does the ability to learn end.

Concept espoused by Hubel and Weisel

Penfield Maps of brain

Dr. Wilder Penfield used an image to illustrate the relationship between the motor cortex and the areas of the body it controlled



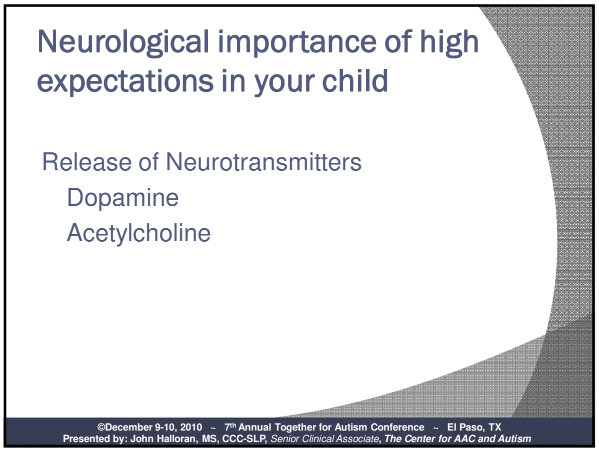
Treatments need to always address the integration of the sensations and see the brain as a whole rather than isolated regions.

Wobblers
Paul Bach-y-Rita

-Gentamicin kills vestibular system

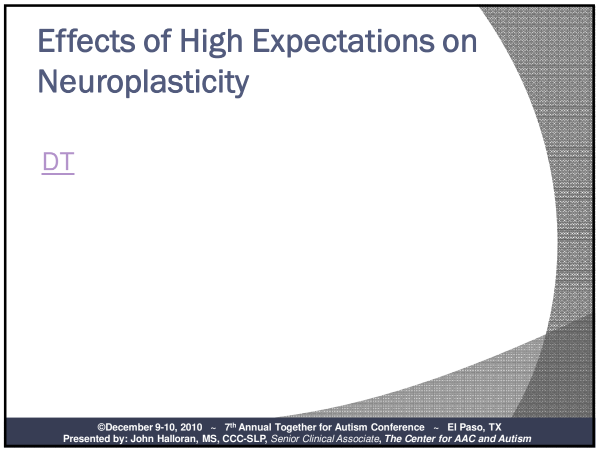
-Accelerometer

-Marvel of neuroplasticity- If certain key pathways are blocked then the brain uses older pathways to get around them.



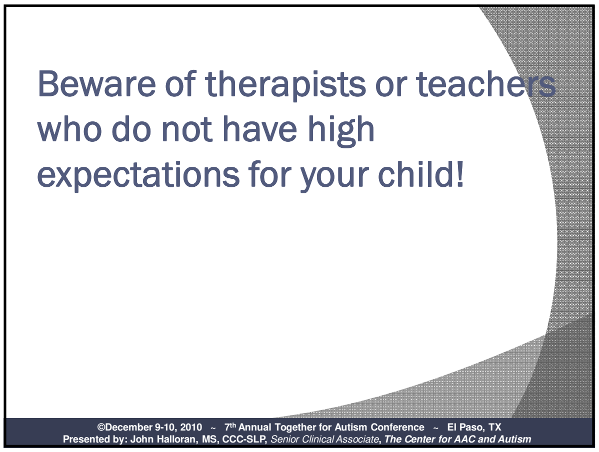
Neurological importance of high expectations in your child

Release of Neurotransmitters
- Dopamine
- Acetylcholine

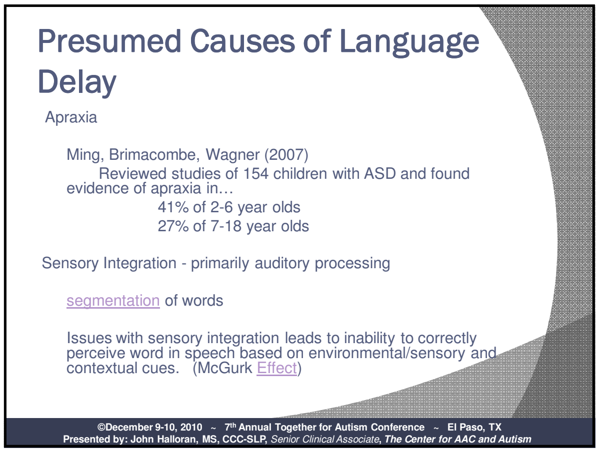


Effects of High Expectations on Neuroplasticity

DT



Beware of therapists or teachers who do not have high expectations for your child!



Presumed Causes of Language Delay

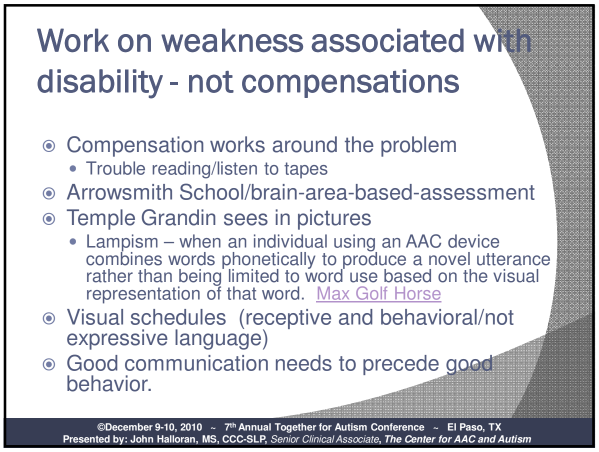
Apraxia

Ming, Brimacombe, Wagner (2007)
Reviewed studies of 154 children with ASD and found evidence of apraxia in…
41% of 2-6 year olds
27% of 7-18 year olds

Sensory Integration - primarily auditory processing

segmentation of words

Issues with sensory integration leads to inability to correctly perceive word in speech based on environmental/sensory and contextual cues. (McGurk Effect)



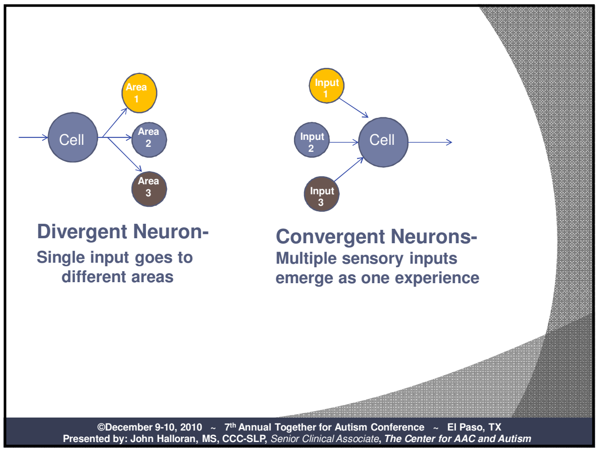
Work on weakness associated with disability - not compensations

- Compensation works around the problem
  - Trouble reading/listen to tapes
- Arrowsmith School/brain-area-based-assessment
- Temple Grandin sees in pictures
  - Lampism – when an individual using an AAC device combines words phonetically to produce a novel utterance rather than being limited to word use based on the visual representation of that word. Max Golf Horse
- Visual schedules (receptive and behavioral/not expressive language)
- Good communication needs to precede good behavior.

Halloran Proposed Rule

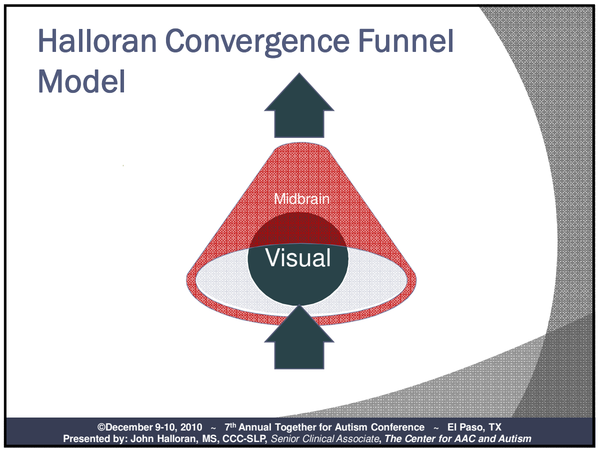
"Treatments that cause neurons to fire in the right order will lesson the disorder."

For language: somatosensory, then auditory and then visual. The hand becomes the articulator

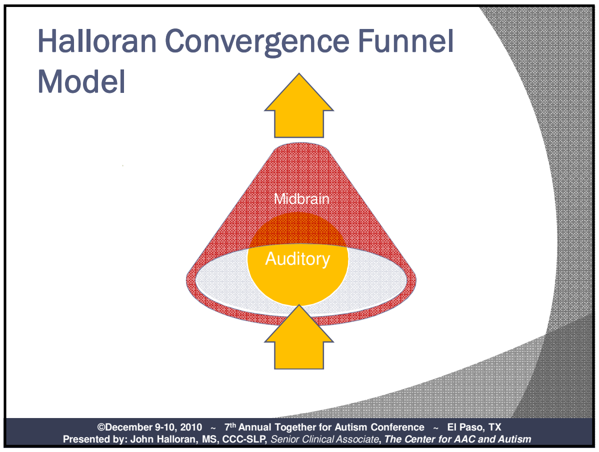


Divergent Neuron-
Single input goes to different areas

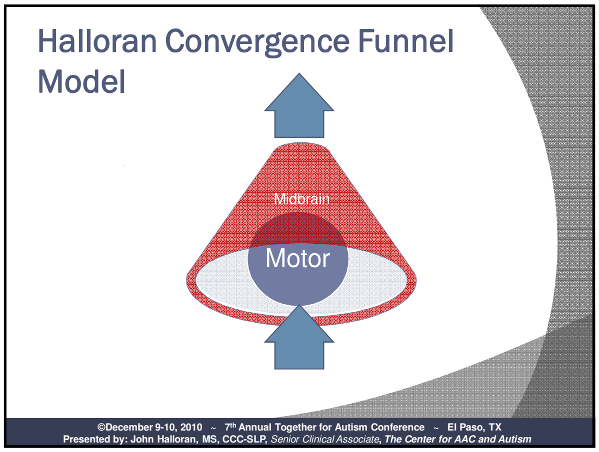
Convergent Neurons-
Multiple sensory inputs emerge as one experience



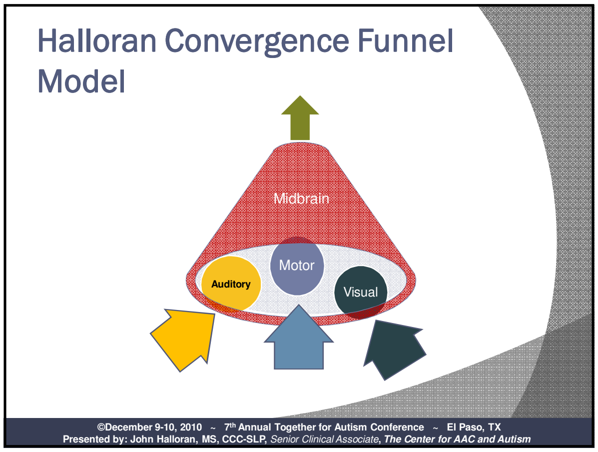
Halloran Convergence Funnel Model



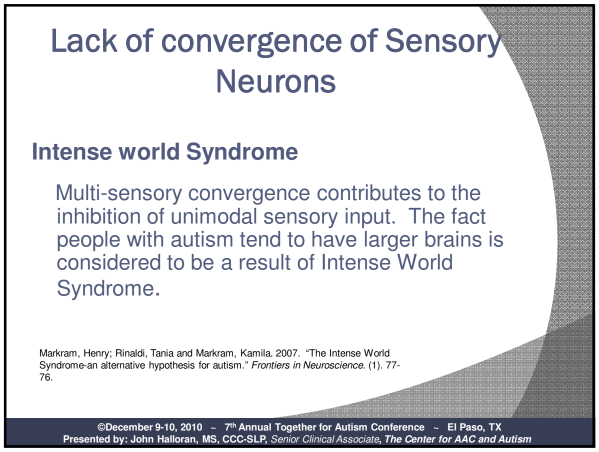
Halloran Convergence Funnel Model



Halloran Convergence Funnel Model



Halloran Convergence Funnel Model

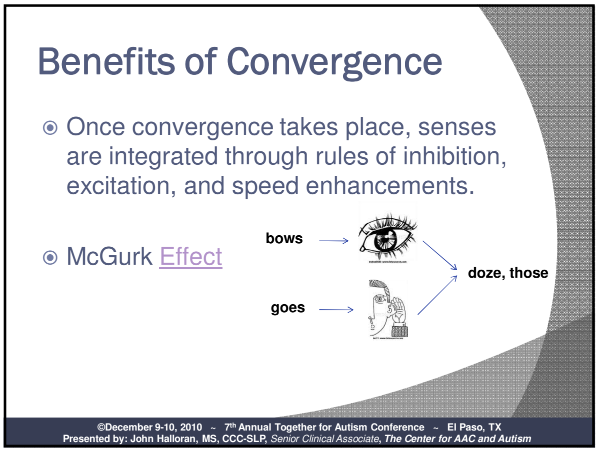


Lack of convergence of Sensory Neurons

Intense world Syndrome

Multi-sensory convergence contributes to the inhibition of unimodal sensory input. The fact people with autism tend to have larger brains is considered to be a result of Intense World Syndrome.

Markram, Henry; Rinaldi, Tania and Markram, Kamila. 2007. "The Intense World Syndrome-an alternative hypothesis for autism." *Frontiers in Neuroscience.* (1). 77-76.



Benefits of Convergence

- Once convergence takes place, senses are integrated through rules of inhibition, excitation, and speed enhancements.
- McGurk Effect

bows →

goes →

doze, those

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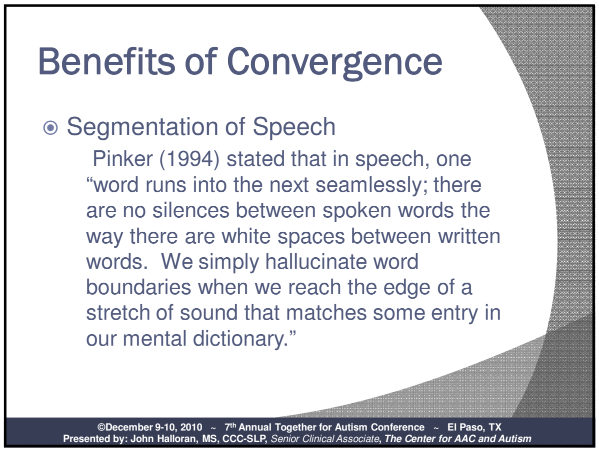

Benefits of Convergence

- Segmentation of Speech

  Pinker (1994) stated that in speech, one "word runs into the next seamlessly; there are no silences between spoken words the way there are white spaces between written words. We simply hallucinate word boundaries when we reach the edge of a stretch of sound that matches some entry in our mental dictionary."

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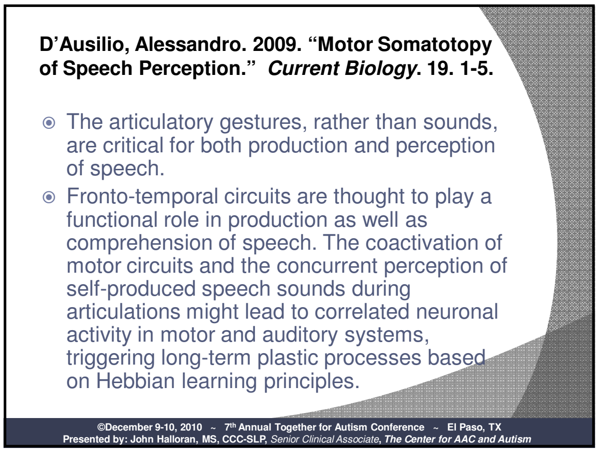

D'Ausilio, Alessandro. 2009. "Motor Somatotopy of Speech Perception." *Current Biology*. 19. 1-5.

- The articulatory gestures, rather than sounds, are critical for both production and perception of speech.
- Fronto-temporal circuits are thought to play a functional role in production as well as comprehension of speech. The coactivation of motor circuits and the concurrent perception of self-produced speech sounds during articulations might lead to correlated neuronal activity in motor and auditory systems, triggering long-term plastic processes based on Hebbian learning principles.

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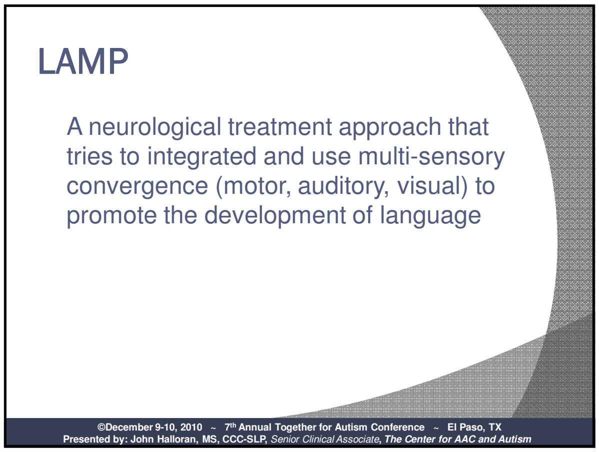

LAMP

A neurological treatment approach that tries to integrated and use multi-sensory convergence (motor, auditory, visual) to promote the development of language

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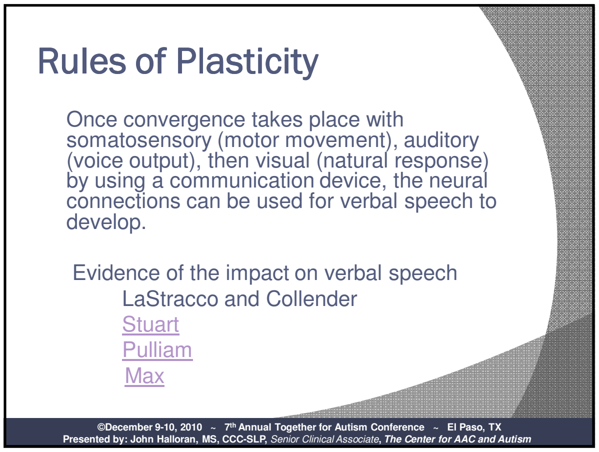

Rules of Plasticity

Once convergence takes place with somatosensory (motor movement), auditory (voice output), then visual (natural response) by using a communication device, the neural connections can be used for verbal speech to develop.

Evidence of the impact on verbal speech
  LaStracco and Collender
  Stuart
  Pulliam
  Max

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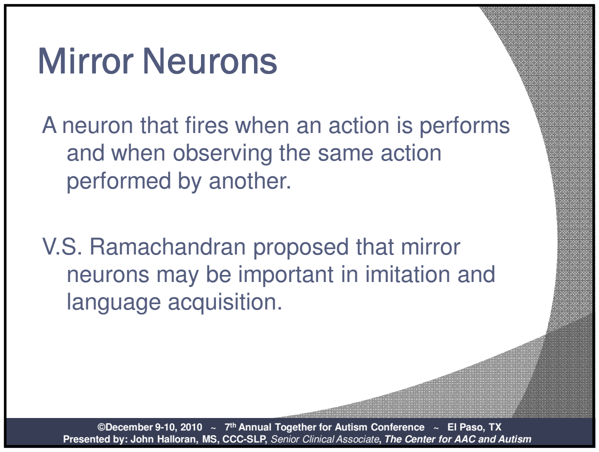

Mirror Neurons

A neuron that fires when an action is performs and when observing the same action performed by another.

V.S. Ramachandran proposed that mirror neurons may be important in imitation and language acquisition.

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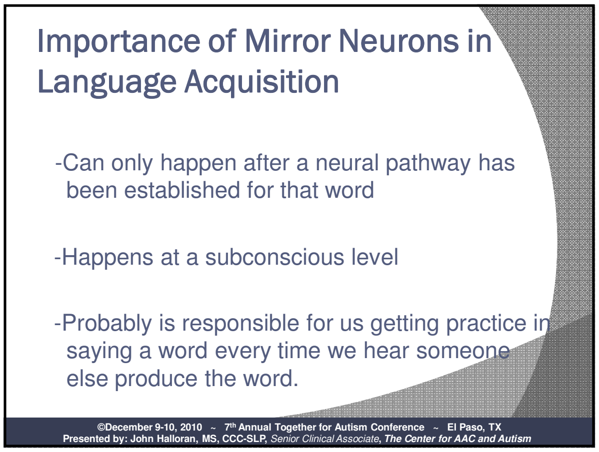

Importance of Mirror Neurons in Language Acquisition

-Can only happen after a neural pathway has been established for that word

-Happens at a subconscious level

-Probably is responsible for us getting practice in saying a word every time we hear someone else produce the word.

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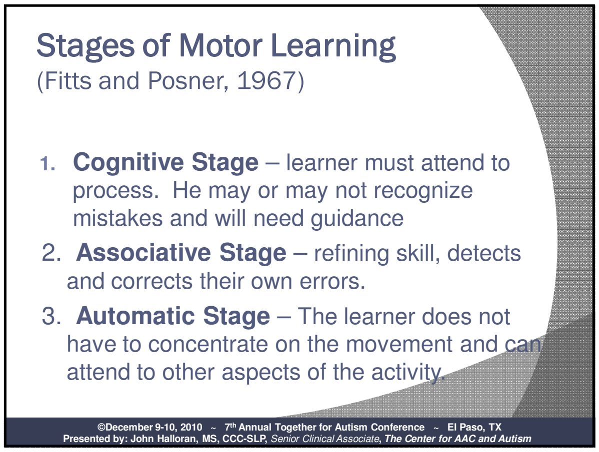

Stages of Motor Learning

(Fitts and Posner, 1967)

1. **Cognitive Stage** – learner must attend to process. He may or may not recognize mistakes and will need guidance
2. **Associative Stage** – refining skill, detects and corrects their own errors.
3. **Automatic Stage** – The learner does not have to concentrate on the movement and can attend to other aspects of the activity.

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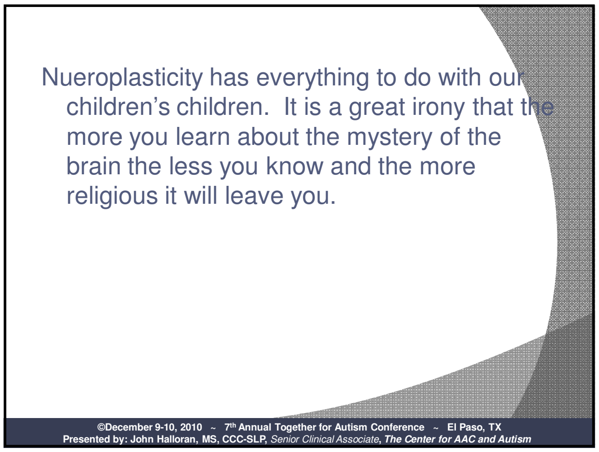

Nueroplasticity has everything to do with our children's children. It is a great irony that the more you learn about the mystery of the brain the less you know and the more religious it will leave you.

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Believe in Your Children but Understand Treatments

What is my therapist doing and how does that lead to a neurological change/improvement ?

Need more than a "stimulating environment."

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Presented by: John Halloran, MS, CCC-SLP, *Senior Clinical Associate*, ***The Center for AAC and Autism***

Families

- Believe
- Understand
- Share

- Lucas Dreams

©December 9-10, 2010 ~ 7th Annual Together for Autism Conference ~ El Paso, TX
Presented by: John Halloran, MS, CCC-SLP, *Senior Clinical Associate*, ***The Center for AAC and Autism***

THE
CHARGE
SYNDROME
FOUNDATION

Grandparents Gathering

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Panzacola F3

Len Stanger
Grandfather

Presenter Information:

Len Stanger is the grandfather of Megan Stanger, a delightful young lady who has CHARGE syndrome.

Presentation Abstract:

The purpose of this “gathering” is to allow grandparents of individuals with CHARGE to connect at the start of the conference and share information. The Foundation recognizes there are unique concerns and challenges for grandparents in the family of an individual with CHARGE syndrome.

THE CHARGE SYNDROME FOUNDATION

Perkins' Pal Program

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Panzacola F1 & F2

Sharon Stelzer, M.Ed.
Mary Gavin, M.Ed.
Perkins School for the Blind

Presenter Information:

Sharon Stelzer is a seasoned teacher at Perkins School for the Blind. She has taught children with Deafblindness from ages 6-18. For the past 25 years Sharon has taught a variety of children with CHARGE syndrome. She is currently teaching middle school and high school age teenagers with CHARGE syndrome. She has presented nationally and internationally on Teaching Strategies for Students with CHARGE as well as literacy and other communication topics.

Mary Gavin has worked with a variety of students at Perkins School for the Blind. She is currently a Preschool Teacher in the Deafblind Program. Mary works with young children with a variety of etiologies including CHARGE syndrome. Mary is currently finishing up her certification as a teacher of the visually impaired. Mary has done several presentations on teaching strategies and assessment for preschool age children.

Presentation Abstract:

An overview of a pilot program to develop social skills and mentorship in adolescent student with CHARGE syndrome. Several teenage students were paired with younger students ages 5-9 years old in the Deafblind Program at Perkins School for the Blind. Students will reflect on the project with presenters and attendees. Video clips of the initial sessions as well as subsequent sessions will be shared. Two of the teachers will share the rubric they developed for teaching social and mentoring skills to the adolescents. The presenters will share results and conclusions of how the program is going.

THE
CHARGE
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FOUNDATION

FATHERS' FORUM

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Panzacola F4

Moderators:
Tim Hartshorne & John Gaudin

Presenter Information:

Dr. Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. His particular interest is in understanding the challenging behavior exhibited by many individuals with CHARGE. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project.

John Gaudin is an auditor for the Windham School District in Huntsville, Texas. He also serves on the Board of the Texas Charger's Inc., a non-profit group dedicated to serving those with CHARGE syndrome along with their families and professionals who support them. He has facilitated the Fathers' Forum at the annual Texas Chargers Retreat for the past 3 years. He is happily married to Crissy, and they have 3 beautiful children – Jacob (10), Rachel(8), and Joey (6).

Presentation Abstract:

Purpose of the Fathers' Forum: The Fathers' Forum is an opportunity for fathers to share some of their unique perspectives on and experiences of having a child with CHARGE syndrome.

THE CHARGE SYNDROME FOUNDATION

CHARGE 101:

Diagnostic and Medical Features of CHARGE for New Families

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Panzacola G1

Meg Hefner, MS Genetic Counselor
St Louis University

Kim Blake, MD
Dalhousie University

Presenter Information:

Meg Hefner has been a genetic counselor for 30 years, most of those working on CHARGE syndrome. She was an author of the Booklet for Families and the Management Manual for Parents and was editor of the American Journal of Medical Genetics special issue on CHARGE and the 2010 book on CHARGE Syndrome from Plural Publishing. Meg was a founding member of the CHARGE Syndrome Foundation and continues as a Special Advisor to the Board and chair of the Research Committee. In July, 2011, Meg and Sandra Davenport started the first CHARGE Syndrome Clinic at Saint Louis University, where they are both faculty members in the Department of Pediatrics.

Dr. Kim Blake began her involvement with CHARGE 30 years ago at Great Ormand Street Hospital in the UK. She published some of the earliest papers on CHARGE and organized the UK family support group. At Dalhousie University in Halifax, she has continued her research on CHARGE, with focus on anesthesia, feeding, sleep, and issues of the adolescent and adult with CHARGE. Kim has recruited local faculty and mentored many medical students in doing research on CHARGE.

Presentation Abstract (see also CHARGE 102 and 103):

Meg will do a 20 minute overview of medical aspects of CHARGE syndrome: diagnostic criteria and other medical features.

Kim will do a 20 minute presentation focusing on three critical aspects of management in CHARGE: anesthesia, Botox injections and sleep apnea

20 minutes will be reserved for questions from the audience and “conference orientation” for new families. Written questions can be submitted for the 103 session.

Anaesthesia, Botox, Sleep Apnea and FAQ's

From Canada and Beyond

Dr. Kim Blake, MB, MRCP, FRCPC

10th International CHARGE Conference
Orlando Florida, July 2011

Objectives

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

There are Always Risks of Complications with Anaesthesia

- "…you sign a consent"
- Are you informed?
- Are Individuals with CHARGE Syndrome More at Risk?
- If yes, what are the risks and who should know?

Kennedy's Four ICU Admissions

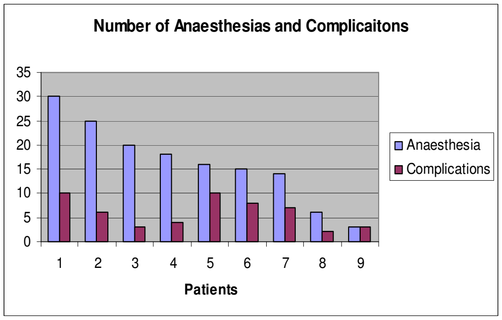
- 2 weeks – open heart surgery
- 6 months – G-tube/fundoplication extubation attempted (x 3)
- 18 months – aspiration pneumonia
- 6 yrs – heart surgery – pneumonia after heart surgery

Postoperative Airway Events of Individuals with CHARGE Syndrome

Population 9 children
Average age 11.8 years (± 8.0)
215 surgeries (average of 22 per child)
147 anaesthesias (average of 16 per child)
Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Blake K, MacCuspie J, Hartshorne TS, Roy M, Davenport SLH, Corsten G. *International Journal of Pediatric Otorhinolaryngology*, *Vo. 73, February 2009*

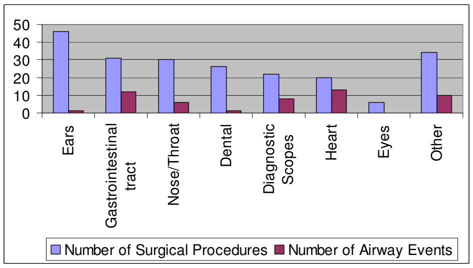
Results



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

Results

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



Results

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

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

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

Results

Feeding procedures and rates of postoperative airway events.

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

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

Summary

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

Blake K, MacCuspie J, Hartshorne TS, Roy M, Davenport SLH, Corsten G. *International Journal of Pediatric Otorhinolaryngology*, *Vo. 73*, *February 2009*

Summary cont'd

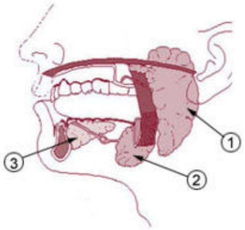
- High risk of complications in individuals with Nissen fundoplication and/or gastrotomy/jejunostomy tube
- What about individuals with CHD7 mutations who have mild clinical criteria?
  - Will they be at risk in the future?
  - Have they actually been challenged with surgeries?

Freddy at 2 Months

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

Site of Botox Injections

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



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

Freddy at 7 Months

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

Botox Injection

Submandibular Gland Via Ultrasound and Parotid Gland by Palpation

10 Botox Units/gland

Summary - Botox

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

MacCuspie, J., Blake, K., Corsten, G. Botulinum Toxin Injections into Salivary Glands to Decrease Oral secretions in CHARGE Syndrome: Prospective Case Study. AJMG (accepted 2010)

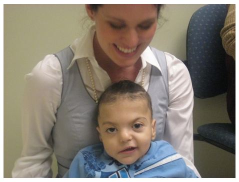
MacKenzie's Story

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

Understanding Sleep Apnea in Children with CHARGE Syndrome

In Children 0-14 years old

Authors:
Carrie-Lee Trider
Dr. Gerard Corsten
Dr. Debra Morrison
Meg Hefner
Dr. Saundra Davenport
Dr. Kim Blake



Carrie-Lee & Freddy

Ongoing study to participate contact:
Carrie-Lee Trider carrie_trider@hotmail.com
Kim Blake kblake@dal.ca 902-470-6499

Please feel free to contact us during this conference

Objectives

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

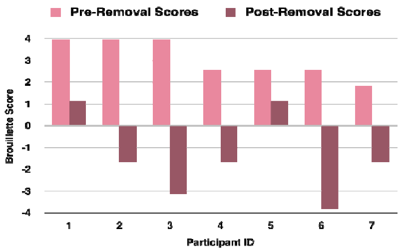
Thank you to the 34 Participants Results

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

Tonsils and Adenoids

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

Removal of Tonsils & Adenoids (Preliminary Results)



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

Take Home Messages

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

Take Home Messages

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

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So Many Ways to Have a Conversation

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Panzacola G2

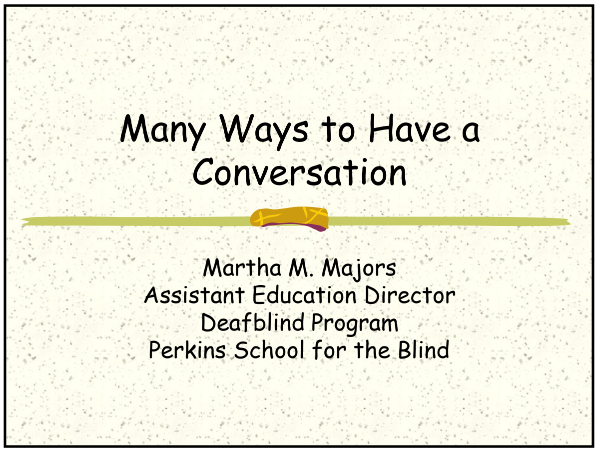
Martha Majors
Assistant Education Director
Deafblind Program
Perkins School for the Blind

Presenter Information:

Perkins School for the Blind developed a series of 3 webcasts related to CHARGE syndrome; Martha participated in these webcasts focusing on the educational implications for a child with CHARGE syndrome.

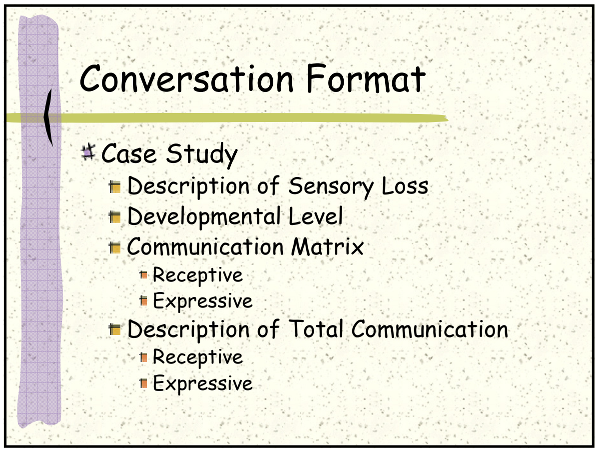
Presentation Abstract:

The development of communication for children with CHARGE syndrome can be challenging for both the child and their team (families and educators). For most students, receptive language is the area of strength; the use of expressive communication can be delayed and as a result there is a level of frustration that builds within the child. Current thinking includes the consistent use of total communication as well as assistive technology as a support. The best solutions come from a team approach where several disciplines come together to consider the strengths of the child that not only includes communication but use of vision, hearing, and physical presentation. In this way, the whole child is taken into consideration and outcomes can be more child specific and focused.



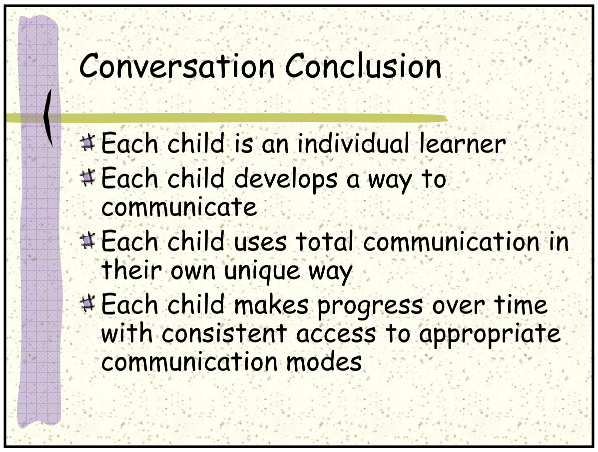
Many Ways to Have a Conversation

Martha M. Majors
Assistant Education Director
Deafblind Program
Perkins School for the Blind



Conversation Format

- Case Study
  - Description of Sensory Loss
  - Developmental Level
  - Communication Matrix
    - Receptive
    - Expressive
  - Description of Total Communication
    - Receptive
    - Expressive



Conversation Conclusion

- Each child is an individual learner
- Each child develops a way to communicate
- Each child uses total communication in their own unique way
- Each child makes progress over time with consistent access to appropriate communication modes

Technology for Fun and Learning

Friday, 07/29/11
Breakout Session #1: 10:45-11:45
Wekiwa 5

Holly Cooper, Ph.D.
Deafblind Educational Consultant
Texas Deafblind Outreach
Texas School for the Blind
and Visually Impaired

Presenter Information:

Holly Cooper was a classroom teacher of students with multiple disabilities and later an itinerant teacher of students with visual impairments, including deafblindness. She joined Texas School for the Blind and Visually Impaired Outreach program in 2001 as an assistive technology consultant, and in 2006 joined the deafblind team as an educational and assistive technology consultant. She travels the state of Texas supporting students with deafblindness, their teachers and families. Holly has a special interest in CHARGE syndrome, assistive technology and teacher preparation. She has also done adjunct teaching for Texas Tech University.

Presentation Abstract:

This presentation will be an overview of assistive technology and computer applications which are both fun and useful in the home or classroom. It will include a discussion of assistive technology with an emphasis on low vision accessibility, and accessibility for users with learning differences. It will include a brief overview of computer low vision accessibility settings and applications, magnification devices, alternative input devices, electronic book readers. Voice output switches and medium-tech augmentative communication devices will also be discussed.

Technology for Learning and Fun: Using Assistive Technology in the Home and Classroom

Holly Cooper, Ph.D.
Deafblind educational specialist, Texas Deafblind Project
Texas School for the Blind and Visually Impaired

1. Using Technology

It's not about what cool products are out there,
It's about what your student wants and needs to do.

- Vision magnification technology
- Amplification technology
- Computer assisted learning
- Internet use for research and learning
- Augmentative Communication devices
- Electronic books
- Assistive devices for daily living activities
- Hobbies and entertainment

2. Computer Use

A large monitor is a good thing, but there are other, sometimes better options.

- Built-in magnification and accessibility features
- Add-on software to magnify the screen image
- Easy key commands

3. Computer Magnification

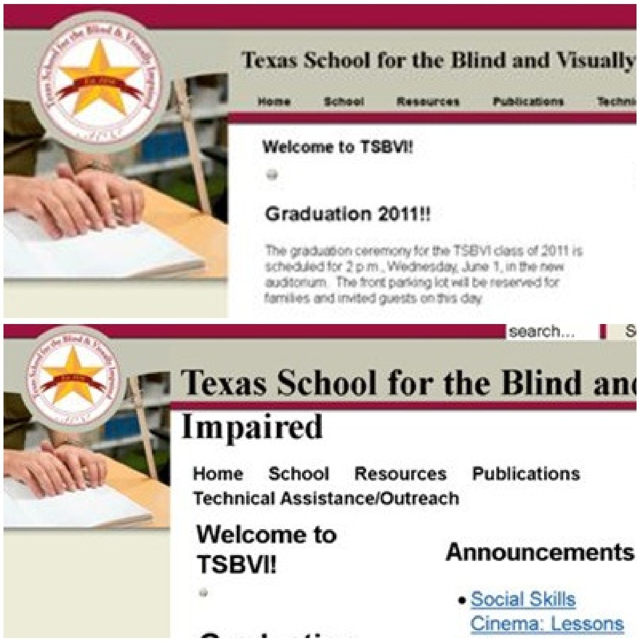
Windows and Mac have accessibility and magnification features.
Accessibility features make the icons, menus, text size and other controls larger
Magnification makes a selected area larger, but the user must scroll to see all areas of the screen.

4. Computer Magnification

Windows showing built-in magnification of a webpage.
Top half of screen is magnified
Bottom half of screen is the standard view
This can be confusing to inexperienced users!

5. Keyboard Controls

When using an internet browser, users can make the print size larger by
Holding down the “Ctrl” key and pressing “+“
The page will automatically re-format so the text doesn’t run off the screen



Texas School for the Blind and Visually

Home School Resources Publications Techni

Welcome to TSBVI!

Graduation 2011!!

The graduation ceremony for the TSBVI class of 2011 is scheduled for 2 p.m., Wednesday, June 1, in the new auditorium. The front parking lot will be reserved for families and invited guests on this day.

search... S

Texas School for the Blind an
Impaired

Home School Resources Publications
Technical Assistance/Outreach

Welcome to TSBVI!

Announcements

- Social Skills Cinema: Lessons

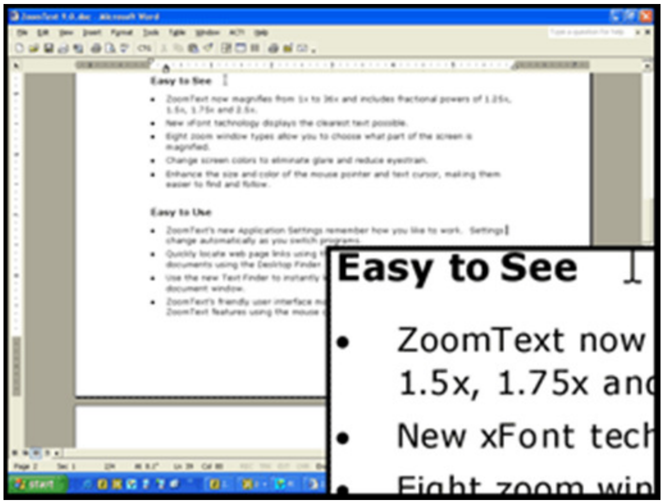
6. Mouse Pointer

Sometimes just making the mouse pointer larger can be helpful.
In windows, find the control panel, and select “mouse properties”
Some free customizable mouse pointers are available

7. Screen Magnification Software

For users who are using the computer at a middle or high school grade lever for research or learning applications, screen magnification software may be the best choice.

ZoomText
www.aisquared.com
Magic
www.freedomscientific.com
Supernova Magnifier
www.yourdolphin.com



Easy to See

- ZoomText now
  1.5x, 1.75x and
- New xFont tech
- Eight zoom win

8. Touch Screen

For younger users, or those who are less academic, a touch screen can make a computer much easier and more understandable.

9. Adapted Input Devices

Keyboard and mouse alternatives:
Zoomcaps stickers for a standard keyboard
Various vendors
Big keys
www.bigkeys.com
Intellitools keypad
www.intellitools.com
Track balls, etc.
www.infogrip.com

10. Tablet Computers

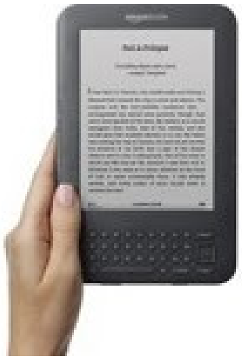
Tablet computers are light and portable and can be handy for games, web browsing, reading ebooks, email and texting:

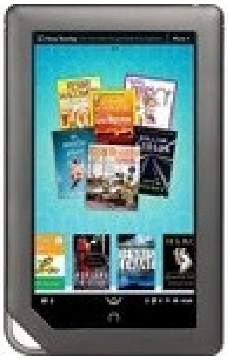
- iPad 2
- Tablet PC's

11. Electronic book readers

Ebook readers such as the

- Kindle
- Nook





Allow text size adjustment for large print options
Kindle is easier to see in bright light such as outdoors
Kindle has long battery life
Nook and others have a backlit screen, color display

12. Electronic book readers

Electronic book readers use special files known as ePub.
Free file converters are available
A textbook on CD in a .pdf format can be converted to ePub format and loaded into the electronic reader from a computer.

13. Magnifiers

- Optical magnifiers
- Video magnifiers

14. Amplification

- FM amplification
- Sound field generators
- Audio input alternatives to headphones

15. Communication

- Picture symbol displays
- Talking switches
- Voice output switch activated devices
- Dynamic display augmentative communication devices

www.enablingdevices.com
www.ablenetinc.com
www.dynavoxtech.com
www.rjcooper.com

16. Communication

An inexpensive solution to try for communication needs is the Proloquo2Go software for use on an iTouch or iPad.
www.proloquo2go.com

17. Sharing and Social Use of Technology

Computers can be used to share experiences, build functional literacy skills and have fun.
PowerPoint presentations can be created with added voice, video clips with sign language, and switch accessibility option.

18. Sharing and Social Use of Technology

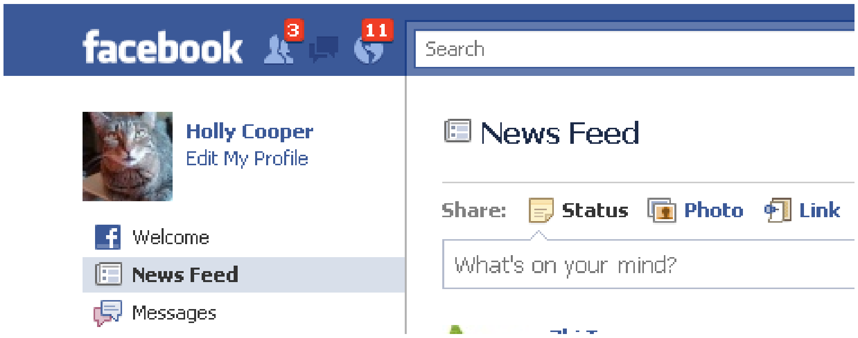
Scrapbooking Software can be an easy and fun way to embellish photos, keep picture journals and share stories.

- My Memories Suite
- www.mymemories.com
- Memory Mixer
- www.memorymixer.com
- Digital Scrapbook Artist
- www.daisytrail.com

19. Sharing and Social Use of Technology

When appropriately supervised, social media can be good ways to support functional use of literacy and age-appropriate social communication

- Facebook
- Text messaging
- Instant messaging via the computer



THE
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CHARGE: Personal Case Study of Post Secondary Transition

Friday, 07/29/11
Breakout Session #2: 1:00-2:15
Panzacola F4

Christopher Sence, Sr. at Sam Houston State University

Kathleen Sheriff, Special Education Specialist, Education Service Center, Region VI, Huntsville, Texas

Presenter Information:

Christopher (Chris) Sence is a 29 year old adult with CHARGE syndrome who is currently a senior at Sam Houston State University majoring in Technical Theatre.

Kathleen Sheriff is Chris' mother and a Special Education Specialist in Texas.

Presentation Abstract:

A Senior student with CHARGE syndrome at Sam Houston State University (SHSU) shares his personal post secondary transition process. Interview video of professors at SHSU discussing accommodations and communication concerns is included in this presentation. His mother shares interventions and strategies used to help him be successful.

CHARGE: Personal Case Study of Post Secondary Transition

Christopher Sence – Sam Houston State University
Kathleen Sheriff – Texas Education Service Center, Region VI

Today's Agenda

- A little about me
- My High School Preparation
- Post Secondary Transitions Process
- My College Life

MY SPECIFIC DISABILITIES

- I HAVE ALL 6 MAJOR SYMPTOMS OF CHARGE (& additional)
- C: 20/400 in left eye and 20/30 in right eye with glasses, with field loss and limited peripheral vision: Optic Nerve Colobomas
- H: Aortic Valve Replacement (2 open heart surgeries): Sub-aortic stenosis
- A: No sinus cavity on right, no sense of smell or taste: I also have an unilateral cleft palate and lip
- **R:** I AM STILL GROWING AT 29
- **G:** JUST BEGINNING PUBERTY
- **E:** PROFOUND NERVE LOSS IN MY LEFT EAR MODERATE LOSS w/Hearing Aid (CONDUCTIVE) IN RIGHT EAR

My Developmental Progression

- FEEDING TUBE AT 5 WEEKS UNTIL 3 ½ YEARS
- HAD OPEN HEART SURGERY AT 6 MONTHS AND BEGAN TO GAIN A LITTLE BIT OF WEIGHT
- GOT MY FIRST HEARING AIDES AT 10 MONTHS; BEGAN SCHOOL WITH CHILDREN THAT HAD INTELLECTELUAL DISABILITIES TO LEARN SIGN LANGUAGE (TWICE A WEEK)

My Developmental Progression

- NEVER CRAWLED – ROLLED AROUND THE HOUSE LIKE A LOG AT 15 MONTHS AND THEN PULLED UP TO MY KNEES WHILE HOLDING ON
- AT AGE 5 I WENT TO A SPECIAL ORAL SPEECH CLASS (NO SIGNING), ONLY SPEAKING AND I BEGAN TO SPEAK SIMPLE WORDS

My Developmental Progression

- STARTED INCLUSIVE KINDERGARTEN AT THE AGE OF 6 YEARS OLD
- I BEGAN TO USE SIMPLE SENTENCE STRUCTURE AT THE AGE OF 7 YEARS OLD

My Developmental Progression

- IN 4TH GRADE I MOVED OUT OF RESOURCE CLASSES INTO REGULAR EDUCATION CLASSROOMS - AT THIS TIME MY I.Q. WAS TESTED AT 104 BECAUSE I COULD UNDERSTAND A LITTLE BETTER.

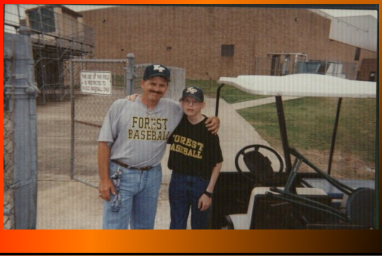
6TH GRADE

MY DEVELOPMENTAL PROGRESSION

- I BEGAN TO UNDERSTAND AND ENJOY MUSIC IN THE 6TH GRADE BY JOINING THE CHURCH CHIOR
- STARTED BEING OCD
- BEGAN MAKING A SOCIAL LIFE IN THE SUMMER BETWEEN MY 7TH AND 8TH GRADE YEARS
- HOME SCHOOLED BETWEEN 8TH AND 9TH USING COMPUTER SOFTWARE AND WORKED ON VOCABULARY AND MATH

My Developmental Progression

- AT AGE 16, I ADVOCATED FOR MY RIGHT TO ATTEND REGULAR HIGH SCHOOL (GO TO A SMALLER PRIVATE SCHOOL).



My Developmental Progression

- GOT MY FIRST JOB AT CHICK FIL A IN THE FALL OF 1999
- LETTERED AS THE BASEBALL MANAGER ALL 4 YEARS IN HIGH SCHOOL (HELPED OUT WITH THE FOOTBALL AND WRESTLING TEAMS AS WELL)
- GRADUATED HIGH SCHOOL IN MAY 2002 WITH A GPA OF 3.33

My Developmental Progression

High School Interventions for Post Secondary School

- Computer Teacher allowed me to use the computer technology to organize my school work and managing the baseball team was a real life learning opportunity
- This is when my OCD really started to kick in and became a good strategy
- My first job - Chick Fil A (6 Years)
- ASL became available for my Foreign Language Credit in 11th grade rather than me taking Spanish
- Church and Camp – Social Activities

High School Interventions for Post Secondary School

- By high school we had dropped a lot of the itinerant services because I would miss too much lecture time.
- Was able to help out in the main office doing mail and taking deliveries to classrooms.
- I was able to put a few things on note cards to use during tests in certain classes.
- I had to be in classes with good behavior management so that I could see and hear

Post Secondary Transition

I Walk the Lonely Road: song by Green Day

BUT WAIT!: College Experiences are about to change the lonely road into a winding way!

May 2002 until Jan 2004 - Worked 3 days a week in a Deaf Ed preschool program as a volunteer para professional … worked roughly 12 hours a week at Chick Fil A … taught 3-5 year olds at Sunday school … walked to the movies …. Took 4 classes at local community college for para professional certificate .Isolated myself to my bedroom and the baseball field at high school on weekends. Also kept scorebooks for Astros Games I watched alone in my room. Got my first cell phone at the age of 20. started playing online spades and met people through internet

Had only one TRUE friend throughout high school and still is to this day my BEST FRIEND EVER!

Post Secondary Transition - Jay

June - 2006 ACCIDENT

Christmas 2003 – Jay visited and told my mom about TSTC (Texas State Technical College) … Mom went with the idea and asked “How easy is it to get into TSTC?” … ROAD TRIP TO WACO TEXAS (3 hours)
January 2004 – Summer 2006 …
Enrolled in TSTC, Computer Technology Program ..
Took remedial writing and math courses
Lived in the dorm next to office … rode a church van to church once in awhile … campus nurse monitored my health, local doctor … ate in cafeteria … small fridge and microwave in dorm room. Internet and cell phone kept me in touch with mom (called every 3 hours) saying “ARE YOU OKAY? WHAT ARE YOU WEARING? WHAT ARE YOU DOING? WHAT ARE EATING THIS VERY SECOND? WHERE ARE YOU LOCATED ON CAMPUS? AND MANY MORE …” in reality parents came every ten days to do shopping and cleaning and tutoring and medical appointments.

Post Secondary Transition - SHSU

- Fall 2006 – Current (2 semesters at Texas Tech University .. Mom went for Doctorate)
- 1st semester lived with my sister for support
- Learning to live in apartment
- SHSU Disability Office (DARS) for accommodations
- Mom tutored – spreadsheets for class schedules and grade point support and medical issues
- I will graduate December 2011 with a BFA in technical theater design
- Mild stroke in 2007,Major abdominal surgery in 2008, and Transient Ischemic Attack in 2011

Post Secondary Transition - SHSU

- David McTier, Ph.D (Dramaturgy and Director)
- Linda Byrd Cook, Ph.D (English)
- Maureen McIntyre, M.F.A (Mrs. Mac) (Directing and Acting)
- Eric Marsh, M.F.A. (Lighting Designer)
- Penelope Hasekoester, M.F.A (Dean of SHSU Theater and Dance)





My Post Secondary Life: Church and Friends

University Heights Baptist Church

“But those who hope in the LORD will renew their strength. They will soar on wings like eagles; they will run and not grow weary, they will walk and not be faint.” - Isaiah 40:31 (NIV)

Post Secondary Life: Lions Club and Camp Counselor

Post Secondary Life: Advocacy (DBYAA and AER)

AER Little Rock

DBYAA

DBYAA Senator Markey

Texas Represenative Micheal T. McCaul

White House Visit ADA Signing
President Barak Obama

Post Secondary Life: Crystal ☺

CONTACT INFO

- If you have questions or would like a copy of this presentation please feel free to contact me
- Cell phone – 936 662 8702
- Email – css006@shsu.edu
- FACEBOOK - CHRISTOPHER SENCE or SHSU Email

Sammy the Bearkat & me

SHSU Bling

THE CHARGE SYNDROME FOUNDATION

CHARGE 102: Sensory Deficits and Communication

Friday, 07/29/11
Breakout Session #2: 1:00-2:15
Panzacola G1

Sandra Davenport, MD
Genetics and Sensory Development
Bloomington MN and Saint Louis University

Rob Last
CHARGE Syndrome Association of Australia

Presenter Information:

Sandra Davenport is a Pediatric Geneticist and Developmental Pediatrician who specializes in sensory losses -- conditions which cause combined vision and hearing loss. She has been involved with the Minnesota Deafblind Project since its inception. As a founding member of the Foundation, she has been instrumental in guiding research in areas of development, communication and education in CHARGE syndrome. Dr. Davenport is a member of the team starting the first CHARGE Syndrome Clinic at Cardinal Glennon Children's Medical Center at Saint Louis University.

Rob Last is an Early Childhood Educator who has worked with the Royal Institute for Deaf and Blind Children in New South Wales, Australia and Vision Australia. He has specialized in working with children with sensory deficits in particular hearing and vision loss. He has developed communication systems to meet individual needs of children with CHARGE since the mid 1980's. His focus has been home-based early intervention with families. Rob has been involved with every Australasian CHARGE conference.

Presentation Abstract:

Dr. Davenport will spend 20 minutes talking about the sensory deficits in children with CHARGE syndrome and how they affect development: How the combined features of hearing loss, vision loss, poor balance, decreased sense of smell, altered pain threshold and multiple hospitalizations and surgeries affect the developing baby and child. The importance of early assessment and early intervention in these areas will be presented.

Rob Last will spend his 20 minutes talking about how to begin establishing a communication system with your child: what is needed in terms of assessment, resources and environment. He will also be presenting 'Children with CHARGE – A Longitudinal Study 1993 – 2010', discussing and showing excerpts from his DVD of 10 young people with CHARGE in 1993, 2000 and 2007 – focusing on their progress. There will be time for questions; you can also leave written questions for the 103 session

Sensory Loss: How does that Change Early Development?

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

952-831-5522
slhdaven@umn.edu

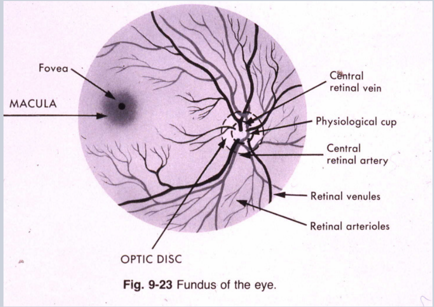
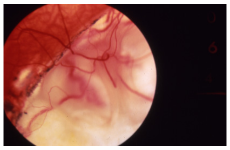
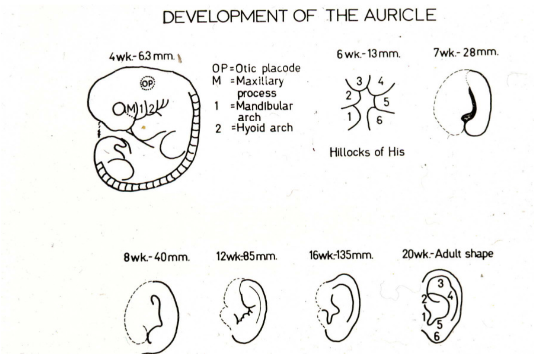


Fig. 9-23 Fundus of the eye.

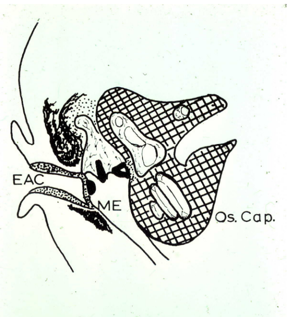
Coloboma – cleft of the eye



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

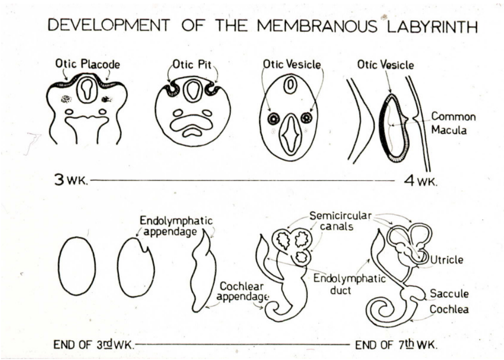


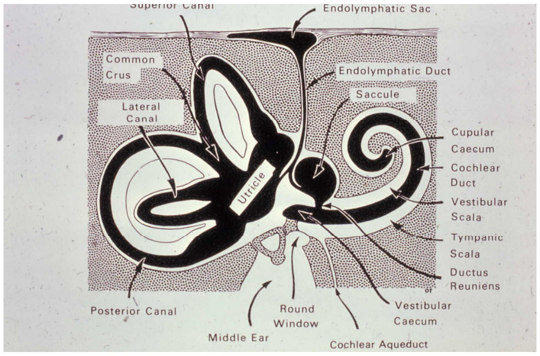
Characteristic CHARGE Ear



20 Weeks

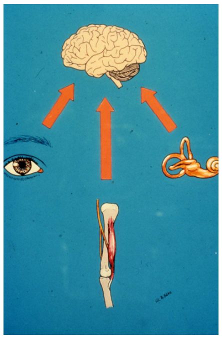
FIG. 39–37. Development of the external and middle ear in relationship to the labyrinth (diagrammatic). *E.A.C.*, external auditory canal; *M.E.*, middle ear; *Os.Cap.*, osseous capsule.





Components of Balance

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



Communication Bubble

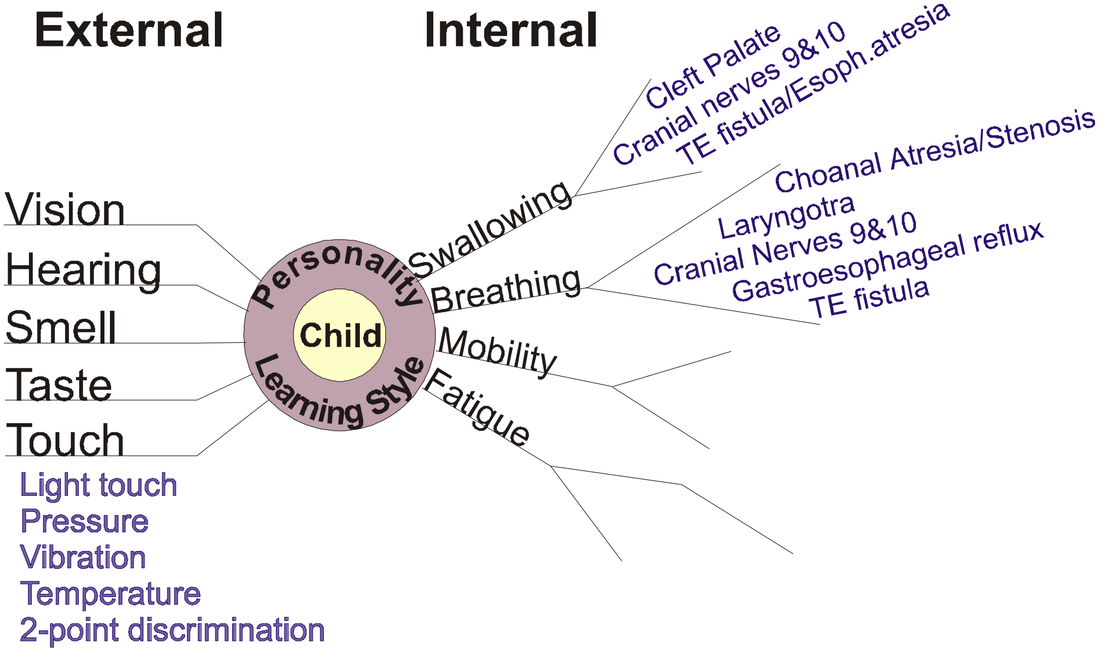
- The area within which the individual can
  - See
  - Hear
  - Smell
- Touch and taste do not give information at a distance
- Anything outside the bubble is “off the radar”

Kids with CHARGE may be “Hard of”

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

CHARGE 102: PHYSICAL INFLUENCES ON DEVELOPMENT IN CHARGE

Sandra L.H. Davenport, M.D.
SensoryGenetic/Neuro-development,
5801 Southwood Drive, Bloomington MN 55437-1739
952-831-5522 slhdaven@umn.edu



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

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

A more complete discussion of this is in the Manual

INFLUENCE OF SENSORY LOSS ON DEVELOPMENT:
The Communication Bubble

Are all five major senses working?

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

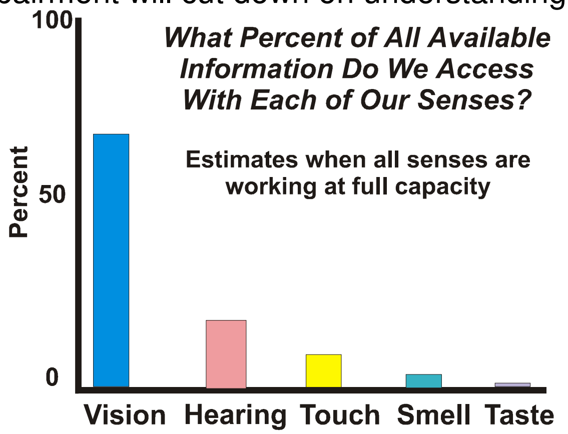
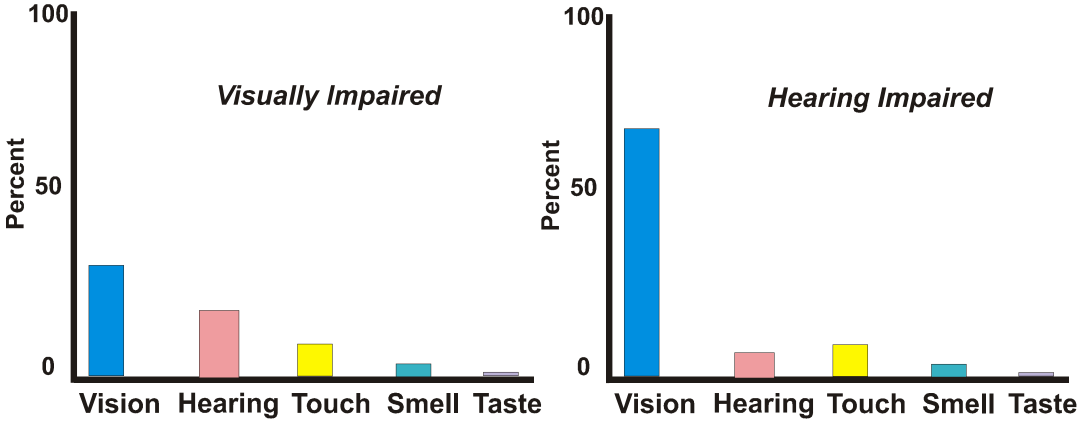


Figure 1: Full access to information from all senses.

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



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

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

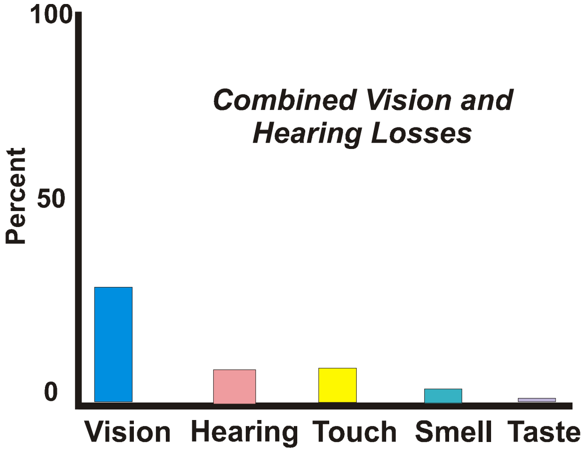


Figure 4: Combined vision/hearing loss with half of each gone.

What is Deaf-Blind?

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

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

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

What combination of vision & hearing is present?

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

The Need to Establish A Communication Bubble

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

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

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

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

Setting The Scene
Babies and Toddlers

Rob Last

CHARGE Syndrome Association of Australasia

PO Box 404
Somers Vic 3927
Australia
email: roblast@iinet.net.au

Early Considerations

Educators and therapists may assist in:

- Positioning of the crib, people and toys
- Contrast
- Lighting
- Provision of communicative cues
- Alerting to events
- Early literacy

> "For children who have CHARGE syndrome there is strong evidence that diagnosis and intervention before six months of age significantly improves outcomes in communication, behavior and learning."
>
> Jan van Dijk, Arno de Kort

Early Communication

- Contentment
- Distress
- Wakeful
- Sleepy
- Pain
- Comfort
- Hunger
- The first smile

The Early Months

- Medical issues take precedence
- A large team involved
- Life and death issues dominate
- Outcomes are unknown
- Major surgery's take place
- Bonding between child and parents is interrupted
- Medical procedures may occur without warning or preparation

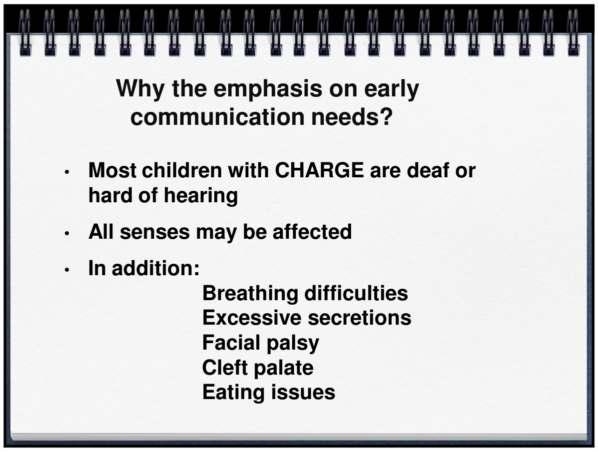
Speech/Communication

Communication includes:

- body language
- facial expression
- touch cues
- gestures
- situational cues

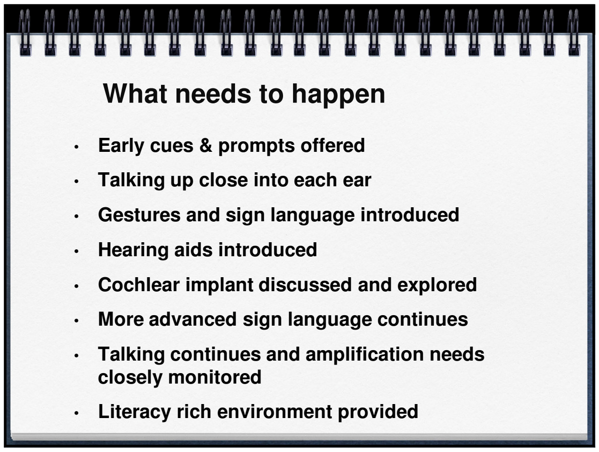
and

- speech



Why the emphasis on early communication needs?

- Most children with CHARGE are deaf or hard of hearing
- All senses may be affected
- In addition:
  - Breathing difficulties
  - Excessive secretions
  - Facial palsy
  - Cleft palate
  - Eating issues

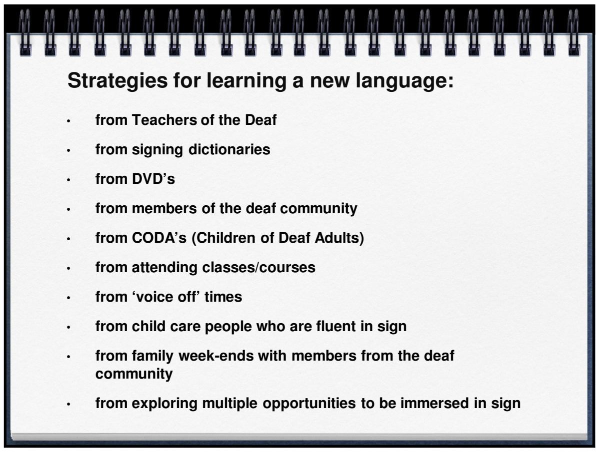


What needs to happen

- Early cues & prompts offered
- Talking up close into each ear
- Gestures and sign language introduced
- Hearing aids introduced
- Cochlear implant discussed and explored
- More advanced sign language continues
- Talking continues and amplification needs closely monitored
- Literacy rich environment provided

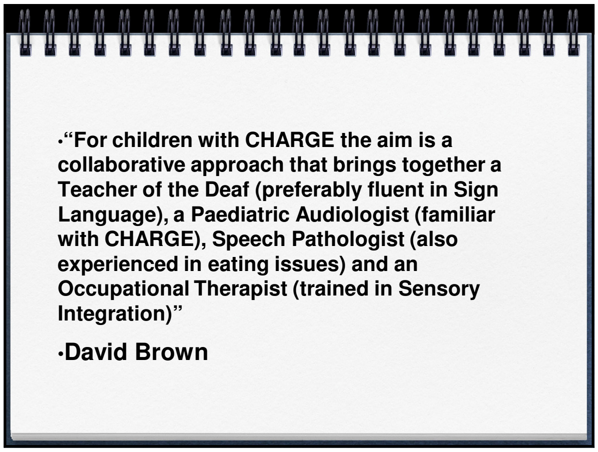
Communication Embraces

- Sign language - the sign language of the country where the child lives
- Speech with hearing aids
- Speech with a cochlear implant
- Speech assisted by BAHA (Bone Anchored Hearing Aid)
- Visual programs
- Signed English
- Reading and writing



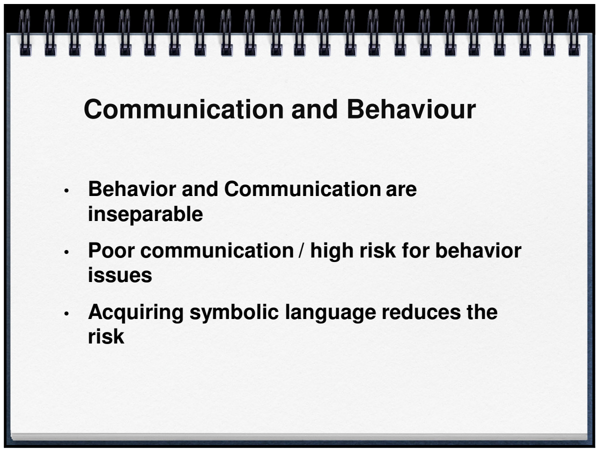
Strategies for learning a new language:

- from Teachers of the Deaf
- from signing dictionaries
- from DVD's
- from members of the deaf community
- from CODA's (Children of Deaf Adults)
- from attending classes/courses
- from 'voice off' times
- from child care people who are fluent in sign
- from family week-ends with members from the deaf community
- from exploring multiple opportunities to be immersed in sign



•"For children with CHARGE the aim is a collaborative approach that brings together a Teacher of the Deaf (preferably fluent in Sign Language), a Paediatric Audiologist (familiar with CHARGE), Speech Pathologist (also experienced in eating issues) and an Occupational Therapist (trained in Sensory Integration)"

•David Brown



Communication and Behaviour

- Behavior and Communication are inseparable
- Poor communication / high risk for behavior issues
- Acquiring symbolic language reduces the risk

Communication and Gross Motor Development

"Through parent surveys it was found that children with CHARGE achieve gross motor developmental milestones but do so at a slower rate than unaffected children. A correlation between gross motor development and communication abilities was also found."

James Thelin and Lori Travis

What is considered best practice:

- for parents, extended family and professionals to embrace bilingualism
- for the best amplification to be provided
- to provide an environment of visual language and amplified speech

What then is the aim?

- To provide a range of communications
- To ensure the communication is meaningful
- To provide an environment of immersion in a meaningful language
- To aim for communication competence which provides the opportunity and ability to express thoughts, ideas, emotions and humor i.e. symbolic language

Outcomes

"We have become a bilingual family. Sean is able to access both oral and signed communication. Both modes of communication are essential to his overall acquisition and use of language."

Madelene Rich, Mother of Sean who has CHARGE

"All who are in contact with persons with CHARGE need to adapt to each child's communicative development. They need to help establish the most favorable social contexts for making the most of their communication competencies to enrich their social lives."

Jacques Souriau, et al

'Children with CHARGE Seven Years Later'

The DVD

- First filmed in 1993
- Follow up in 2000
- Further follow up in 2007

Available free from Rob Last

email: roblast@iinet.net.au

THE CHARGE SYNDROME FOUNDATION

Problems with Self-Regulation and Behavior in CHARGE

Friday, 07/29/11
Breakout Session #2: 1:00-2:15
Panzacola G2

Tim Hartshorne, Ph.D.
Central Michigan University

Presenter Information:

Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. His particular interest is in understanding the challenging behavior exhibited by many individuals with CHARGE. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project.

Presentation Abstract:

The proposed CHARGE behavioral phenotype includes problems with self-regulation. This presentation explores the nature of self-regulation, its role in the behavioral challenges found with CHARGE, problems with the regulation of learning, behavior, emotions, and sensations, and how parents and teachers might use scaffolding to assist individuals with CHARGE to learn to self-regulate.

10th International CHARGE Syndrome Conference
Rosen Shingle Creek Resort, Orlando, FL, July 28-31, 2011

Problems of self-regulation in the behavior of individuals with CHARGE syndrome

Tim Hartshorne

Central Michigan University

Two ends of the spectrum

- Totally regulated
- Completely dysregulated

Self-regulation Scale

I have a hard time paying attention and my mind tends to wander.

When I really need to pay attention I can focus my mind.

I can readily prioritize the things I need to get done in a day.

I become overwhelmed when faced with too many things to take care of.

I get upset a lot and cannot find any way to get rid of those feelings.

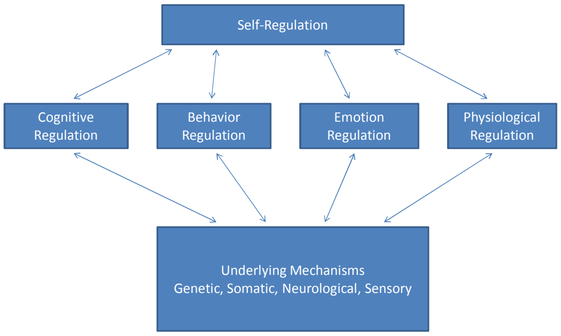
When I really need to control my feelings I can do it.

When there is nothing going on I have to create it.

When I am in a noisy crowd I have to find a way to leave.

Self-Regulation

- Managing the threshold of arousal
- Processes of self-control
- Both suppresses and encourages; inhibits and promotes
- Supports homeostasis of the system
- Critical to development



Self-Regulation

Cognitive Regulation

Behavior Regulation

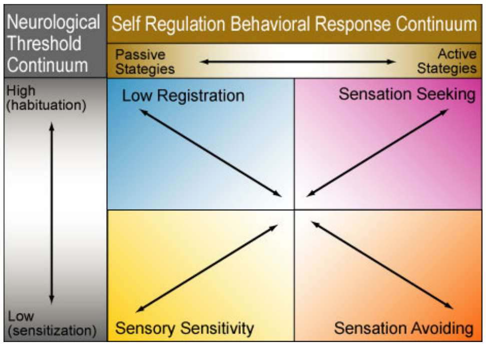
Emotion Regulation

Physiological Regulation

Underlying Mechanisms
Genetic, Somatic, Neurological, Sensory

Jude Nicholas and Tim Hartshorne, 2009

Dunn Conceptual Model



The extremes

- If a system cannot self-regulate, we have to provide external systems of regulation
- Too much regulation can stifle innovation
- Too little regulation can lead to chaos and abuse

Diagnoses in CHARGE

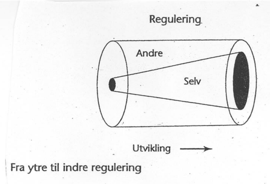
- OCD – a way to reduce stimulation and exercise control
- ADHD – a problem with regulating sensory stimulation and focusing on a problem
- Tic disorder – a stress response to lack of control over environment
- Autistic-like behavior – the failure of regulation strategies

Scaffolding

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

Scaffolding for self-regulation

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

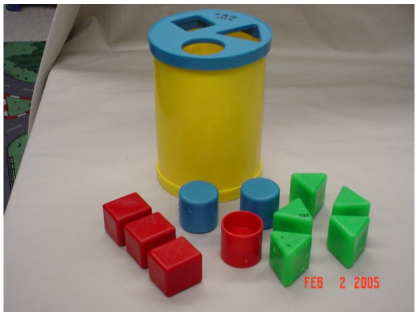


Fra ytre til indre regulering

Components of Scaffolding

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

The Shape Sorter



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

The four areas of self-regulation

- Define each area of self-regulation
- What is involved?
- Describe scaffolding strategies

Cognitive Regulation

- Motivated to think about a problem
- Being precise and accurate
- Comparing alternative choices
- Adapting prior learning to the problem

How learning changes

- Concrete reasoning
  - Objects and events available to the senses
- Rote learning and memorization
  - Alphabet
  - Multiplication table
  - Names of things
- Abstract reasoning
  - Ideas or concepts with no physical referents

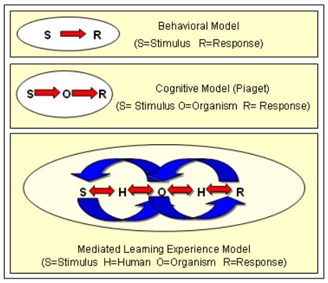
Executive Function

- Initiate – goal, planning, getting started
- Sustain – staying on task, moving toward goal
- Inhibit – avoiding getting side tracked
- Shift – changing directions when needed

These functions continue to develop into early adulthood and can be improved.

Cognitive Scaffolding

- Mediated Learning Experience



Example

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

- Motivated to think about a problem
- Being precise and accurate
- Comparing alternative choices
- Adapting prior learning to the problem

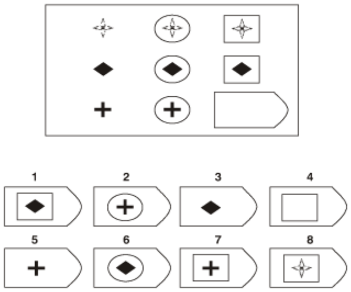


Figure 2 - Raven's progressive matrices

Behavior Regulation

- What is the purpose of the behavior?
- Is it consciously planned and intentional?
- Well regulated behavior is both intentional and goal directed.

The Self-Regulation of Behavior

- Too often we tell children what we do not want them to do, and not what we very much do want them to do

- Strategies for building self-regulation
  - Offering choices (shared control)
  - Rehearsing behavior options
  - Building communication
  - Delay of reinforcement
  - Embedding a positive context

Behavior Scaffolding

- Supporting what we want the child to do
- Positive Behavioral Supports
  - How does the social environment support positive behavior?
  - How does the physical environment support positive behavior?
  - What skills does the child possess for positive behavior?

Social Environment

- Social embeddedness
- Social skills
- Negative relational schemas
- Circle of Friends

Physical Environment

- Supporting what we want the child to do
- Responsive to sensory needs of the child
- Responsive to physical limitations
- Reducing complexity

Child’s Behavior

- Supporting what we want the child to do
- Reading behavior as communication
- Understanding the purpose of behavior
- Functional Communication Training

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

Emotion Regulation

- What a person does to manage his or her emotional states
    - Regulate both negative and positive emotions
    - Decrease emotions or increase emotions
    - May be conscious or unconscious
    - May be internal or external
    - Are generally goal directed

Learning to regulate emotions

- “She didn’t know what to do with her emotions”
    - Emotional expression
      (What does it look like to be angry, sad, etc.?)
    - Emotional intensity
      (How worried, sad or mad would you feel in this situation?”)
    - Emotional self-efficacy
      (How could you make yourself feel better in this situation?)

Emotion Scaffolding

- Social referencing
- Attachment
- Talking about how you feel
- Soothing
- Positive face to face play
- Distraction
- Problem-solving
- Altering interpretations
- Suggesting better ways to respond
- Creating daily routines that make emotional demands predictable and manageable

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

Physiological Regulation

- Sensory
- Pain
- Fatigue
- Eating
- Sleeping
- Respiratory/Digestive/Temperature/Other systems

The Self-Regulation of Physical States

- Relaxation
- Tuning in to our bodies
- Bio-feedback – being aware of control
- Management of arousal
    - Timeout
    - Sensory room

Physiological Scaffolding

- Developmental Care
- Sensory Integration
- Physical responses
    - Hug
    - Squeeze
    - Touch
    - Rock
    - Tickle

Fun Chi

- Reduced stress
- Reduced anxiety
- Reduced depression
- Increased self-esteem
- Increased energy/focus/concentration
- Increased positive mood
- Better balance
- Improved sleep
- Improved immune system

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

Summary

- Children with CHARGE often have poorly regulated systems
- They will do better socially and academically if they can learn to self-regulate
- They can only develop self-regulation skills slowly unless they experience a lot of scaffolding from the adults in their lives

Thanks to my Lab

- Tasha Nacarato
- Maria Ramirez
- Rachel Vert
- Stephanie Budde
- Valerie Webber
- Kasee Stratton

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THE
CHARGE
SYNDROME
FOUNDATION

A Disturbance in the Force: Sensory processing differences and what they mean in everyday life

Friday, 07/29/11
Breakout Session #2: 1:00-2:15
Wekiwa 3 & 4

Kate Beals, OTR/L
Registered Occupational Therapist,
Technical Assistance Specialist, South Carolina Interagency Deaf-Blind Project

Presenter Information:

Kate Beals is a registered occupational therapist with 14 years of experience working with children who have multiple disabilities, and 21 years of experience being the mother of a son who has autism. Kate has spent the past 9 years working at the South Carolina School for the Deaf and the Blind, and the last 2 and half of those years as the Technical Assistance Specialist for the South Carolina Interagency Deaf-Blind Project. Because Kate has both personal and clinical experience addressing sensory processing and integration issues, she brings a unique perspective to the discussion of her favorite topic.

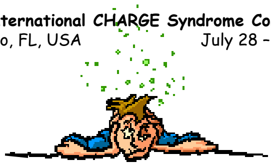
Presentation Abstract:

Individuals with CHARGE syndrome often have differences in the way they receive, process, and integrate information from the seven sensory systems. Children with CHARGE syndrome may not experience the world through their senses in the same way as their parents, family members and friends. Becoming aware of the differences in sensory processing (input) may help families achieve a better understanding of some of the behaviors (output) that occur when a child has CHARGE syndrome.

A Disturbance in The Force:

Sensory Processing Differences
and what they mean
in everyday life

10th International CHARGE Syndrome Conference
Orlando, FL, USA July 28 – 31, 2011



Kate Beals, OTR/L
South Carolina Interagency Deaf-Blind Project

Session Objectives

1. Understand the relationship between sensory input and motor output.
2. Know the names, structures and functions of the seven sensory systems.
3. Be aware of sensory differences, including structural, likely to be associated with CHARGE Syndrome in general, and their impact on function and behavior.
4. Identify resources for additional information and therapeutic intervention if needed.

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

1. Visual – what we see
2. Auditory – what we hear
3. Gustatory – what we taste
4. Olfactory – what we smell
5. Light touch – what we feel on our skin (protective touch)
6. Proprioception – what we feel in our joints and muscles (body position)
7. Vestibular – movement and position of the head in space

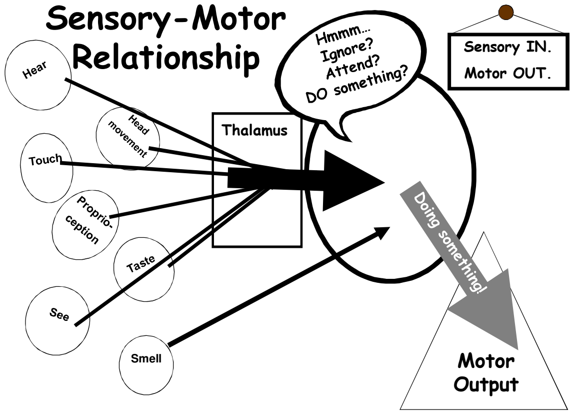
Have you ever said this?

Hey, quiet down back there so I can see where I'm going!

Sensory Integration

Brain takes in information from all 7 systems and decides what is relevant, irrelevant, and whether an active motor response is called for. We all have times when we need to "turn off" one sensory channel so we can attend better to a different one.

Sensory-Motor Relationship



Sensory IN.
Motor OUT.

Why do Sensory Differences in CHARGE Syndrome matter?

They matter because…

- Studies of the process of human development reveal that a certain amount of stimulation (meaning **sensory** stimulation) is necessary for the brain to grow and develop.
- "If 2 or more **(sensory)** systems are functioning below normally expected levels…then the baby usually experiences delays in adaptation to the environment." (Human Development textbook)
- Children with CHARGE Syndrome are likely to have impairment of not only 2 out of 7, but 6 out of 7, or even 7 out of 7 sensory systems.

Sensory IN.
Motor OUT.

If the sensory information going into the brain is absent, insufficient, inaccurate or distorted…

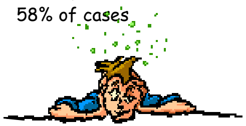
…what do you suppose happens to the motor output?

Cranial Nerve Involvement

Cranial nerve anomalies are very frequent in CHARGE.

- 12 cranial nerves
- Begin in the brain
- Extend to structures in the head and neck
- Both motor control and sensation
- Nerves involved in the senses of smell, taste, hearing, and vision (sensory)
- Movements of the eyes, face, tongue, palate and swallowing (motor)

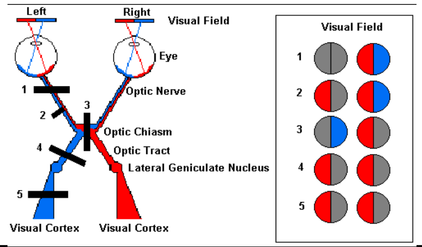
- Some cranial nerves are involved in control of heart rate and movements of the gastrointestinal tract.
- **At least 1 cranial nerve** is involved in 75% of CHARGE cases
- **2 or more** are involved in 58% of cases



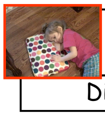
Bruce R. Korf, MD, PhD; *CHARGE Syndrome Manual*

Visual System - 1

The Visual System is very complex!



Receptors called **rods** respond to movement.
Receptors called **cones** respond to color and detail.
Pathway: Optic nerve **Function:** Seeing



Vision in CHARGE

Differences

- Coloboma: 80 – 90%
  - iris, retina, optic nerve
- Field loss – usually upper
- Possible microphthalmia
- Acuity deficits – 90%
- Light sensitivity
- Strabismus or ambliopia
- Sometimes CVI
- Facial Palsy – 40%
  - Lack of blinking causes dry cornea, leading to corneal scarring

What it means

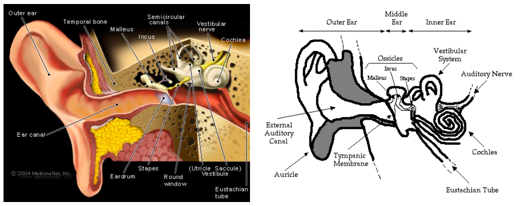
- May need a "quiet" visual environment
- Present materials in best (usually lower) visual field for access
- Seeing is HARD WORK:
  - might need supported body position or to "turn off" other senses
- Contributes to problems with **Balance**

Roberta Pagon, MD, CHARGE Syndrome Manual

“Look at the camera.”

Photo courtesy of California Deaf-Blind Services

Auditory System - 2



Sound waves enter the ear, the eardrum vibrates which moves the three tiny bones in the middle ear. These connect via the oval window to the inner ear, where the hair cells in the cochlea transform the waves into electrical impulses the brain can perceive and understand.

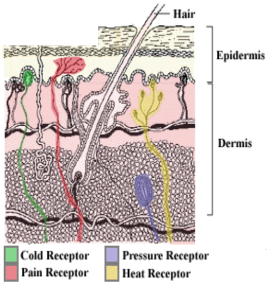
Pathway: Vestibulocochlear Nerve. **Function:** Hearing

Hearing in CHARGE

Differences	What it means
Structural differences of inner ear	May need to explore alternate forms of receptive and expressive communication
Cranial nerve VIII, “Auditory Nerve” involvement	May need to explore supports for auditory function such as cochlear implant, hearing aids, FM system, etc.
Significant hearing impairment is common	May not be able to use hearing for echolocation during travel

*NOTE Cranial nerve VIII is also called the “Vestibulocochlear nerve”.

Tactile System - 3



There are many different types of sensory receptors in the skin, specialized for pain, heat, cold, light touch, firm touch (pressure), static touch and moving touch.

Pathway:

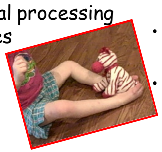
- Pain,temperature, nondiscriminative (crude) touch, pressure, and some proprioceptive sensation travel on the Anterolateral (ALS) system.
- Discriminative (fine) touch, vibratory sense, position sense travel on the Dorsal Column– Medial Lemniscal (DCML) system.

Two functions of touch:

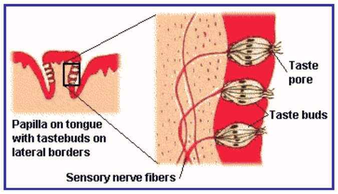
Protection and discrimination.

Tactile in CHARGE

Differences	What it means
Possible increased **or** decreased tactile awareness	May use feet to explore
Tactile defensiveness due to early frequent hospitalizations, surgeries, and/or neurological processing differences	May dislike unexpected touch, other kids getting too close, being touched around face or trach
	May benefit from Wilbarger therapeutic brushing protocol
	Don’t force tactile exploration
	Hand-UNDER-hand technique is recommended

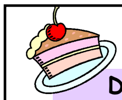


Gustatory System - 4



We have almost 10,000 taste buds inside our mouths. A single taste bud contains 50–100 taste cells representing all 5 taste sensations: salty, sour, sweet, bitter, and umami (savoriness).

Pathway: Facial, Glossopharyngeal and Vagus Nerves (Cranial Nerves VII, IX and X).



Taste in CHARGE

Differences

Three of the four cranial nerves most likely to be involved in CHARGE Syndrome affect taste:

VII. Facial:
- controls most facial expressions
- secretion of tears & saliva
- **taste**

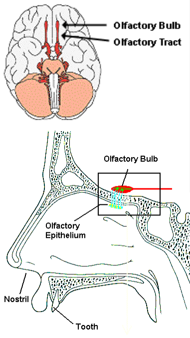
IX. Glossopharyngeal:
- **taste**
- senses carotid blood pressure

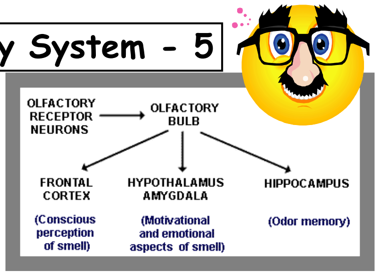
X. Vagus:
- senses aortic blood pressure
- slows heart rate
- stimulates digestive organs
- **taste**

What it means

- **Good taste** is not a motivator
- **Bad taste** is not a deterrent
- Limited exposure to taste due to g-tube feedings
- Oral defensiveness is likely due to early g-tube feedings (not used to food textures in mouth)

Olfactory System - 5





OLFACTORY RECEPTOR NEURONS → OLFACTORY BULB

FRONTAL CORTEX (Conscious perception of smell)

HYPOTHALAMUS AMYGDALA (Motivational and emotional aspects of smell)

HIPPOCAMPUS (Odor memory)

Pathway: Unlike the other senses, olfactory information goes straight to the frontal cortex of the brain without going through the thalamus first. It travels on the Olfactory Nerve (Cranial Nerve I).

Smell in CHARGE

Differences

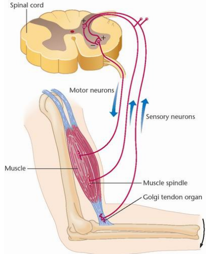
- Anosmia (inability to smell) is a frequent feature of CHARGE Syndrome
- Structural differences – many are missing olfactory bulbs and stems
- Nasal blockages are common (bilateral choanal atresia)
- Cranial Nerve I (Olfactory) involved in approx. 40%
- Study showed 100% correspondence between anosmia and problems with onset of puberty

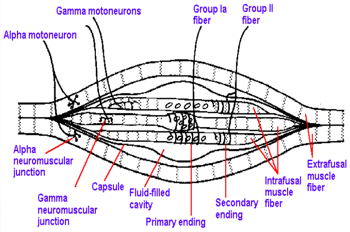
What it means

- What we think of as a "bad" smell may not be aversive
- Safety – can't smell smoke from a fire, gas leak, spoiled food
- Social – unaware of body odors
- Smell is powerful - linked to memory, learning, and bonding – reduced access

Jorieke Bergman, MD, and
Conny von Ravenswaaij-Arts, MD, PhD

Proprioceptive System - 6





Proprioception is awareness of body position. Receptors are in the muscles, joints, and tendons. Receptors react to stretch and compression. It is an unconscious feedback system between the body and the brain. **Pathway:** Spinocerebellar Tract. **Function:** Body position, graded movement (how much force?), awareness of gravity.

Proprioception in CHARGE

Deep pressure; discriminative touch; position of the BODY in space; awareness of gravity; body awareness

Differences

- Little research, but behavior and motor patterns suggest diminished proprioceptive awareness.

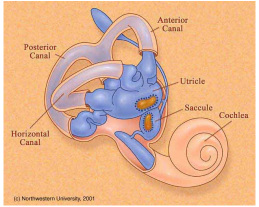
What it means

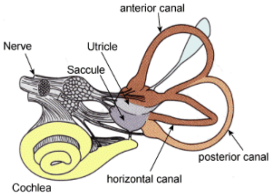
- Movement patterns resemble those of a person with reduced proprioception (video)
- Late to walk
- Needs extra time to learn new motor routines
- Contributes to problems with **Balance**

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*

Vestibular System - 7





The Vestibular System responds to changes in gravity and acceleration of the head (up/down, forward/back, side to side). Fluid inside the canals sloshes against hair cells, causing them to bend. The degree and direction of the bend translates to position of the head in space. Semicircular canals sense rotation. Utricle and saccule sense up and down.

Pathway: Vestibulocochlear Nerve. **Function:** Sense of Balance

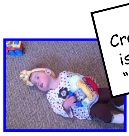
---

Vestibular in CHARGE

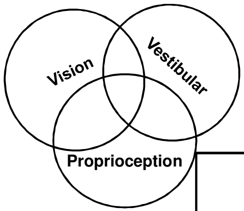
Movement; position of the head in space

Differences	What it means
ABSENCE OR UNDERDEVELOPMENT OF SEMI-CIRCULAR CANALS Cranial nerve VIII* ("Vestibulocochlear") may be absent or displaced	Difficult to keep head upright against gravity Will often prop head in hand Can't process rotation – the stimulation from swinging or going in circles with the walker is probably more visual than vestibular Contributes to problems with **Balance**

*NOTE Cranial nerve VIII is also called the "Auditory" nerve.



Balance



The **visual**, **vestibular**, and **proprioceptive** systems work together to enable us to achieve upright balance and to maintain it as we move through space. Postural control relies primarily on these three systems functioning well TOGETHER.

Balance in CHARGE

Vision Prop.

- Vestibular: little or none
- Vision: impaired
- Proprioception: impaired

Vest.

Walking occurs against all odds!

- Walking emerges later than for typically developing children.
- It takes a lot of energy to stay upright, so many children with CHARGE Syndrome need some literal "down time" lying down on their backs.

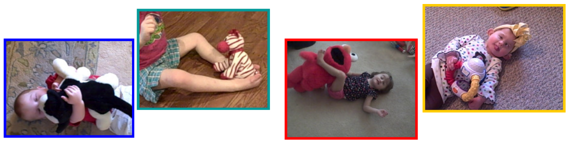
Does this sound like someone you know?

In the spring 2003 issue of the *California Deaf-Blind Services Newsletter,* David Brown states:

"Children with CHARGE are likely to be among the most truly multi-sensory impaired people you will ever meet, having difficulties not just with vision and hearing, but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell."

What does it mean for the child?

- The child with CHARGE experiences the world differently from the way others do.
- He faces ENORMOUS challenges in perceiving (taking in and "making sense" of sensory inputs) and responding to (motor output) just about every Life Experience he encounters.



What does it mean for us?

- We, as parents, educators and service providers, face an enormous challenge as well.
- To help the child develop the **skills** (which are a kind of **"motor output"**) he or she needs to live the most meaningful, productive, enjoyable life possible, we must first **recognize and respect** those differences in **"sensory input"** that define the child's experience of Self and Self in relation to the environment.

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Sensory system graphics are from a number of online sources.

What's Next?

How will
increased awareness
of the unique and complex
sensory issues in
CHARGE Syndrome
influence our expectations
about behavior,
and the types of
interventions we employ?

Self-Portrait by Andrew Hippenstiel

Let's wrap it up.

SOUTH CAROLINA INTERAGENCY DEAF-BLIND PROJECT

Evaluations, please.

Thank you for coming!

THE CHARGE SYNDROME FOUNDATION

Understanding My Team. The Roles and Responsibilities of the Team Members

Friday, 07/29/11
Breakout Session #2: 1:00-2:15
Wekiwa 5

Gloria Rodriguez-Gil
Educational Specialist
California Deaf-blind Services

Presenter Information:

Gloria Rodriguez-Gil has been an educational specialist for California Deaf-Blind Services for the last eleven years, and has worked in the field of visual impairments, multiple disabilities and deaf-blindness for close to twenty. She provides technical assistance and training for educational teams, and she has written several articles and blogs on ways the educational team can better support students who are deaf-blind in the classroom setting. You can read them at www.cadbs.org

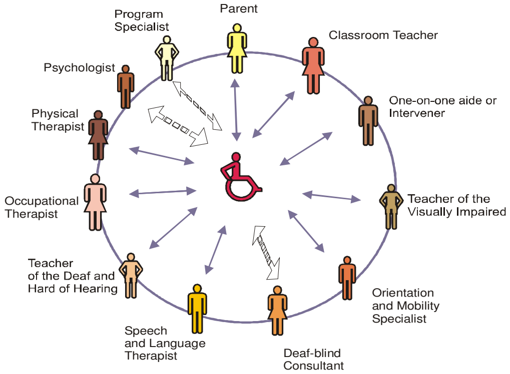
Presentation Abstract:

Understanding the roles and responsibilities of the team members is critical for good collaboration because parents can know what to expect and ask from each team member. This session will describe the collective roles and responsibilities of all the team members and of each member individually. These include: parent, classroom teacher, the intervener or one-to-one aide, teacher for students who are visually impaired, teacher for students who are deaf and hard of hearing, the orientation and mobility specialist, the deaf-blind consultant, speech and language therapist, occupational therapist, physical therapist, and school psychologist.

Understanding My Team.
The Roles and Responsibilities of the Team Members.

Gloria Rodriguez-Gil
California Deaf-Blind Services
10th International CHARGE Syndrome Conference
Orlando, Florida

The Educational Team



Parent
Classroom Teacher
One-on-one aide or Intervener
Teacher of the Visually Impaired
Orientation and Mobility Specialist
Deaf-blind Consultant
Speech and Language Therapist
Teacher of the Deaf and Hard of Hearing
Occupational Therapist
Physical Therapist
Psychologist
Program Specialist

> ***"They all have an expertise—but no team member can work autonomously of the others."***
>
> **Terry Boisot, parent**

Parent

- Is the driving force, one of the leaders of the team
- Holds the big picture, consistent team member
- Is a unique source of information: on the child, on CHARGE Syndrome
- Shares information and helps develop goals the child will accomplish during the school year
- Advocates strongly but also needs to be very sensitive to educators needs and fears
- Promotes team collaboration

Program Specialist

- Responsible to manage individual cases
- Generally has basic knowledge of the child's disabilities
- Attends educational decision-making meetings, where services for the student are discussed

Program Specialist

- Supports the educational team work to better serve the student
- Develops the IEP document fully describing the services that will be delivered and how services these will be implemented
- Provides the student with an educational program that not only addresses academic achievement but social-emotional

Classroom Teacher

- Person in charge in the classroom
- Must work in collaboration with the IEP team
- Must collaborate with team members to adapt the classroom environment, activities, materials to be accessible to the student

Classroom Teacher

- Must promote socialization amongst classmates and schoolmates
- Ideally must work directly with the child

One-to-one Assistant/ Intervener

- Deaf-blindness is a disability of access to visual and auditory information
- Individuals with deaf-blindness need environmental access, communication and social-emotional support
- The role of an intervener is to facilitate this process

One-to-one Assistant/ Intervener

- The intervener is a paraprofessional with abilities and specialized training to provide direct support to a deaf-blind individual
- The decision of having an intervener is based upon the level of support needed by the student to access the information
- The intervener is the bridge/connection between the individual with deaf-blindness and the world

Teacher of the Visually Impaired

- Performs functional visual assessment
- Refers the student to a low vision clinic for evaluation
- Evaluates and supports with visual aids
- Determines which visual materials are appropriate for the student. (i.e., size, color, contrast)
- Adapts written materials, (i.e., large print, Braille)

Teacher of the Visually Impaired

- Evaluates and modifies visual environment
- Provides Braille instruction
- Many VI Teachers have limited experience working with deaf or hard of hearing or students with multiple disabilities.

Orientation and Mobility Specialist

- Helps a student who is blind or with low vision know where he is in space and where he wants to go (orientation)
- Helps a student who is blind or with low vision to have a plan to get there (mobility)
- This specialist has experience traveling in space blindfolded or with low vision goggles

Orientation and Mobility Specialist

- Teaches student the concepts of space, environment and how to use the information received through other senses (sound, temperature, vibration) to maintain or re-direct orientation
- Teaches the student how to use the white cane and dog guide as support or as the only instrument of guidance

Orientation and Mobility Specialist

- Teaches the student how to use residual vision and low vision aids when traveling
- Helps the student to learn about his environment and the language that the student needs to use in that environment

Orientation and Mobility Specialist

- Develops adaptations that will allow the student to interact with the public (e.g., communication cards in public transportation)

Deaf-Blind Specialist (CA)

- Provides statewide and regional trainings to IEP teams
- Provides technical assistance in person, via telephone, Internet and video
- Provides urgent technical assistance
- Helps create and participate in a family-to-family support network
- Disseminates information, resources and products

Speech-Language Therapist

- Identify student’s speech problems
- Evaluates students with speech problems
- Refers students to medical professional for further evaluation
- Provides speech-language services to improve or prevent communication issues

Speech-Language Therapist

- Provides counseling and guidance to parents, children and teachers in the area of speech-language
- Some therapists know about other modes of communication such as Sign Language and augmentative and alternative communication

Deaf and Hard of Hearing Teacher

- Observes the classroom environment to see what can be done to improve the student's learning
- Checks for properly functioning hearing aids, FM systems, cochlear implants
- Determines with the educational team the best way to communicate with the student (speech, volume, distance, Sign Language, object cues, etc.)

Deaf and Hard of Hearing Teacher

- Knows about formal language development in hard of hearing people (spoken and written, comprehension and use)
- Many of these professionals have limited experience working with students who are blind or with low vision or have multiple disabilities and the information given by other team members is important.

Occupational Therapist

- Works with fine motor abilities, coordination and strength to improve mobility and physical abilities
- Helps the student to work with available senses
- Works on feeding issues, hand use (manipulatives) and physical independence

Occupational Therapist

- Knows about adapted equipment that can help the student be as independent as possible
- Creates a "sensory diet" (coined by OT Patricia Wilbarger) which is a carefully designed, personalized activity plan that provides the sensory input a person needs to stay focused and organized throughout the day.

Physical Therapist

- This service generally works on posture, muscle strengthening, mobility and endurance
- Works on gross motor abilities
- Provides experiences and practice to improve motor development.
- Monitors the appropriate use of prescribed durable equipment

Psychological Services

- Performs evaluations/assessments
- Interprets evaluation/assessment results
- Provides consultation to educational team members, to plan educational programs that meet the unique needs of students, according to psychological evaluations, interviews and behavior assessments

Psychological Services

- Plans and manages psychological services, including counseling to be provided to students and parents
- Assists in the development of positive behavior plans

Educational Team Members *Should…..*

- Collaborate as a team
- Provide support to each other
- Share good teaching/working strategies to meet the student's needs
- Develop goals and objectives together
- Provide services to meet the unique needs of the student
- Provide training as appropriate
- Make sure the IEP is implemented

THE
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Transition to Independent Living

Friday, 07/29/11
Breakout Session #3: 2:30-3:45
Panzacola F4

Tim Hartshorne, Ph.D.
Central Michigan University

Nancy Salem-Hartshorne, Ph.D.
Delta College

Presenter Information:

Dr. Tim Hartshorne is a professor of psychology, specialized in school psychology, at Central Michigan University. He has been researching and presenting about CHARGE syndrome since 1993, motivated by the birth of his son with CHARGE in 1989. His particular interest is in understanding the challenging behavior exhibited by many individuals with CHARGE. He is the grant holder for DeafBlind Central: Michigan's Training and Resource Project.

Dr. Nancy Salem-Hartshorne is an instructor at Delta College located in central Michigan. Her research has focused on developmental outcomes for individuals with CHARGE syndrome. Her published work has focused on cognitive and adaptive behavior skills in CHARGE, as well as on adolescent and adult medical and life issues. She has a son, Jacob, aged 22, who has CHARGE syndrome.

Presentation Abstract:

Many parents worry about where their child will live when he or she grows up. This presentation reviews the major choices and then proposes a four step process for making a decision: identify desired life experiences & goals, determine the pattern and intensity of support needs, develop an individualized plan, and monitor progress. These steps will be illustrated by the presenters' personal experience with their son.

10th International CHARGE Syndrome Conference
Rosen Shingle Creek Resort, Orlando, FL, July 28-31, 2011

Transition to Independent Living

Tim Hartshorne
Central Michigan University
Nancy Salem-Hartshorne
Delta College

Primary Alternatives

- Live with parents in parents' home
- Live in group home or institution
- Live independently with supports

Remaining with Parents

- Familiar and comfortable
- Parents can keep the person safe
- Parents can advocate
- Costs
  - What happens when parents are incapacitated?
  - Possible restrictions to developing independent living skills
  - Restrictions to parents' freedom

Group Home

- A safe, comfortable environment
- Able to live with peers
- Little choice regarding structure and amenities
- Costs
  - No choice in who you live with
  - Restrictions on freedom based on house rules
  - May tend to community isolation

Own Home

- Maximum freedom in how to live
- Staff totally focused on one individual
- Can design the home to meet the preferences of the one person living there.
- Costs
  - May be expensive to establish
  - Must rely on staff to insure community inclusion
  - Issues related to quality of staff
  - Loneliness?

How to decide?

- The availability of local models
- Parent instinct to protect and keep close
- Guilt factors
- What would the person prefer? How to know?
- General philosophy and beliefs

Support Plan Development

Four Component Process:

1. Identify desired Life Experiences & Goals
2. Determine Pattern & Intensity of Support Needs
3. Develop Individualized Plan
4. Monitor Progress

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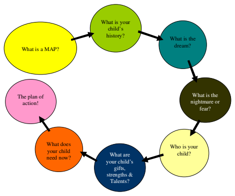
1. Identify desired life experiences and goals

Person Centered Planning

- A tool for advocacy
- A tool for inclusion
- A tool for planning and setting the path



Planning

- Traditional Planning
  - Formal assessments
  - Deficiencies and needs
  - Professional teams, roles and reports
- Person Centered Planning
  - Discovering and learning who the person really is
  - Finding capacities and gifts in the other person
  - Using family and friends and other connections

GOALS OF PERSON CENTERED PLANNING

- Being Present And Participating In Community Life
- Gaining And Maintaining Satisfying Relationships
- Expressing Preferences And Making Choices
- Continuing To Develop Personal Competencies
- Fulfilling Respected Roles And Living With Dignity

Making Action Plans

- What is this person's history and your history with this person?
- What are your dreams for this person?
- What are your nightmares for this person?
- Who is this person (one or two words/phrases)
- What are this person's strengths?
- What are this person's needs?
- What can we do to plan for the future this person wants?

ACTION PLAN

- Move toward the dream
- What would a perfect day be like for this person?
- Who is willing and able to support?

A goal for Jacob

- Jacob will live in his own home
- A consistent goal from pre-Kindergarten
- But how much support would he need?

Guiding Philosophy

The best (effective & efficient) way to meet people's needs is by supporting them in the life they want!

2. Determine Pattern & Intensity of Support Needs

Defining Supports

Resources and strategies that promote the interests and welfare of individuals and that result in:

- Enhanced personal independence and productivity
- Greater participation in society
- Increased community integration
- Improved quality of life

Intensity of Supports

- The range could be from mostly independent to requiring 24-7 support
- Areas to consider
  - Home living (love)
  - Community living (friendship)
  - Lifelong learning (work)
  - Employment (work)
  - Health and safety (love)
  - Social activities (friendship)
  - Protection and advocacy (love)

Supports Intensity Scale

- The SIS is a planning tool developed by the American Association on Intellectual and Developmental Disabilities to assess needs of people with intellectual and developmental disabilities

Supports Intensity Scale

Interview and Profile Form

AAMR

What does the SIS offer?

- Direct, reliable, and valid measure of an individual's support needs across life areas.
- Evaluation of the impact of significant medical and behavioral conditions.
- Profile of an individual's support needs in comparison to a national sample of adults with developmental disabilities.
- Process/tool for assisting in planning of an individual's supports needs.
- Useful tool to be included in individualized resource allocation.

3. Develop Individualized Plan

Developing the Plan for Jacob

- Identifying funding
- Finding the right house
- Remodeling the house
- Finding the right staff

Identifying Funding

- In Michigan key agency is Community Mental Health
- Jacob already received respite care funding from CMH
- Availability of Habilitation Waiver
- Intensity of needed supports
  - High

Finding the Right House

- The house next door!
  - Owned by an elderly lady
  - Same floor plan
as our main floor
  - We can keep an
eye on things

Remodeling the house

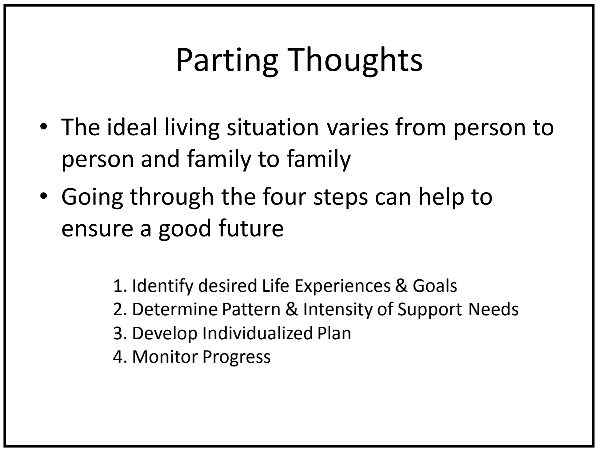
- Meeting to brainstorm fixing up Jacob's house
- Finding contractors who
  - Believe in the project
  - Are willing to find ways to save money
- Volunteers
- Donations

Finding the Right Staff

- "Jacob was the best teacher I had at CMU."
- CMU students from many departments
- We hire and train and support
- Money comes from CMH
- Paid through a fiscal intermediary

4. Monitor Progress

- Is Jacob happy in his home?
- Does Jacob continue to make progress with independence?
- Is Jacob progressing in terms of pre-vocational skills?
- Does Jacob continue to experience community inclusion?



Parting Thoughts

- The ideal living situation varies from person to person and family to family
- Going through the four steps can help to ensure a good future

1. Identify desired Life Experiences & Goals
2. Determine Pattern & Intensity of Support Needs
3. Develop Individualized Plan
4. Monitor Progress

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CHARGE 103: Questions and New Family Orientation

Friday, 07/29/11
Breakout Session #3: 2:30-3:45
Panzacola G1

Meg Hefner, MS, Genetic Counselor
Kim Blake, MD, Pediatrician
Sandra Davenport, MD, Geneticist
Rob Last, Communication Specialist

Please Note: CHARGE 101, 102 and 103 are meant as an introduction to CHARGE and to the Conference for New Families (and those who want a refresher)

Presenter Information:

These four presenters have more than 100 years of combined experience with CHARGE syndrome as well as experience and expertise in other areas of pediatrics, genetics and development. See the information at 101 and 102 for more information on each presenter. Between the four of us, our experience and expertise ranges from prenatal genetics to medial issues to language development.

Presentation Abstract:

This session of the CHARGE 101, 102 and 103 series has three goals:
1. Ask all the questions you want of the four presenters, including written questions submitted during the 101 and 102 sessions.

2. Get oriented to the conference. If you have questions about or want more information about a particular topic, we can direct you to presentations later in the conference or tell you whom to corner to ask your questions…. Where should you go when, what should you be doing… we can help.

3. Meet other families who have been to conference before. At this session, we have asked some “experienced” family members to come by to meet and greet and help you find your way around. We hope this will make you feel more at home among the big crowds at conference and find your way to sessions which will best meet your needs.

To accomplish these goals, this session will be a combination of “whole group” and small group sessions at the individual tables.

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What worked for you?

Friday, 07/29/11
Breakout Session #3: 2:30-3:45
Panzacola G2

Marlyn Minkin, LMFT/LMHC

Presenter Information:

Marlyn Minkin began her professional career decades ago as a Teacher of the Deaf. Soon after, she began providing parent support groups as a part of educational programs. Her experience includes Directing Programs and Agencies serving Deaf, Hard of Hearing and Deaf-Blind Children. She has facilitated hundreds of parent groups worldwide for families who have children that are Deaf-Blind. Marlyn is currently in private practice as a therapist in the Seattle area.

Presentation Abstract:

This session is a facilitated discussion that allows parents to share their experiences, ask questions and discover what strategies other parents may have used in similar situations

THE
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Advances in Understanding CHD7 through Use of Genetically Engineered Mice

Friday, 07/29/11
Breakout Session #3: 2:30-3:45
Wekiwa 3 & 4

Donna Martin, MD, PhD,
Elizabeth A. Hurd, PhD,
Wanda S. Layman, PhD,
Yehoash Raphael, PhD

The University of Michigan

Presenter Information:

Donna M. Martin is a Physician-Scientist and Associate Professor at The University of Michigan Medical School in the Departments of Pediatrics and Human Genetics. Her expertise is in Medical Genetics of developmental disorders including CHARGE syndrome.

Elizabeth A. Hurd is a Senior Research Associate working in Dr. Martin's laboratory. Dr. Hurd generated *Chd7* mutant mice and is analyzing them for inner ear defects and hearing abilities.

Wanda S. Layman is a recent PhD graduate of the Department of Human Genetics at The University of Michigan. She worked in Dr. Martin's laboratory and generated all of the data on endocrine and olfactory systems in *Chd7* mutant mice.

Yehoash Raphael is Professor of Otolaryngology at The University of Michigan. He specializes in studies of the inner ear, with a special focus on CHARGE syndrome.

Presentation Abstract:

CHD7, the gene mutated in human CHARGE syndrome, encodes a chromodomain DNA-binding protein that is highly expressed in specific tissues of the developing embryo. Our laboratory has generated and analyzed several different strains of mice with mutations in the mouse *Chd7* gene, with the goal of exploring the underlying mechanisms by which CHD7 regulates organ growth and development. We will discuss recent findings and roles for CHD7 in the development of several organs and tissues, including neurons that influence hearing, balance, and olfaction.

Advances in Understanding CHD7 through Use of Genetically Engineered Mice

CHARGE Syndrome Conference
July 28-31, 2011

Donna M. Martin, MD, PhD
Elizabeth A. Hurd, PhD
Wanda S. Layman, PhD
Yehoash Raphael, PhD

Departments of Pediatrics, Human Genetics, and Otolaryngology

The University of Michigan

Outline

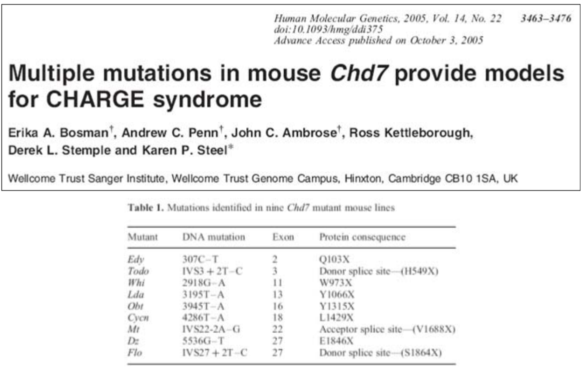
- *Chd7* deficient mice
  - – ENU mutants
  - – Gene trapped allele
  - – Conditional (flox) allele
- Organ system-specific defects
  - – Olfactory
  - – Endocrine
  - – Inner ear

Mouse models of CHARGE Syndrome

- ENU-derived mutants (10 alleles) with single base pair heterozygous loss of function mutations in *Chd7*
- *Chd7*$^{Gt/+}$ gene trapped loss of function allele
- Phenotypes of *Chd7* heterozygous mutant mice are consistent with those observed in CHARGE patients

Hurd et al., *Mammalian Genome*, 2007; Bosman et al., *Human Mol Gen 2005*

First report of *Chd7* mutant mice



Human Molecular Genetics, 2005, Vol. 14, No. 22 ***3463–3476***
doi:10.1093/hmg/ddi375
Advance Access published on October 3, 2005

Multiple mutations in mouse *Chd7* provide models for CHARGE syndrome

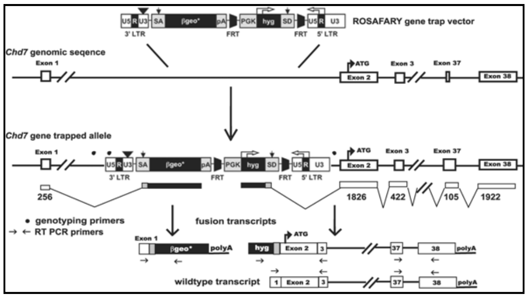
Erika A. Bosman[†], Andrew C. Penn[†], John C. Ambrose[†], Ross Kettleborough, Derek L. Stemple and Karen P. Steel[*]

Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge CB10 1SA, UK

Table 1. Mutations identified in nine *Chd7* mutant mouse lines

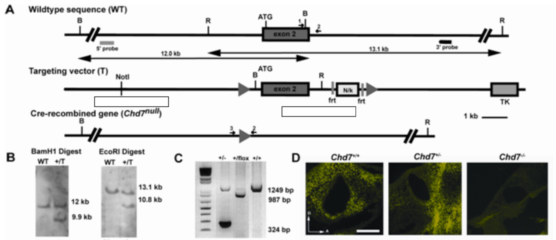
Mutant	DNA mutation	Exon	Protein consequence
Edy	307C–T	2	Q103X
Todo	IVS3 + 2T–C	3	Donor splice site—(H549X)
Whi	2918G–A	11	W973X
Lda	3195T–A	13	Y1066X
Obt	3945T–A	16	Y1315X
Cycn	4286T–A	18	L1429X
Mt	IVS22-2A–G	22	Acceptor splice site—(V1688X)
Dz	5536G–T	27	E1846X
Flo	IVS27 + 2T–C	27	Donor splice site—(S1864X)

Chd7$^{Gt/+}$ mice are a model for CHARGE



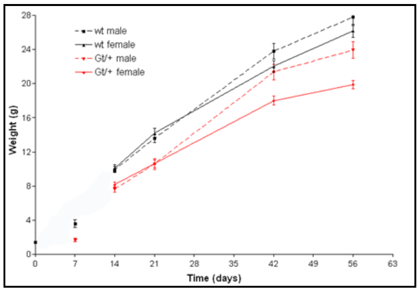
Hurd et al., *Mammalian Genome*, 2007

Generation of a *Chd7*^{flox} allele



Hurd et al., *Development* 2011

Chd7$^{Gt/+}$ mutants have postnatal growth delays and circling



Hurd et al., *Mammalian Genome*, 2007

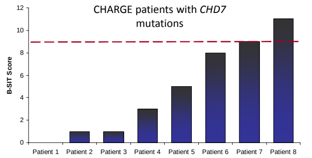
Outline

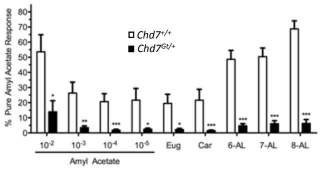
- *Chd7* deficient mice
  - – ENU mutants
  - – Gene trapped allele
  - – Conditional (flox) allele
- Organ system-specific defects
  - – Olfactory
  - – Endocrine
  - – Inner ear

Olfaction and CHARGE syndrome

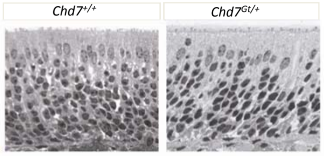
- Olfactory bulb defects (33/33) and olfactory impairment (18/19) are common features of CHARGE
- *Chd7* is expressed in olfactory epithelium and olfactory bulb in humans and mice

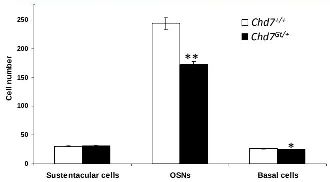
Layman et al., Human Molecular Genetics 2009





Olfactory sensory neurons are reduced in *Chd7*$^{Gt/+}$ mice

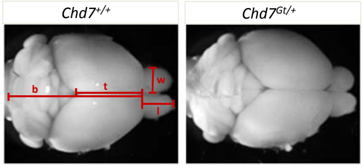


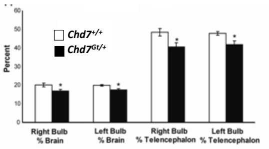


* p<0.05
** p<0.001

Layman et al., Human Molecular Genetics 2009

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





* p < 0.05
** p < 0.01

Layman et al., Human Molecular Genetics 2009

Conclusions (olfactory)

- *Chd7*$^{Gt/+}$ mice have olfactory defects similar to human CHARGE individuals
- Olfactory sensory neurons are reduced in *Chd7*$^{Gt/+}$ mice
- Cellular proliferation is reduced in *Chd7*$^{Gt/+}$ olfactory epithelium

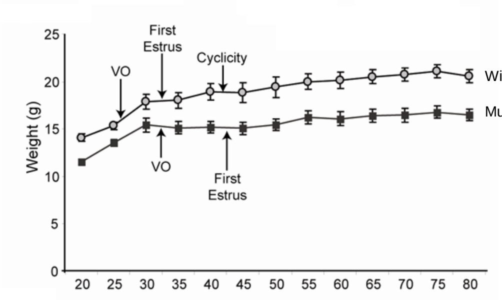
Outline

- *Chd7* deficient mice
  - – ENU mutants
  - – Gene trapped allele
  - – Conditional (flox) allele
- Organ system-specific defects
  - – Olfactory
  - – Endocrine
  - – Inner ear

Endocrine dysfunction and CHARGE

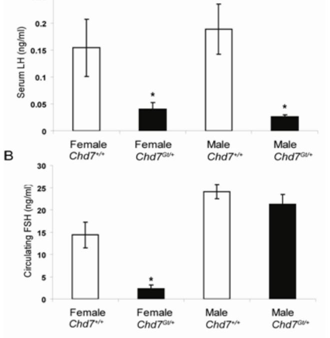
- 81% of males and 93% of females with CHARGE have LH and FSH are deficient
- Genital hypoplasia including cryptorchidism and micropenis occurs in 62% of CHARGE individuals with confirmed *CHD7* mutations
  - – Females often have hypoplastic labia
- Anosmia and hyposmia can predict idiopathic hypogonadotropic hypogonadism in CHARGE individuals (Bergman et al., 2010)

Chd7$^{Gt/+}$ female mice have delayed puberty



Layman et al., Human Molecular Genetics 2011

Chd7$^{Gt/+}$ mice have decreased levels of LH and FSH

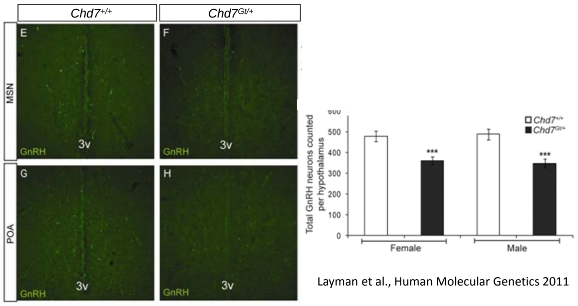


Chd7$^{Gt/+}$ females and males have reduced LH

Chd7$^{Gt/+}$ females have reduced FSH

Layman et al.,
Human Molecular Genetics 2011

GnRH neurons are reduced in *Chd7*$^{Gt/+}$ mice



Layman et al., Human Molecular Genetics 2011

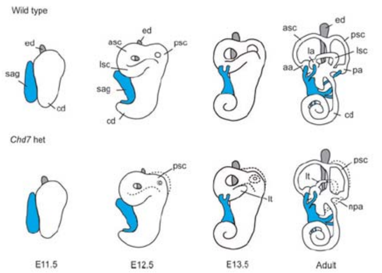
Conclusions part II (endocrine)

- *Chd7*$^{Gt/+}$ mice have pubertal defects and decreased LH, FSH similar to human CHARGE individuals
- GnRH neurons are reduced in *Chd7*$^{Gt/+}$ embryos and adults
- Cellular proliferation is reduced in the olfactory epithelium of *Chd7*$^{Gt/+}$ embryos
- Reduced CHD7 dosage lowers expression of *Bmp4*, *Fgfr1*, *Otx2*, *GnRH1*, and *GnRHR*

Outline

- Chromatin remodeling proteins
  - – Classification and roles in human disease
  - – CHD7 and CHARGE Syndrome
- Organ system-specific defects
  - – Olfactory
  - – Endocrine
  - – Inner ear

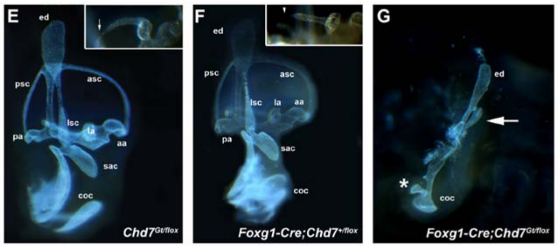
Chd7$^{Gt/+}$ mice have defects in inner ear morphogenesis



Elizabeth Hurd

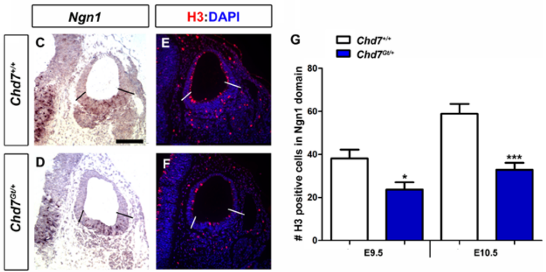
Layman et al, *Clin Gen* 2010; Hurd et al, *Mamm Gen* 2007;
Bosman et al, *HMG*, 2005; Adams et al, *JCN* 2008

FoxG1cre-Chd7 conditional mutants have severe semicircular canal and cochlear defects



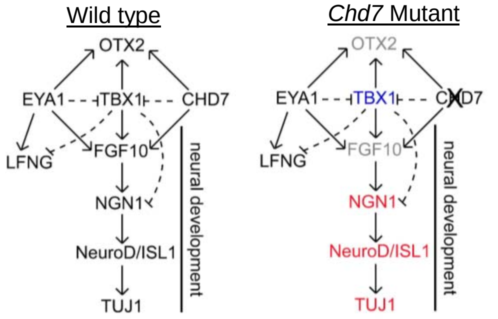
Hurd et al., *Development*, 2010

Chd7 mutants have reduced proliferation in the neurogenic domain



Hurd et al., *Development*, 2010

Model for CHD7 Developmental Gene Regulation in Inner Ear



Wild type

OTX2
EYA1
TBX1
CHD7
LFNG
FGF10
NGN1
NeuroD/ISL1
TUJ1
neural development

Chd7 Mutant

OTX2
EYA1
TBX1
CHD7
LFNG
FGF10
NGN1
NeuroD/ISL1
TUJ1
neural development

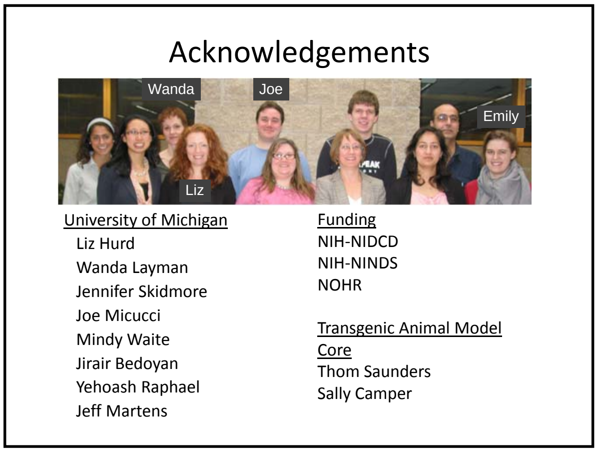
Hurd et al., *Development*, 2010

Conclusions part III (inner ear)

- *Chd7*$^{Gt/+}$ mice have inner ear defects and hearing loss similar to human CHARGE individuals
- Inner ear neuroblast proliferation is sensitive to CHD7 dosage
- CHD7 likely acts upstream of proneural genes to regulate inner ear neurogenesis

Take-home points

- CHD7 deficiency affects development of multiple similar tissues in humans and mice
- Neurogenesis in the olfactory epithelium and inner ear requires appropriate CHD7 dosage
- Mouse mutants are a powerful tool for exploring CHD7 function during development and beyond



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Jirair Bedoyan
Yehoash Raphael
Jeff Martens

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NIH-NINDS
NOHR

Transgenic Animal Model Core
Thom Saunders
Sally Camper

THE CHARGE SYNDROME FOUNDATION

Entendiendo Mi Equipo. Los Roles y Responsabilidades del los Miembros del Equipo

Friday, 07/29/11
Breakout Session #3: 2:30-3:45
Wekiwa 5

Gloria Rodriguez-Gil
Especialista Educativa
Servicios de Sordoceguera de California

Información del Presentador:

Gloria Rodriguez-Gil ha sido una especialista educativa para los Servicios de Sordoceguera de California por los últimos once años, y ha trabajado en el campo de la discapacidad visual, discapacidad múltiple y sordo-ceguera casi veinte. Ella proporciona asistencia técnica y entrenamiento a equipos educativos, y ha escrito varios artículos y blogs sobre formas en cómo el equipo educativo puede mejor apoyar a estudiantes con sordo-ceguera en el salón de clase. Usted los puede leer en Español en http://www.cadbs.org/es/

Resúmen de la presentación:

Entender los roles y responsabilidades de los miembros del equipo es crítico para una buena colaboración porque entonces los padres de familia pueden saber qué esperar y pedir a cada uno de los miembros del equipo. Esta sesión describirá los roles y responsabilidades de todos los miembros del equipo y de cada miembro individualmente. Estos incluyen: El padre de familia, el maestro del salón de clase, el mediador o interventor o asistente uno-a-uno, el maestro de estudiantes con problemas visuales, el maestro de sordos y duros de oído, el especialista en orientación y movilidad, el consultor en sordo-ceguera, el terapista de lenguaje, el terapista ocupacional, el terapista físico y el psicólogo escolar.

Entendiendo Mi Equipo. Los Papeles y Responsabilidades de los Miembros del Equipo.

Gloria Rodriguez-Gil

California Deaf-Blind Services

10th International CHARGE Syndrome Conference

Orlando, Florida

El Equipo Educativo

Especialista del Programa

Padre de Familia

Maestro del Salón de Clase

Psicólogo

Interventor o mediador

Terapista Físico

Terapista Ocupacional

Maestro de Estudiantes con Discapacidad Visual

Maestro de Sordos y Duros de Oído

Especialista en Orientación y Movilidad

Terapista de Lenguaje

Especialista en Sordo-ceguera

> ***“Todos tienen una especialidad— pero ningún miembro del equipo puede trabajar autónomamente de los otros.”***
>
> **Terry Boisot, madre de familia**

Padre de Familia

- Es la fuerza propulsora, uno de los líderes del equipo
- Ve la totalidad, es un miembro consistente del equipo
- Es una fuente única de información: sobre el niño, sobre el Síndrome de CHARGE
- Comparte información y ayuda a desarrollar metas que el niño alcanzará durante el año escolar
- Aboga fuertemente pero debe ser muy sensible a los temores y necesidades de los educadores
- Promueve la colaboración en el equipo

Especialista del Programa

- Responsable del manejo de casos individuales
- Generalmente tiene conocimento básico de las discapacidades del niño
- Asiste a las reuniones educativas de toma de decisiones, donde se discuten servicios para el estudiante

Especialista del Programa

- Apoya el trabajo del equipo educativo para mejor servir al estudiante
- Desarrolla el documento del IEP describiendo en detalle cuáles servicios se proporcionarán y cómo se realizarán los servicios
- Provee al estudiante con un programa educativo que cubre logros académicos así como logros sociales y emocionales

Maestro(a) del Salón de Clase

- Persona a cargo del salón de clase
- Debe trabajar en colaboración con el equipo del IEP
- Debe colaborar con los miembros del equipo para adaptar el ambiente, actividades, y materiales del salón de clase para que sean accessibles al estudiante

Maestro(a) del Salón de Clase

- Debe promover la socialización entre compañeros de clase y de escuela
- Idealmente debe trabajar directamente con el niño

Asistente Uno-a-uno/ interventor o mediador

- La Sordo-Ceguera es una discapacidad de acceso a información visual y auditiva
- Los individuos con sordo-ceguera necesitan acceso al ambiente, comunicación y apoyo social y emocional
- El papel de un interventor o mediador es el de facilitar este proceso

Asistente Uno-a-uno/ interventor o mediador

- El interventor o mediador es un para-profesional con habilidades y entrenamiento especializados para proveer apoyo directo a un estudiante con sordo-ceguera.
- La decisión de usar un interventor o mediador se basa en el nivel de apoyo requerido por el estudiante para tener acceso a la información
- El interventor o mediador es el puente o conexión entre el individuo con sordo-ceguera y el mundo

Maestro(a) para Estudiantes con Discapacidad Visual

- Realiza la evaluación funcional visual
- Refiere al estudiante a una clínica de baja visión para su evaluación
- Evalúa y apoya con ayudas visuales
- Determina cuáles materiales visuales son apropiados para el estudiante (tamaño, color, contraste).
- Adapta materiales escritos (tamaño de letra, Braille).

Maestro(a) para Estudiantes con Discapacidad Visual

- Evalúa y modifica el ambiente visual
- Provee instrucción en Braille
- Muchos maestros para estudiantes con discapacidad visual tienen experiencia limitada trabajando con estudiantes sordos o duros de oído, o con discapacidades múltiples.

Especialista en Orientación y Movilidad

- Ayuda a un estudiante que es ciego o con baja visión a saber dónde está en el espacio y dónde desea ir (orientación)
- Ayuda a un estudiante que es ciego o con baja visión a tener un plan para llegar a donde desea (movilidad)
- Este especialista tiene experiencia moviéndose en el espacio con ojos vendados o con gafas de baja visión

Especialista en Orientación y Movilidad

- Enseña al estudiante los conceptos de espacio, ambiente y a cómo usar la información recibida a través de otros sentidos (sonido, temperatura, vibración) para mantener o re-dirigir la orientación
- Enseña al estudiante cómo usar el bastón blanco y perro guía como ayuda o como solo instrumento de guía

Especialista en Orientación y Movilidad

- Enseña al estudiante cómo usar apoyos a la visión residual y la baja visión cuando viaja
- Ayuda al estudiante a aprender sobre su ambiente, y el lenguaje que el estudiante necesita usar en ese ambiente

Especialista en Orientación y Movilidad

- Desarrolla adaptaciones que le permitirán al estudiante relacionarse con el público (v.g., tarjetas de comunicación en el transporte público)

Especialista en Sordo-Ceguera (CA)

- Provee entrenamiento a los equipos de IEP a nivel estatal y regional
- Provee asistencia técnica en persona, por vía telefónica, Internet y video
- Provee asistencia técnica urgente
- Ayuda a crear y participa en una red de apoyo familia-a-familia
- Disemina información, recursos y productos

Terapista de Habla y Lenguaje

- Identifica los problemas de habla del estudiante
- Evalúa estudiantes con problemas de habla
- Refiere estudiantes a profesionales médicos para más profunda evaluación
- Provee servicios de habla y lenguaje para mejorar o prevenir problemas de comunicación

Terapista de Habla y Lenguaje

- Provee consejo y guía a padres, niños y maestras en el área de habla y lenguaje
- Algunos terapistas conocen sobre otras modalidades de comunicación tales como Lenguaje de Señas y comunicación aumentativa y alternativa

Maestro(a) de Sordos y Duros de Oído

- Observa el ambiente de clase para ver qué se puede hacer para mejorar el aprendizaje del estudiante
- Revisa que dispositivos de ayuda al oído, sistemas de FM e implantes cocleares funcionen bien
- Determina con el equipo educativo la mejor manera de comunicarse con el estudiante (habla, volumen, distancia, Lenguaje de Señas, claves-objeto, etc.)

Maestro(a) de Sordos y Duros de Oído

- Conoce sobre el desarrollo formal del lenguaje en personas duras de oído (hablado y escrito, comprensión y uso)
- Muchos de estos profesionales tienen experiencia limitada trabajando con estudiantes que son ciegos o con baja visión, o con discapacidades múltiples, y la información que dan otros miembros del equipo es muy importante

Terapista Ocupacional

- Trabaja con las habilidades motoras finas, la coordinación y la fortaleza para mejorar la movilidad y las habilidades físicas
- Ayuda al estudiante a trabajar con los sentidos disponibles
- Trabaja en asuntos de alimentación, uso manual (manipulativos) e independencia física

Terapista Ocupacional

- Conoce sobre el equipo adaptado que puede ayudar al estudiante a ser tan independiente como sea posible
- Crea una “dieta sensorial” (palabra originada por la TO Patricia Wilbarger) que es un plan de actividades cuidadosamente diseñado y personalizado que provee los estímulos sensoriales que la persona necesita para mantenerse enfocado y organizado a través del día.

Terapista Físico

- Este service generalmente trabaja en postura, fortalecimiento muscular, movilidad y resistencia
- Trabaja en habilidades motoras gruesas
- Provee experiencias y práctica para mejorar el desarrollo motor.
- Monitorea el uso apropiado del equipo durable indicado

Servicios Psicológicos

- Realiza evaluaciones/estudios
- Interpreta los resultados de evaluaciones/estudios
- Provee consulta a los miembros del equipo educativo, para planear programas educativos que llenen las necesidades únicas de los estudiantes, de acuerdo con las evaluacions psicológicas, las entrevistas y los estudios de comportamiento.

Servicios Psicológicos

- Planea y administra Servicios Psicológicos, incluyendo el consejo que se deba proveer a padres y estudiantes
- Asiste en el desarrollo de planes de comportamiento positivo

Los Miembros de Equipo Educativo *DEBEN…..*

- Colaborar como un equipo
- Proveerse apoyo los unos a los otros
- Compartir las buenas estrategias de trabajo o enseñanza para llenar las necesidades de los estudiantes
- Desarrollar juntos metas y objetivos
- Proveer servicios para llenar las necesidades únicas del estudiante
- Proveer entrenamiento donde sea apropiado
- Asegurarse de que el IEP sea llevado a cabo

THE CHARGE SYNDROME FOUNDATION

Taking Back the IEP and Making It Work for Our Kids

Saturday, 07/30/11
Breakout Session #5: 10:45-11:45
Panzacola F1 & F2

Ellen Steinbrick
Mom

Presenter Information:

I have been Mom to Alex for almost 14 years. I have learned a lot through the process that needs to be shared. We have been through the wringer as far as obtaining a public school education. Alex is the very first child with the deaf/blind classification to attend school in our town. I have used a few of the remedies (complaint process) to make the program work for Alex instead of a cookie cutter program that was thrown together. Through the complaint process I have been able to meet people who have shared vital information that I was able to use to create a workable IEP.

I have also had my share of failures that have been wonderful learning opportunities that have allowed me to learn the law and its applications.

Presentation Abstract:

My objective is to share the information gathered through the complaint process. This information was all about the details. Often the districts write the IEPs in a vague format left up to interpretation. I would like to help parents prepare for IEP and participate in the meeting. Help parents question the information districts put in the IEP and what they leave off. I will help parents understand the remedies set forth through IDEA.

IEPs were designed to be the maps to our kid's education. IEPs should be customized to your child's exact needs to allow our kids to make meaningful progress. All too often IEPs are written with cookies cutter goal and objectives pulled off IEPgoalbank.com.

When speaking with the attorneys from Office of Civil Right I found the importance of the language written on the following pages of the IEP: These pages vary from state to state

Prior written notice page: the page that will list all actions proposed and denied

Present levels of Academic Achievement and Functional Performance: the page that lists strengths, concerns and how the disability impact the child's ability to access the general education curriculum.

Program Accommodations and Modifications: the page that list modifications and accommodations to help the student make progress and be involved in general education. This is a VERY important page. (I did not prevail on certain areas of our complaint due to vague language on this page)

Special Education, Related Services, and Regular Education: the page list how the service hours are broken down, who implements goals, the frequency, the responsible staff, and where the services will take place.

These pages are vital to the development of the IEP. Districts often use vague language that leaves them with little or no responsibility. During my OCR complaint an accommodation was listed “as needed” and did not list “all classes” OCR reluctantly found that although it was common sense for it to be used to the benefit of the student in all classes the district was not required if they felt it was unnecessary. The lesson learned is that these pages need to use exact language so that all using this IEP can implemented.

Many drafters of IEPs have explained they know what they mean when they write goals. I have learned the hard way that when a teacher would not be following my son to the next year as planned the new teacher did not know where to begin and it was a frustrating process. Working to keep the language clear and concise would have alleviated some of the delay that was experienced.

The page that reflect present level of performance written using exact language can assist the team in writing good measurable goals and objectives. Listing strengths, concerns and impact accurately without using vague statements allow a new team member to pick up the IEP and being implementation.

Districts often use words and statements that are specifically vague: These are excerpts actually written in my son’s IEPs.

“As needed” for modifications(What criteria being used to determine the need)

“When appropriate” (What criteria being used to determine appropriate)

“When feasible” (What criteria being used to determine feasible)

“The student will use relaxation techniques to manager anxiety” (What are the relaxation techniques and how does this help manage anxiety)

Often I have experienced goal writers to “know what they mean” but some goals are not able to be implemented:

“The student will safely cross the street”… Mastery criteria were listed as 80% of trials. (Wow what happens to the other 20% of the time) I kept reading this to the OT and she did not get it. I finally said are you expecting him to get hit the other 20% of time?

When you read the IEP out loud and can say….

How can this be done?
When will this be used?
Why is this being used?
Is this a vague goal/objective that doesn’t reflect my child’s actual need?
Will this be a life time skill or does it just fit district curriculum?

We all have these glitches in our IEPS and they are easily fixed. I have been lucky this year I asked for the case manager to give the goals ahead of time and we were able to discuss some changes that will make it a better IEP. We are all a team working to help children get an education.

THE
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Language Acquisition through Motor Planning (LAMP) – Part 1

Saturday 07/30/11
Breakout Session #5: 10:45 – 11:45
Panzacola F3 & F4

John Halloran M.S.CCC-SLP
Center for AAC and 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. He also finds much reward in exploring ways to best implement assistive technology with children who have autism. John Halloran graduated from the University of Arkansas at Little Rock in 1990 with a bachelor's degree in Communication Disorders. He 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 is the primary developer of Language Acquisition through Motor Planning(LAMP) and presents internationally. John maintains membership in several professional organizations, including the American Speech Language Hearing Association and the Arkansas Speech Language Hearing Association.

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 developmental disabilities a method to develop independent and spontaneous communication. Strategies to teach language/communication skills within this framework will be discussed.

Thank You

Alex Halloran

Alex meets Dad

Alex and Ellie

- If this were my child I would….

- Making effective communication, a human right, accessible and achievable for all.
- (ASHA 2009)

The Center for AAC & Autism Mission:

- To improve public awareness of the unique qualities of the power of AAC to change the lives of non-verbal individuals with autism and other developmental disabilities by:
  - Providing specialized clinical training to health care professionals, teachers, and parents
  - Supporting clinical research
  - Supporting clients and families with education, resources, and information

Frigid Mother Syndrome

- 1. Talk softly
- 2. Look into your child's eyes
- 3. Be gentle
- 4. Do not be so rigid
- 5. Get on the floor and play
- 6. Sabotage their environment

Lazy eyed John

Presumed Causes

- 1. Inadequate sensory processing
- 2. Poor auditory processing
- 3. Apraxia

Ming, Brimacombe, Wagner 2007

- Reviewed studies of 154 children with ASD and found that…
  - 41% 2 – 6 year olds
  - 27% 7-18 year olds
- Showed clear evidence of oral motor and/or hand muscle apraxia

LAMP LAW

Every word has a unique motor plan

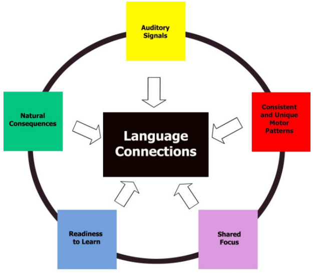
- **"In the practiced automatic** movements **of daily life attention is directed to the sense impression and not to the movement. So, in piano playing, the beginner may attend to his fingers but the practiced player attends only to the notes or to the melody. In speaking, writing and reading aloud, and in games and manual work, attention is always directed to the goal, never to the movement. In fact, as soon as attention is directed to the movement, this becomes less automatic and less dependable."**
-
- **Cattell, J.M.**

1893

Max Conway

www.aacandautism.com

Five key elements



Readiness to Learn

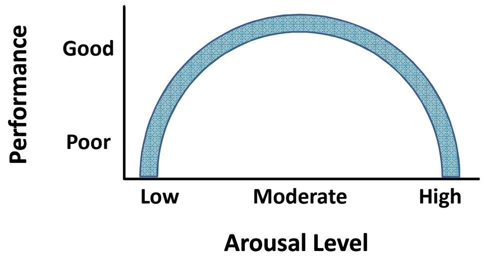
Some children with ASD need to be calmed in order to be in a state of readiness to interact. Other children need to have their level of alertness increased to be ready to learn. In either instance, the child must be ready at emotional and sensory levels before learning can occur.

Sensory Strategies to Ready the Child to Learn

- Calming sensory input
  - Slow repetitive movements
  - “Heavy work”/ resistive activities
  - Deep pressure

- Alerting sensory input
  - Quick, unexpected movements
  - Loud noises, music
-

The Inverted U-Principle



(Duffy, 1962)

Too Much

Brandy

" Brandi is becoming spontaneous. She likes it a lot" –Jill Carpenter, Speech Therapist

Child Directed

- Follow the child's lead:
  - Build on the child's interests
  - Carefully use barriers
  - Join in with the child
  - Let the child make the moves
  - Use of Movement

Shared Focus

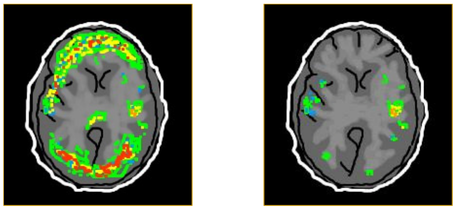
- **Joint attention**: the process by which one alerts another to a stimulus via nonverbal means, such as gazing or pointing. For example, one person may point to another, and then point to an object. In this case, the pointing person is trying to get the other to look at the object. The person seeing the other point responds to the gesture by looking at the object. It typically develops around one year of age in human infants and is essential to building strong social connections. Before one year of age, infants merely look at the hand of the person pointing; after developing joint attention, they look in the direction of the pointing. Chimpanzees also show some understanding of joint attention, although they primarily use it as a means to an end, rather than for pure communicative purposes.[1] Although chimps do display joint attention, the general consensus is that only humans use it in a purely altruistic way. (Wikipedia)

Consistent and Unique Motor Patterns

- Levelt's (1993) model of spoken language focuses on the cognitive processes associated with utterance generation. Levelt argued that "normally speakers have no conscious access to language encoding or articulation. For most speakers, language production is relatively effortless." (The Efficacy of Augmentative and Alternative Communication, Schlosser, 2003 p. 48).

- Fitts and Posner (1967) proposed a three stage model of motor learning. The first stage is the cognitive stage in which the learner has to attend to the process of learning a motor action. This stage is marked by highly variable performance. The learner may or may not know what they are doing wrong or how to correct their performance and will need guidance to assist them. In the associative stage, he works on refining his skill and is able to detect and correct their errors. The autonomous stage is the result of a lot of practice. At this stage, the learner does not have to concentrate on the movement and can attend to other aspects of the activity.

Automaticity



- EMG brain activity: new vs. automatic tasks
- Habitual movements become subcortical
- Cortical areas can then be put to “better use”

Automaticity: The Great Equalizer

- Natural Language
  - Cognitive activity: Formulation of thoughts
  - Motor activity: Speech & Gesture (automatic)
- AAC Language
  - Cognitive activity: Formulation of thoughts
  - Motor activity: Device activation (automatic???)

Pattern not Metaphor

Auditory Signal/Feedback

La Sorte (1993), he found that synthetic speech facilitated natural speech production. He found that, “Synthetic speech can facilitate the segmenting of speech into word units since the boundaries are more clearly defined than in human speech, and stress is not an important aspect of synthesized speech.”

- Prizant (1983): “Individuals with autism have trouble segmenting incoming speech into meaningful word units.”

- Pinker (1994): In speech, one "word runs into the next seamlessly; there are no silences between spoken words the way there are white spaces between written words. We simply hallucinate word boundaries when we reach the edge of a stretch of sound that matches some entry in our mental dictionary."

- In the *Language Instinct* by Steven Pinker, he credits Chomsky for our understanding of "two fundamental facts about language. First, virtually every sentence that a person utters or understands is a brand new combination of words appearing for the first time in the history of the universe. Therefore, a language cannot be a repertoire of responses. The brain must contain a recipe or program that can build an unlimited set of sentences out of a finite list of words. That program may be called a mental grammar

- **According to Pinker (1994), if a person started memorizing all the possible 10-word sentences, it would take, at a rate of five seconds a sentence, a hundred trillion years** (with no time for eating or sleeping).

Pinker: 2 tricks to language

- "The first is the arbitrariness of the sign." The wholly conventional pairing of a sound with meaning. The word dog does not look like a dog, walk like a dog, or woof like a dog, but it means dog……for the price of this standardized memorization, the members of a language community receive an enormous benefit….

Second trick

- The second trick behind language instinct is captured in a phrase from Wihelm Von Humbodt that presaged Chomsky,

- "Language makes infinite use of finite media."

- Tigers Woods
- Grapes Soda
- Golf Horse
- "Ocean" for "lotion
- "Fix bird" for "Vicksburg"
- No more go away (Karli sign)

Motor movements affect perception

Articulatory gestures, rather than sounds, are critical for both production and perception of speech. On neurobiological grounds, fronto-temporal circuits are thought to play a functional role in production as well as comprehension of speech. The coactivation of motor circuits and the concurrent perception of self-produced speech sounds during articulations might lead to correlated neuronal activity in motor and auditory systems, triggering long-term plastic processes based on Hebbian learning principles (D'Ausilio et al. (2009). "The Motor Somatotopy of Speech Perception." *Current Biology*. 19, 1-5.)
Piano

Integrating vision and hearing

Random Selection and Perseveration

- Hide
- May be what they are wanting to say

How to Model Vocabulary to Achieve Motor Automaticity

- The goal is that the child will press the correct icon or icon sequence spontaneously and independently. However, to get to that end, you may have to help the child through the movement initially. You want to back off the amount of cuing that you are giving as soon as possible so that the child does not become cue dependent. Remember, it is a lot easier to remember how to get somewhere if you were the driver last time rather than the passenger.

Levels of motor assistance:

- Hand over hand
- Point to icon
- Point to general area of icon
- Point to device
- Wait for child to activate

How Automaticity Works

- Motor Learning
  - Acquisition & retention of movement for task
- Repetition of a neuromotor pathway
  - Eventually requires less energy
  - Eventually enhances performance
  - Example: touch typing

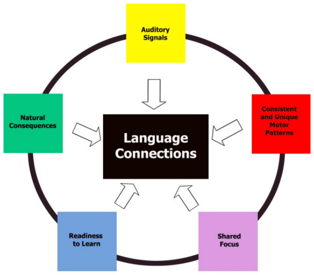
Automaticity on an AAC Device

- Possible when:
  - Location of icons is consistent
  - Small icon set allows for immediate recognition
  - Moving from one icon to the next is predictable
- Not Possible when:
  - Significant Navigation is required
  - Continuous visual refocus & reorientation is required

Language Connections

- The AAC device is a tool that allows the child to “babble” and learn about language. For example, after saying “more” to get bubbles, the child might think that pressing the key for “more” means “bubbles.” A beginning talker might make the same conclusion. However, in another instance when the child says “more” and gets more juice, the meaning of “more” is revised by the child.
- As the child learns with the AAC device, he/she is learning consistent motor patterns that result in an auditory signal. Depending on the natural consequence triggered by the auditory signal, the child may modify his/her perception of what the auditory signal means.

Language Connections



Child Directed/ Therapist Guided

- Natural environments
- Treat the child as a communicator
- Quality vs. quantity

Selecting Vocabulary

- Use core words
- Broad meaning vs. specific
- Verbs vs. nouns
- Frequency of use vs. picture predicting ability
- Words vs. phrases

Access to Single Words

- *“Communication is based on the use of the individual words of our language. True communication is spontaneous and novel. Therefore, communication systems cannot be based significantly on pre-stored sentences. Communication requires access to a vocabulary of individual words suitable to our needs that are multiple and subject to change. These words must be selected to form the sentences that we wish to say.”*

American Speech-Language-Hearing Association

ASHA’s AAC Glossary

Why does everyone teach sign for more?

- “Certain words such as not or more have broader application to objects and events than other words, such as cookie or car, and thus may be heard more often and will serve the child more frequently in his or her effort to communicate”

(Bloom and Lahey, 1979)

Passion not Obsession

- Train
- Disney
- Beads
- Rope
- Vacuum Cleaner

Give 10 examples of using go and stop

- 1
- 2
- 3
- 4
- 5
- 6
- 7
- 8
- 9
- 10

10 examples of how to teach core words “go” and “stop” in the classroom

1. Go vs. stop at circle time to start pledge
2. Kids all dance/jump around until they hear stop then they all freeze
3. On slide, child says “go” and “stop” for next child to come down slide
4. Therapist gets child or self in rolling chair/wagon
5. Spinning/swinging
6. Musical chairs go go go go go go stop
7. Doll that you squeeze and it pee’s on “go” quits on “stop”
8. Video go = play and stop = pause
9. Animated pictures on power point of things going and stopping
10. Make fan stop and go on command
11. Computer games with mouse click to make something stop and go

Core Vocabulary has few Picture Producers

- Fewer than 5 percent of all words used by toddlers are picture producers
  (Banajee, Dicarlo & Stricklin, *AAC*, 2003)
- Concrete graphics can only be made for picture producers
- More than 90 percent of core vocabulary words are not picture producers
  (Hill, Dissertation, Establishment of Performance Indices, 2001)

Generalize Vocabulary in a Single Session

- Use word in a variety of activities
- Use language to request, tell, and limited labeling

Expand Vocabulary Learning

- Add new words as automaticity develops
- Add fringe words
- Allow miss-hits

Over generalize

- Over generalize words to give them more meanings
- Visual reactions to words is the best method to teach new words
- 3 sensory systems used to teach words
    1. tactile
    2. auditory
    3. visual

Model Vocabulary for Automaticity

- Decrease cuing to encourage independence
- The location of a learned icon or icon sequence should not change

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- **501-951-1588**

THE
CHARGE
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Bringing You Protected Tomorrows: Your Journey Through an Eight-Step Special Needs Planning Program

Saturday, 07/30/11
Breakout Session #5: 10:45-11:45
Panzacola G1

Mary Anne Ehlert
President & Founder
Protected Tomorrows, Inc.

Presenter Information:

Mary Anne Ehlert is a financial professional and sister to an individual with disabilities. She is highly regarded as a specialist in working with families of individuals with disabilities, and serves on the boards of several organizations, including; Vice-Chair, National Disability Institute in Washington DC, Illinois Special Olympics and Gateway to Learning School for Special Needs. She is a member of the Alliance for the Mentally Ill, ARC of Illinois, and the National Council on Aging. She speaks to conferences and other audiences on financial planning, and has recently completed her book, entitled “The Gift I Was Given.”

Presentation Abstract:

As family members of a person with a disability, we so often tend to view life in light of the challenges presented to us. Bringing You Protected Tomorrows looks at our lives in a whole new light, as Mary Anne Ehlert provides a passionate glimpse of her life with her sister, Marcia. The presentation will be uplifting and entertaining, as well as provide a good basic understanding of some important facts. Participants will develop the foundation for their family’s own personal Eight Step Future Care Plan™. Some topics of discussion include: how to create a map of the future; how to prepare legally and financially; how to incorporate and maximize government benefits; how to prepare for transition; and how to communicate your Future Care Plan™ to your family.

BRINGING YOU PROTECTED TOMORROWS

Your Journey Through An Eight Step Special Needs Planning Program

My Take-Aways: What I have accomplished and what I need to do next.

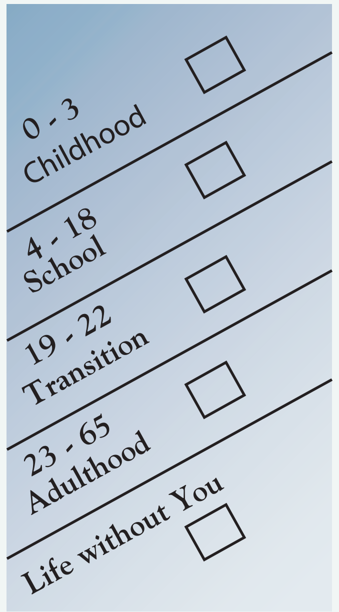
PROTECTED TOMORROWS®

STAGE 1:
TAKE a Candid Look™

Dreams & Goals

Concerns & Fears

STAGE 2:
CREATE the Future Map™

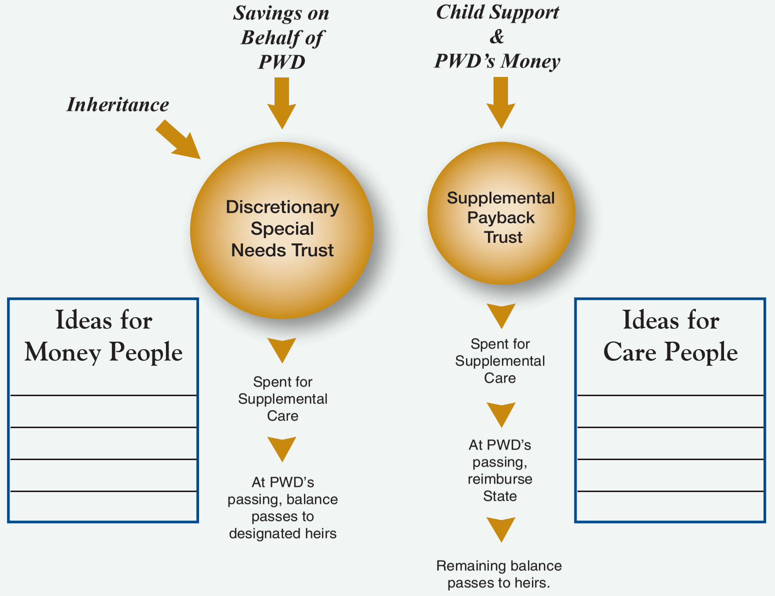

0 - 3 Childhood ☐
4 - 18 School ☐
19 - 22 Transition ☐
23 - 65 Adulthood ☐
Life without You ☐

Income
SSI ____________
Medicaid ____________
Family ____________
Other ____________

Expenses
Living ____________
Medical ____________
Recreation ____________
Education ____________
Other ____________

STAGE 3:
FILTER the Legal Options™

SPECIAL NEEDS TRUSTS


Inheritance
Savings on Behalf of PWD
Discretionary Special Needs Trust
Spent for Supplemental Care
At PWD's passing, balance passes to designated heirs

Child Support & PWD's Money
Supplemental Payback Trust
Spent for Supplemental Care
At PWD's passing, reimburse State
Remaining balance passes to heirs.

Ideas for Money People

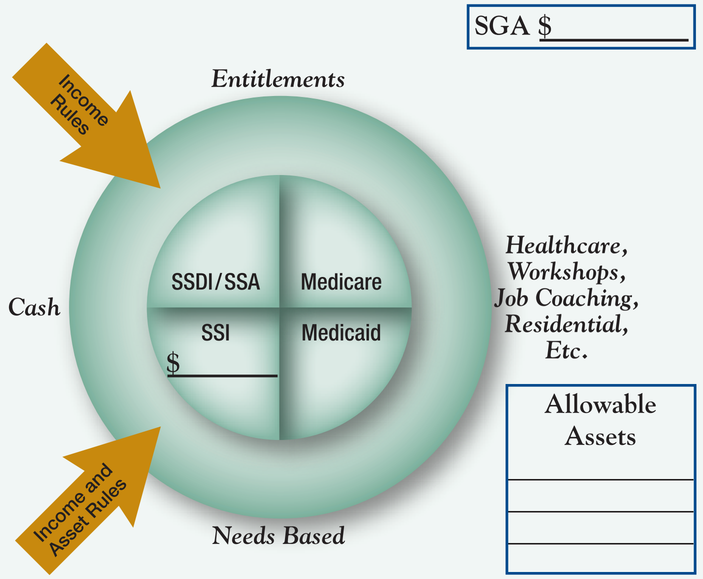
Ideas for Care People

*** PWD - Person With Disabilities*

STAGE 4:
CAPTURE Potential Benefits™

BENEFIT PROGRAMS

SGA $ ____________


Income Rules
Entitlements
Cash
SSDI/SSA
Medicare
SSI
$ ________
Medicaid
Income and Asset Rules
Needs Based
Healthcare, Workshops, Job Coaching, Residential, Etc.

Allowable Assets

STAGE 5:
DOCUMENT the Wonder™

Residential	*Education / Employment*
Social Life & Routines	*Medical Care*

STAGE 6:
BEGIN the Transition™

	Your Life	*PWD Life*
Live		
Learn		
Work		
Play		

STAGE 7:
FUND the Future™

TODAY

Assets	*Liabilities*

FUTURE NEEDS

Savings ____________
Family ____________
Insurance ____________
Other ____________

STAGE 8:
REVIEW and Renew™

I Promise to . . .

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Presenter's Information

MARY ANNE EHLERT

Mary Anne Ehlert, CFP®, is a financial professional and sister to an individual with disabilities. She is highly regarded as a specialist in working with families of individuals with disabilities and the elderly and speaks to conferences and television audiences on financial planning.

President and Founder of both Protected Tomorrows, Inc. and Ehlert Financial Group, Inc., Mary Anne is a member of the Board of Directors of many organizations providing services to children and adults with special needs, including; Vice-Chair, National Disability Institute in Washington DC, Illinois Special Olympics, Gateway to Learning School for Special Needs, and Illinois St. Coletta's. She is a member of the Alliance for the Mentally Ill, The ARC of Illinois and the National Council on Aging.

Mary Anne has been honored for her contributions to the Lake County Advocacy Conference for Persons with Disabilities.

"My sister Marcia, who was born with cerebral palsy, changed my life forever. Who would I have become without her? My close relationship with my sister taught me what really matters, what really is important in life, not only day-to-day, but also for the long term.

"Marcia taught me that saying how one feels is important. She taught me not to be afraid of trying something new even though I might fail. And she helped me discover that there is a way I can make a difference in the lives of others.

"I've learned that sharing my own background and feelings about growing up with Marcia helps other families focus on the positives while they deal with the challenges. They respond to my candor about the dynamics of my own family .... I can see them nod their heads when I talk about how family members coped differently with my sister's disabilities .... they feel secure in planning with someone who knows what they're going through.

"I've also learned how Marcia impacted my entire family, especially my parents. In addition to helping the other five of us get on our feet and reach responsible adulthood, they devoted their lives to Marcia for 34 years. They aged 10 years in one week after deciding to give Marcia the opportunity to live residentially with her peers. They ultimately learned to find joy in her growth at Clearbrook, even though they missed her at every turn.

"Being able to make a difference in the lives of other families and helping them solve some of their unique challenges brings me great comfort. Marcia is no longer of this earth but I'm convinced she looks over my shoulder every day and smiles at her legacy."

Mary Anne Ehlert is founder of Protected Tomorrows, Inc.
Protected Tomorrows, Inc. is not a Registered Investment Adviser and does not engage in offering financial or legal advice.

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ADVOCATING FOR FAMILIES WITH SPECIAL NEEDS

When someone has a family member with a disability, they often depend upon their family to help them live the best life they can. Protected Tomorrows Advocates work with the family to design a Future Care Plan™, starting with the big picture and working down to the smallest details. We focus on quality of life and peace of mind for the entire family. The Future Care Plan must be both practical and sustainable. We collaborate with the family's entire team, which may incorporate attorneys, financial advisors, banks, trust departments, insurance agents, schools, residential facilities, social workers, and government agencies.

THE PROCESS FOR PROTECTED TOMORROWS™

- **Stage 1: Take a Candid Look™:** Look honestly and comprehensively at your future care needs, in light of your own future needs and those of other family members. The results of this stage become the foundation for the assessment of planning needs for the family and the individual with special needs.
- **Stage 2: Create the Future Map™:** Identify the options and create a plan to protect your entire family's future in light of your needs. This Future Map focuses on the different areas of life, including Residential, Education, Employment, and Recreation.
- **Stage 3: Filter the Legal Options™:** Evaluate estate solutions, focusing on the various options available through professional legal resources. Implement the steps to take advantage of your decisions. In this step, we make sure all legal arrangements are made to accomplish goals identified in Stage 2.
- **Stage 4: Capture Potential Benefits™:** Identify and apply for supplemental programs to enhance and supplement the Future Care Plan you have created for your loved one. In this Stage, we identify resources available to supplement Stage 2 plans. This step may also include the actual application for applicable benefits.
- **Stage 5: Document the Wonder™:** Chronicle and treasure your loved one's special story and gifts so that others can carry forward in building your family member's abilities and self-esteem. Many times individuals want to share what they want to take place in their future; documenting it to ensure it is carried out is an important step.
- **Stage 6: Begin the Transition™:** Identify and evaluate the future residential, care and recreation options for your future. As each step needs additional services brought in, the coordination of these resources is important in order for the steps be carried out as desired and planned.
- **Stage 7: Fund the Future™:** Utilize financial solutions to complement the Future Care Plan designed for your future. This step is coordinated with your Financial Advisor to ensure proper funds are available for your care.
- **Stage 8: Review and Renew™:** Review annually your Future Care Plan to accommodate and address life's inevitable change.

103 Schelter Road, Lincolnshire, IL 60069 | 847.522.8086 | Fax 847.522.8081
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Rev 4/2011

PROTECTED TOMORROWS®

ADVOCATING FOR FAMILIES WITH SPECIAL NEEDS

The Protected Tomorrows Team recognizes that there are many different types of families and many types of needs. We are not one size fits all, but rather customize our services to the needs of the client. We provide services to families in the following ways:

➤ One-on-One

When working with an Advocate, families receive personal assistance with their planning. The Licensed Advocate will review all eight stages of the planning process, beginning with a complimentary meeting to Take A Candid Look™ at the families current situation, an honest appraisal of future care needs, and the entire family's financial situation.

➤ Workshop Setting

At a Family Workshop, Protected Tomorrows Advocates guide participants as a group through its planning process to begin the development of a Future Care Plan™. After this interactive and entertaining session, participants will understand what is needed to prepare a plan for a safe and fulfilling future for their loved one with special needs. A Family Workshop is typically 6 to 8 hours long.

➤ Online Planning System

Protected Tomorrows has developed an online Special Needs Planning System which provides families with the tools they need to plan for a safe and fulfilling future for their loved one with special needs. This online system allows subscribers to enter information into a database guiding them through the 8-step planning process. This information may be accessed by other family members, caregivers, etc., as you deem appropriate.

➤ Book with Online Assistance

The book "**THE GIFT I WAS GIVEN**" by Mary Anne Ehlert, provides hope to families of individuals with special needs who often feel lost, confused and alone. Most caregivers know how to be a good caregiver, but they don't know where to begin to look for help. The feeling of helplessness can be staggering. Knowing you are not alone and that there are many to reach out to is a huge help. "The Gift I Was Given" provides hope to those who are walking this path. The book also has a companion website to help families with a step-by-step process through the many areas addressed in the book.

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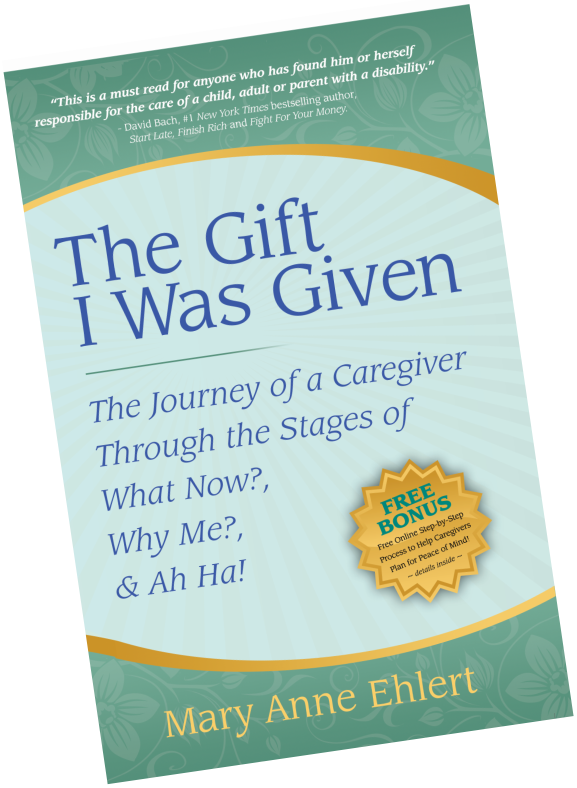
Rev 4/2011

NOW AVAILABLE!



"If you are blessed to have a family member with a disability, this book will give you hope for the future as well as the strategies to create a financial plan that will last beyond your lifetime. Mary Anne knows from experience how to make this happen."

Terry Savage, Nationally Syndicated
Sun-Times Financial Columnist

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THE
CHARGE
SYNDROME
FOUNDATION

Social Skills: The Entrée into Adult Life

Saturday, 07/30/11
Breakout Session #5: 10:45-11:45
Panzacola G2

Sharon Zell Sacks, Ph.D.
Director of Curriculum, Assessment, & Staff Development, California School for the Blind

Presenter Information:

Sharon Sacks, Ph.D. is the Director of Curriculum, Assessment, & Staff Development at the California School for the Blind. Prior to her appointment at CSB, Sharon was a full professor and the coordinator of the teacher preparation program in blindness & visual impairment at California State University, Los Angeles, Also, Sharon coordinated the teacher preparation program for students with moderate to severe disabilities at San Jose State University. Sharon has published widely and presented nationally and internationally in the areas of social skills instruction, psycho-social implications of visual impairments, and transition from school to adult life. She was the President of AERBVI, and has served on numerous boards.

Presentation Abstract:

This workshop will focus on how families can support their students in developing positive social skills that promote interdependence and social competence in the home, the school, the community, and the workplace. The presenter will share practical strategies that can be applied for all students with CHARGE.

Social Skills & Realistic Expectations: Keys to Interdependence

Dr. Sharon Zell Sacks
Director of Curriculum, Assessment, & Staff Development
California School for the Blind

Why are Social Skills Important?

- Social skills provide a way for students to engage with others.
- A repertoire of social skills help students to develop friendships & secure positive relationships
- The development of social skills is the foundation upon which other skills are acquired.
- Persons who exhibit positive social skills have successful employment & living options.

Why do Students with Visual Impairments & Deafblind Need to Learn Social Skills?

- Most of what is learned socially is acquired through vision.
- Lack of incidental learning requires adults to mediate the students social environment.
- The need for hands-on experiential learning promotes curiosity & a sense of self.
- Developing & acquiring social skills is a life-long process.

The Link Between Social Skills & Expectations

- Clear & realistic expectations promote social competence.
- When adults expect students to act socially appropriate, the student's self-esteem improves.
- Clear expectations provide clear messages for the student & the acquisition of socially acceptable behavior.

Who is Responsible for Teaching Social Skills?

- Families play a critical role.
- Teachers of Students with Visual Impairments & Orientation & Mobility Instructors
- School counselors & school psychologists
- Peers who are sighted & who have visual impairments

What is the Family's Role in Teaching Social Skills?

- Families are the essential key to teaching social skills.
- Families build partnerships with professionals to determine what to teach & to determine what is important to teach.
- Families reinforce & promote the social skills that are taught by professionals in real environments.

How Can Families Promote Socialization?

- Encourage participation in a variety of activities.
- Encourage turn-taking & social conversations.
- Include the child in all family activities.
- Provide opportunities & experiences to promote a can do attitude.
- Provide opportunities for your child to make decisions & choices. Encourage problem solving.
- Encourage responsibility & chores.
- Expect your child to act appropriately in social situations.
- Insist that social goals be part of your child’s IEP.

How to Determine What to Teach?

- Assessment is a key component & a partnership.
  - Observation
  - Analyzing behavior
  - Checklists & Interviews
  - Prioritizing Skills to Teach

Hierarchy of Social Skills

- Awareness Skills
- Interactive Skills
- Perspective-Taking & Problem-Solving Skills

Critical Social Skills

»

Critical Social Skills: Body Language

- Maintain appropriate eye contact
- Demonstrate appropriate body posture
- Maintain personal space
- Utilize & respond to gestures & facial expressions
- Refrain from socially unaccepted mannerisms

Critical Social Skills: Social Communication

- Engage with others
- Express wants & needs
- Makes choices
- Initiates & greets others
- Responds appropriate to positive & negative feedback

Critical Social Skills: Conversation Patterns

- Initiates a conversation with peers & adults
- Has a repertoire of conversation topics
- Maintains & expands a conversation
- Knows how to end a conversation
- Knows how to interrupt a conversation
- Knows how to take turns in a conversation
- Knows how to ask for help or assistance
- Invites friends for special events

Critical Social Skills: Cooperative Skills

- Shares with others in play or with a group
- Knows how to join a group & sustain involvement
- Works with others to accomplish a goal
- Leads groups activities
- Volunteers to complete tasks (class jobs or chores)

Critical Social Skills: Interaction Skills

- Greets others and says hello or good-bye
- Maintains interactions with peers or adults
- Can compromise
- Shows empathy & sympathy for others
- Compliments & encourages the efforts of others

Critical Social Skills: Social Etiquette

- Says/Signs please & thank you in an appropriate manner given the situation
- Uses manners when eating
- Shares, toys, & belongings with others
- Waits for turn
- Reciprocates when others assist

Critical Social skills: Relationships & Friendships

- Knows difference between family, friend, acquaintance, & stranger
- Shows interest in peers & has similar interests
- Shares in group activities
- Demonstrates concern for friends
- Has friends outside of school
- Demonstrates age-appropriate understanding of human sexuality
- Understands dating
- Demonstrate appropriate

Critical Social Skills: Knowledge of Self

- Knows likes & dislikes
- Takes responsibility for one's actions
- Understands & explains visual impairment
- Advocates for self in home, school, & community
- Demonstrates assertiveness in an appropriate manner

Critical Social Skills: Interpreting & Monitoring Social Behavior

- Asking questions at the appropriate time
- Knowing when not too obey an adult
- Interpreting non-verbal cues from others
- Understanding & interpreting sarcasm
- Monitoring one's behavior in a variety of situations
- Interpreting feelings & emotions from others
- Interpreting other's behavior in a social situation

Strategies to Promote Social Competence

Setting the Tone for Promoting Social Competence

- Be consistent
- Interpret & verbalize your child's communicative intent
- Establish routines & structure for expecting appropriate social behavior
- Give your child responsibilities, making him/her a participating member of the household

Developing Play Skills

- Help your child to understand that play has function & meaning.
- Nurture curiosity through exploration.
- Provide opportunities for your child to play with toys & functional objects.
- Encourage your child to share with others.
- Encourage turn-taking: The concept of give & take.
- Model a range of conversations topics & age-appropriate vocabulary.

Developing Play Skills

- Provide a range of real life experiences so that your child can share them in pretend play.
- Encourage your child to try new & adventurous activities.
- Don't say no: If your child expresses a desire to engage in an activity, provide the experience.

Developing Friendship Skills

- Encourage your child to be part of the family milieu.
- Encourage & promote interactions with family members, siblings, & peers.
- Join groups that allow your child to engage with others (play groups, music, art, gymnastics, church groups).
- Promote a genuine desire to engage with others.
- Teach your child to compliment peers & adults.
- Teach your child to take the role of others.

Developing Social Etiquette

- Encourage your child to have good manners. Be polite, but not too polite.
- Encourage your child to develop appropriate eating skills.
- Help your child to become aware of age-appropriate dress. Teach age-appropriate grooming skills.

Developing Non-Verbal Communication Skills

- Help your child to learn typical gestures used by sighted students.
- Interpret & help your child learn facial expressions that match specific emotions.
- Assist your child in learning to interpret non-verbal expressions from others.

Developing Self-Advocacy Skills

Encourage your child to express their wants & needs, while attending to the impact their needs have on others.

Encourage students to take responsibility for their acts & belongings by becoming a contributing family member.

Encourage your child to ask for assistance only when needed & to reciprocate.

Resources

Teaching Social Skills to Students with Visual Impairments: Research to Practice **(Sacks & Wolffe, 2006). AFB Press**

Focused on: Social skills **(Wolffe, & Sacks, 2000) AFB Press**

Social Awareness Curriculum (Crow & Herlich, 2011). APH

Looking Good: A Curriculum for Personal Appearance & Social Presence (Corn, Bina, & Sacks, 2007). Pro Ed

Resources

Independent living: A curriculum with adaptations for students with visual impairments **(Loumiet & Levack, 1993). TSBVI**

Promoting acceptance of children with disabilities: From tolerance to inclusion **(MacCuspie, 1996). AFB Press**

Get out of my life but first could you drive me and Cheryl to the mall? **(Wolf, 1991)**

Contact Information

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California School for the Blind
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ssacks@csb-cde.ca.gov
(510 794-3800 Ext. 313)

THE CHARGE SYNDROME FOUNDATION

Creating Calm from CHARGE Chaos

Saturday, 07/30/11
Breakout Session #5: 10:45-11:45
Wekiwa 3 & 4

Catherine Rose
MOM, PhD, MBA

Presenter Information:

Catherine Rose is mom to Alexis and Jessica. Alexis (5) has CHARGE and Jessica (4) has a heart defect. Although Catherine has a PhD and MBA, it's her MOM degree that has taught her the important life lessons. Catherine worked for Lucent and taught business management classes. Now, Catherine works for Philips Healthcare, bringing Alexis' experiences to a global healthcare company. In her free time, Catherine works to support other families in their challenging medical journeys and hopes to inspire confidence in parents to use their voices.

Presentation Abstract:

We are all on a CHARGE journey – it's a sprint when we're in the hospital or managing a crisis and a marathon when we're juggling daily demands. We must be educated, empowered, enabled and experts as caregivers. This presentation will help caregivers with strategies to simplify managing medical care, build an outstanding team to address their children's needs, and create calm out of CHARGE chaos.

Creating Calm from CHARGE Chaos

Catherine Rose

She Fights, We Fight

She fought to be here…

We fight to keep her safe.

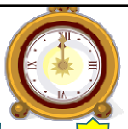
Alexis & Kaitlyn were identical twins.	**High Risk Pregnancy**
At 20 week ultrasound, diagnosed potential heart defect	**Ordered Amnio, No known genetic defects found**
At 29 weeks, Could not find Kaitlyn's heartbeat	**Emergency Transport to Boston**
Alexis had lung steroids as a precaution for premature delivery.	**Sent home for Strict Bedrest.**

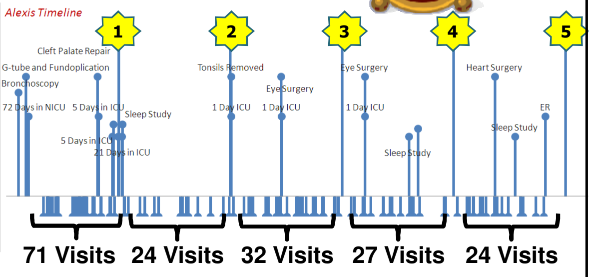
Delivery Day… NICU Time

19 doctors and nurses	**Better Safe than Sorry**
4 days in NICU for monitoring	**Waiting for Poop**
72 days later….	**Discharged with Unexplained Genetic Condition**

Keep her safe. Do no harm.

Hospital-Time





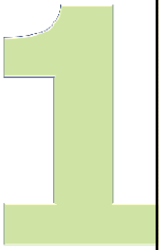
Team of Doctors

- LOCAL PEDIATRICIAN
- COMPLEX CARE SERVICES
- OTOLARYNGOLOGY
- DIAGNOSTIC AUDIOLOGY
- OPHTHALMOLOGY
- PULMONARY
- PEDIATRIC SLEEP DISORDERS
- CARDIOLOGY
- DEAF / HARD OF HEARING PROGRAM
- AUGMENTATIVE COMMUNICATION PROGRAM
- FEEDING AND SWALLOWING PROGRAM
- NEW ENGLAND LOW VISION CLINIC
- GASTROENTEROLOGY
- PHYSICAL MEDICINE / PHYSIATRIST
- GENERAL SURGERY
- ENDOCRINOLOGY
- NEPHROLOGY
- ORTHOPEDICS
- GENETICS
- DENTISTRY
- NUTRITION
- PLASTICS
- NEUROLOGY

Create Outstanding Team

- You are the manager / leader.
- Define the common purpose for a team.
  - Examples: Keep your child alive. Avoid certain treatment. Achieve certain treatment.
- Define performance goals.
  - Examples: No hospitalization rule. Quality of life. Growing and meeting expectations.
- Build team with complementary skills.
  - Examples: We may not be Doctors (we play 1 on TV though). Build your team with people that offer different skills.
- Have mutual accountability.
  - Trust, rely and create an environment of trust and support.

Create Calm from Papers

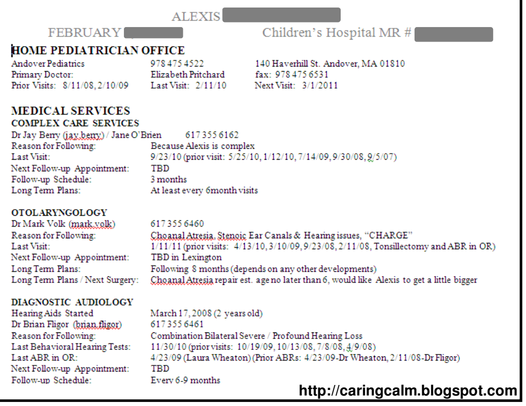


- **Power of One**
  - Keep them in **1** place (bin, room, file cabinet)
  - Review, Purge if possible 1 per year
  - Try to keep it isolated to that 1 place
- Get them from the hospital
  - be diligent about requesting speedier service
- Scan them!
  - Keep on your computer or external drive.
  - Email to doctors, therapists, teachers!

Create Calm from Medical History and Appointments

- Create your own document to keep in 1 place
- Keep it in your language, your understanding
- Include
  - Doctor, Phone number, email
  - Past appointments, Future appointments
  - Summary of last appointment
    - (approximately 1 paragraph)

“Patient Summary” Example



ALEXIS [redacted]

FEBRUARY [redacted] Children’s Hospital MR # [redacted]

HOME PEDIATRICIAN OFFICE

Andover Pediatrics	978 475 4522	140 Haverhill St. Andover, MA 01810
Primary Doctor:	Elizabeth Pritchard	fax: 978 475 6531
Prior Visits: 8/11/08, 2/10/09	Last Visit: 2/11/10	Next Visit: 3/1/2011

MEDICAL SERVICES

COMPLEX CARE SERVICES

Dr Jay Berry (jay.berry) / Jane O’Brien	617 355 6162
Reason for Following:	Because Alexis is complex
Last Visit:	9/23/10 (prior visit: 5/25/10, 1/12/10, 7/14/09, 9/30/08, 9/5/07)
Next Follow-up Appointment:	TBD
Follow-up Schedule:	3 months
Long Term Plans:	At least every 6month visits

OTOLARYNGOLOGY

Dr Mark Volk (mark.volk)	617 355 6460
Reason for Following:	Choanal Atresia, Stenoic Ear Canals & Hearing issues, “CHARGE”
Last Visit:	1/11/11 (prior visits: 4/13/10, 3/10/09, 9/23/08, 2/11/08, Tonsillectomy and ABR in OR)
Next Follow-up Appointment:	TBD in Lexington
Long Term Plans:	Following 8 months (depends on any other developments)
Long Term Plans / Next Surgery:	Choanal Atresia repair est. age no later than 6, would like Alexis to get a little bigger

DIAGNOSTIC AUDIOLOGY

Hearing Aids Started	March 17, 2008 (2 years old)
Dr Brian Fligor (brian.fligor)	617 355 6461
Reason for Following:	Combination Bilateral Severe / Profound Hearing Loss
Last Behavioral Hearing Tests:	11/30/10 (prior visits: 10/19/09, 10/13/08, 7/8/08, 4/9/08)
Last ABR in OR:	4/23/09 (Laura Wheaton) (Prior ABRs: 4/23/09-Dr Wheaton, 2/11/08-Dr Fligor)
Next Follow-up Appointment:	TBD
Follow-up Schedule:	Every 6-9 months

“Patient Summary”

Alexis Example

Microsoft Office
rd 97 - 2003 Docum

Generic Example

Patient Summary
PDF

Adobe Acrobat
Document

Patient Summary
Word Document

Microsoft Office
Word Document

Create Calm from Activities and Schedule

- Keep a calendar of all activities
  - Google Calendar
  - Excel
  - MS Outlook
  - Apple Apps
  - Paper
- Make it visual
- Create reminders for crucial activities

“Calendar” Example

Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
0/1/2006	10/2/2006	10/3/2006	10/4/2006	10/5/2006	10/6/2006	10/7/2006
	9:45 EI PT 11:30 Hearing Tech	Recycling BHI Shipment NOPCO 3 Coordinated Care Appt F10	34 wk! 11:30 GI - Mickey F5 5 EI Beth	VA	VA condo rental $800	NC STEW
Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
0/8/2006	10/9/2006	10/10/2006	10/11/2006	10/12/2006	10/13/2006	10/14/2006
3 month old!	VA - Columbus Day 9:45 EI PT 12 EI Beth	VA	start 40 hrs/wk START DAYCARE 9C5M	TO WORK 7.5C5M	8-12 Heating Company 7.5M12.5C 2pm G-tube evaluation condo	
Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
0/15/2006	10/16/2006	10/17/2006	10/18/2006	10/19/2006	10/20/2006	10/21/2006
	No daycare UML-diploma ready 9:45 EI PT 12 EI Beth 1:45 EI Jessie	Recycling 8:45am Stomach Surgeon F3 TO WORK 11c5m	36 wk! 8:30 - Chimney Cleaning 7.5m5c Pick up dry-cleaning FIX-IT DINNER WITH P&M	7.5m4.75c TO WORK 5:45 Flu Vaccine for Alexis 7pm Sign Language @ House	8:20am Dr Volk Waltham BROCHETTI LUNCH 2-3 mtg, 3-3:30	9am Shared Reading Saturd
Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
0/22/2006	10/23/2006	10/24/2006	10/25/2006	10/26/2006	10/27/2006	10/28/2006
0am Trot for Tots	Lucent Open Enrollment 9:45 EI PT 2 Ultrasound, 3 Swallow DC2	TO WORK	TO WORK 5 EI Beth	RFMV review TO WORK 7-9 Sign Language @ House	9 Dr Cox Genetics - Boston NOPCO	
Sunday	Monday	Tuesday	Wednesday	Thursday	Friday	Saturday
0/29/2006	10/30/2006	10/31/2006	11/1/2006	11/2/2006	11/3/2006	11/4/2006
Go to Steve & Trish for ToT	9:45 EI Janna @ Day Care 12 EI Beth NOPCO-Mike Call BHI for shipment Matt-Guitar	Recycling TO WORK Methuen ToT	38 wk! 9:45 Synagis Shot - And Ped BHI Shipment TO WORK GYM	w2 7-9 Sign Language @ House	TO WORK condo?	

"Calendar"

Family Example

Microsoft Office
:el 97-2003 Workshe

Generic Example

Calendar
PDF

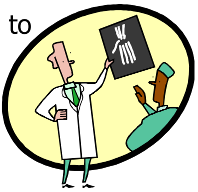
Adobe Acrobat
Document

Calendar
Word Document

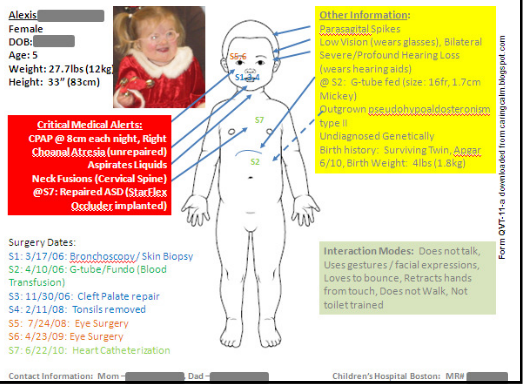
Microsoft Office
Word Document

Create Calm for Chaotic Times

- ER Visits
- New Doctors
- Introducing your child to new caregivers
- Introductions to other families



"Quick View" Example



Alexis [redacted]
Female
DOB: [redacted]
Age: 5
Weight: 27.7lbs (12kg)
Height: 33" (83cm)

Critical Medical Alerts:
CPAP @ 8cm each night, Right Choanal Atresia (unrepaired)
Aspirates Liquids
Neck Fusions (Cervical Spine)
@S7: Repaired ASD (StarFlex Occluder implanted)

Other Information:
Parasagital Spikes
Low Vision (wears glasses), Bilateral Severe/Profound Hearing Loss (wears hearing aids)
@ S2: G-tube fed (size: 16fr, 1.7cm Mickey)
Outgrown pseudohypoaldosteronism type II
Undiagnosed Genetically
Birth history: Surviving Twin, Apgar 6/10, Birth Weight: 4lbs (1.8kg)

Surgery Dates:
S1: 3/17/06: Bronchoscopy / Skin Biopsy
S2: 4/10/06: G-tube/Fundo (Blood Transfusion)
S3: 11/30/06: Cleft Palate repair
S4: 2/11/08: Tonsils removed
S5: 7/24/08: Eye Surgery
S6: 4/23/09: Eye Surgery
S7: 6/22/10: Heart Catheterization

Interaction Modes: Does not talk, Uses gestures / facial expressions, Loves to bounce, Retracts hands from touch, Does not Walk, Not toilet trained

Form QVT-11-a downloaded from caringcalm.blogspot.com

Contact Information: Mom – [redacted], Dad – [redacted]

Children's Hospital Boston: MR# [redacted]

"Quick View"

Alexis Example

Microsoft Office
werPoint Presentati

Generic Example

Quick View
PDF

Adobe Acrobat
Document

Quick View
Powerpoint

Microsoft Office
werPoint Presentati

Create Calm from Communication

- All in 1 place
  - Blogs
  - CarePages
  - CaringBridge
  - Facebook
- Alert friends and families
- Synchronize across teams

"Communication" Example

Saturday, February 19, 2011

Belated Christmas Picture

Alexis came to Philips for Family Fun Day (Open House).
She was photographed with Santa (don't tell Alexis but he works at Philips too!)
She looks like a GIANT to us in this picture.
Absolutely ADORABLE!!! She's a big 5 year old now!
Catherine

Create Calm from Life

- Life is chaotic
- Build simple systems that are easy to maintain
- Build support network for help
  - kids after school
  - prepping a few meals each week
  - respite care

http://caringcalm.blogspot.com

And remember… They are Kids afterall!

- Have fun!
- Our kids are kids, and even our big kids are always kids at heart!

- All is possible – nothing is impossible!

http://caringcalm.blogspot.com

Create Calm for yourself

- Our journey is long.
- Know who you are.
- Be at peace with your best.
- Know that we all struggle, in different ways.
- Reflect, take time and love yourself and your family.

- **Create calm from the chaos.**

http://caringcalm.blogspot.com

For More Information

- On this topic:
  - http://caringcalm.blogspot.com
  - Cat_Rose@comcast.net

- On my family:
  - http://schnaderbeck.blogspot.com
  - http://www.bit.ly/alexis

Molecular Studies to Uncover the Cellular Functions of CHD7

Saturday, 07/30/11
Breakout Session #5: 10:45-11:45
Wekiwa 5

Peter C. Scacheri, PhD
Assistant Professor
Department of Genetics
Case Western Reserve University

Presenter Information:

Peter Scacheri graduated with a BS in Biology from Gettysburg College and earned his Ph.D. in Biochemistry and Molecular Genetics from the University of Pittsburgh. His graduate work was focused on the genetics of muscular dystrophy. His postdoctoral fellowship was at the National Human Genome Research Institute at the National Institutes of Health, where he studied a type of cancer that affects the endocrine organs. Dr. Scacheri is currently an Assistant Professor in the Department of Genetics at Case Western Reserve University School of Medicine. The Scacheri lab uses cutting edge genomics to investigate the function of the CHD7 protein and its role in CHARGE syndrome. Dr. Scacheri's research on CHARGE syndrome is supported by an R01 grant awarded from the National Institute of Child Health and Human Development.

Presentation Abstract:

It is known that DNA mutations in the CHD7 gene cause CHARGE syndrome, but how? My lab has been addressing this question by investigating the function of CHD7 in both normal and CHD7 mutant cells from humans, mice, and zebrafish. Our research indicates that CHD7 functions in the cell nucleus to fine-tune the expression of genes that control the development of organs that are affected in CHARGE syndrome. In addition, CHD7 activates genes that encode components of the protein manufacturing machinery of all living cells. These findings suggest that the multiple anomalies in CHARGE syndrome are due to the combined effects of altered gene expression and reduced protein synthesis.

Molecular studies to uncover the cellular functions of CHD7.

Peter C. Scacheri, PhD
Department of Genetics
Case Western Reserve University
Cleveland, OH

Outline

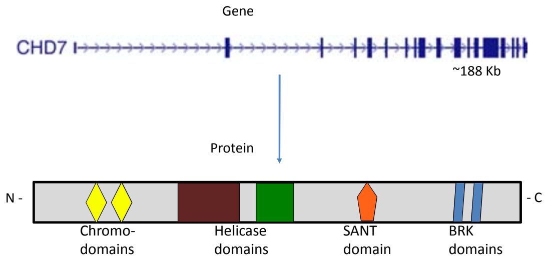
- Overview of the cellular functions of CHD7
    - – Regulator of genes that orchestrate development
    - – Regulator of protein synthesis.
- Zebrafish model of CHARGE syndrome
- Overview of high-throughput sequencing of CHD7 to identify mutations in patient cohorts
- Where we are headed

Mutations in *CHD7* (<u>c</u>hromodomain <u>h</u>elicase <u>D</u>NA-binding protein <u>7</u>) cause CHARGE syndrome

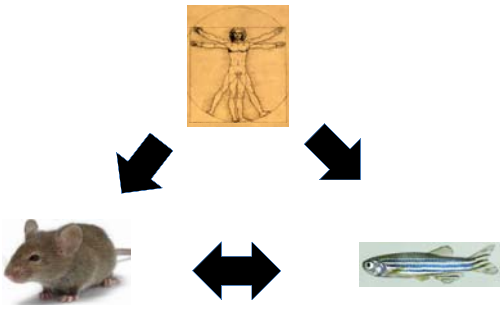
● = nonsense
● = frame-shift
○ = splicing
● = missense

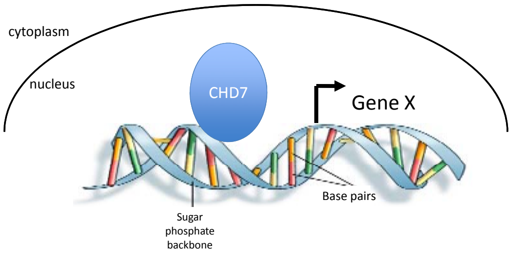
- Mutations in 58-71% of patients
- Arise spontaneously
- Mostly protein truncation mutations (Loss-of-function)
- Haploinsufficiency (one-half the amount of CHD7 protein is made, but half isn't enough for normal development)

What does CHD7 do?



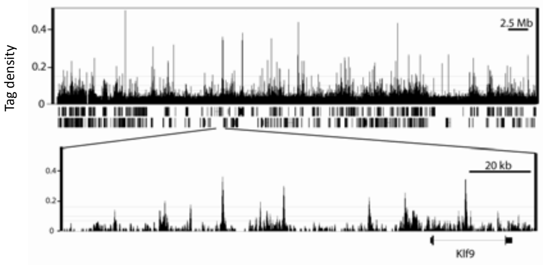
Model Systems for studying CHD7 function





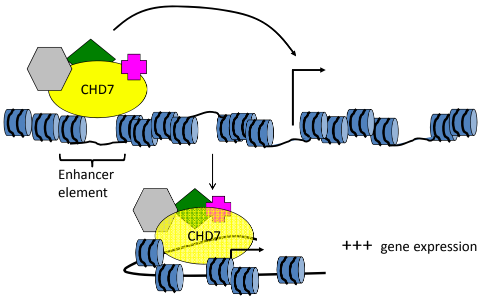
ChIP-seq – A method to find the sites on DNA where CHD7 binds

CHD7 ChIP-seq
(embryonic stem (ES) cells)



> 20,000 CHD7 binding sites across the genome

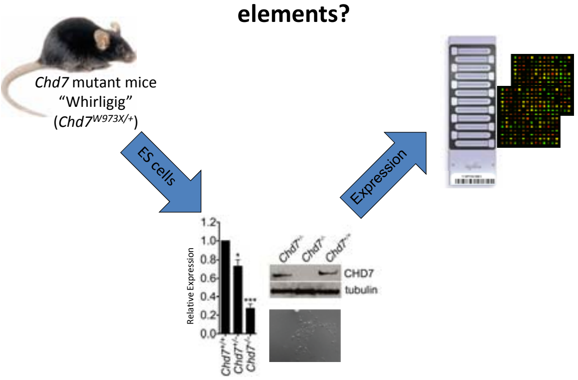
CHD7 binding sites are gene enhancer elements



+++ gene expression

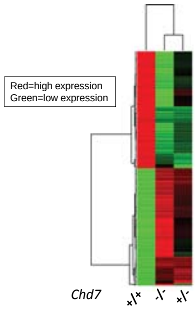
WHAT ARE THESE GENES?
WHICH CELL TYPE AND WHICH STAGE OF DEVELOPMENT ARE THESE GENES AFFECTED?

Is there a regulatory role for CHD7 at enhancer elements?



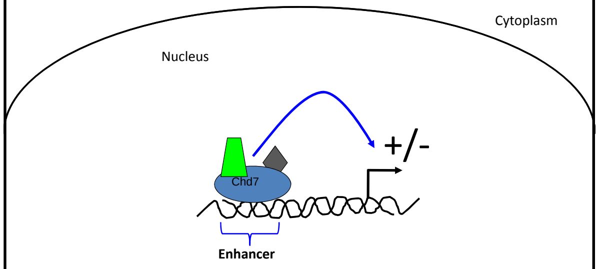
Chd7 mutant mice
“Whirligig”
(*Chd7*$^{W973X/+}$)

CHD7 Mutations Cause Changes in Gene Expression

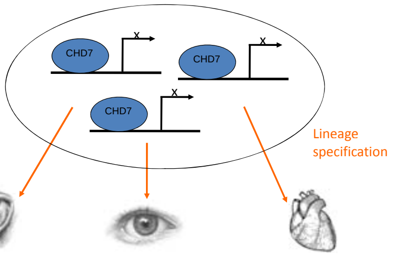


- •CHD7 binds to thousands of genes
- •Only about 10% have altered expression in ES cells
- •We think most CHD7 target genes probably change later in development in the eyes, ears, heart, and other tissues affected in CHARGE syndrome

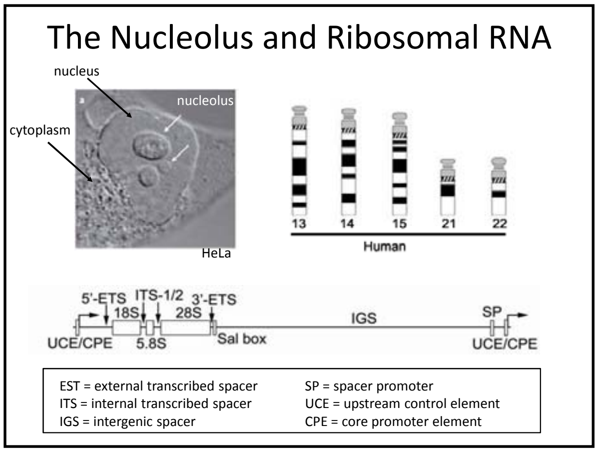
CHD7 as a regulator of tissue-specific genes



Working Model

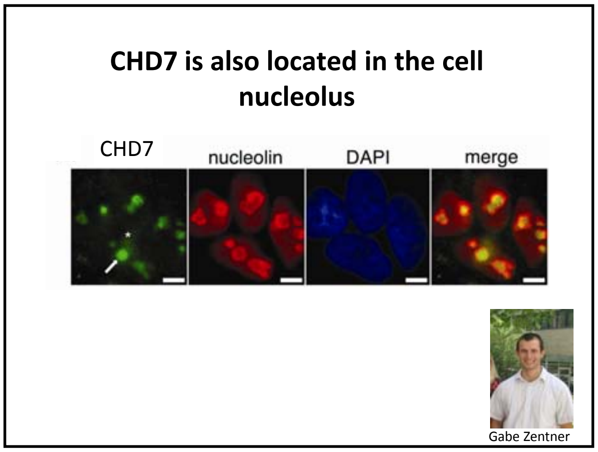


Misexpression of lineage specific genes during embryonic development, due to haploinsufficiency of CHD7, leads to CHARGE syndrome.



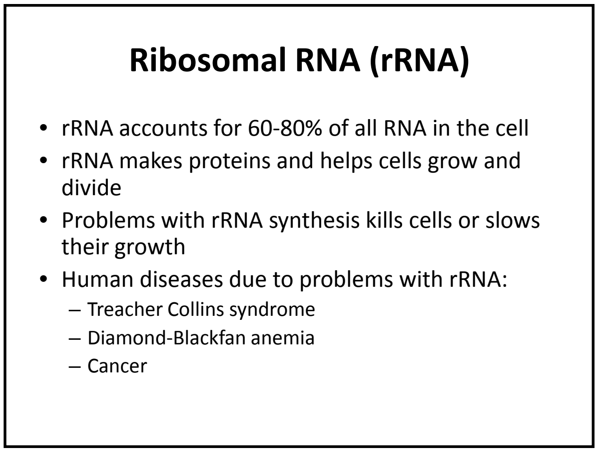
The Nucleolus and Ribosomal RNA

EST = external transcribed spacer
ITS = internal transcribed spacer
IGS = intergenic spacer
SP = spacer promoter
UCE = upstream control element
CPE = core promoter element



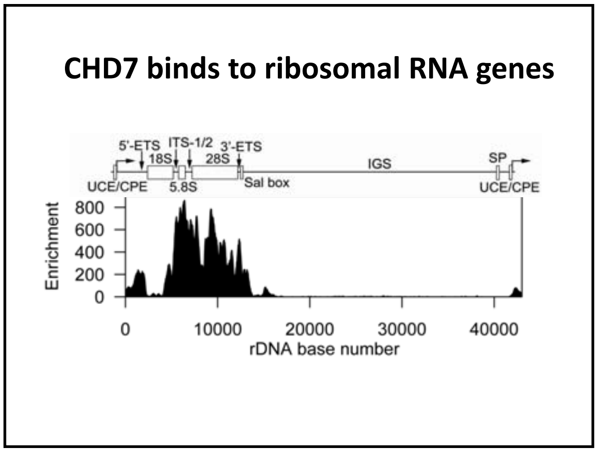
CHD7 is also located in the cell nucleolus

Gabe Zentner

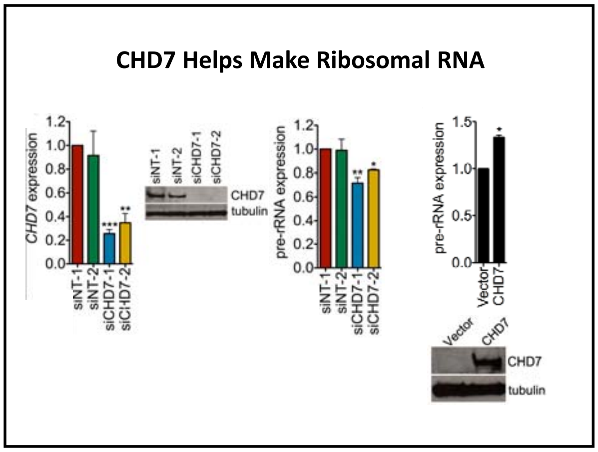


Ribosomal RNA (rRNA)

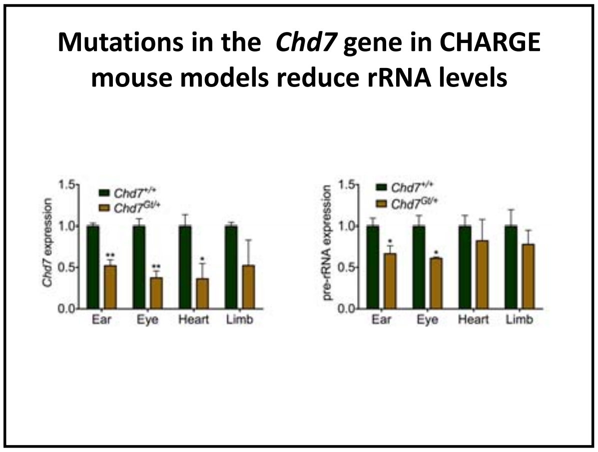
- rRNA accounts for 60-80% of all RNA in the cell
- rRNA makes proteins and helps cells grow and divide
- Problems with rRNA synthesis kills cells or slows their growth
- Human diseases due to problems with rRNA:
  - Treacher Collins syndrome
  - Diamond-Blackfan anemia
  - Cancer



CHD7 binds to ribosomal RNA genes



CHD7 Helps Make Ribosomal RNA



Mutations in the *Chd7* gene in CHARGE mouse models reduce rRNA levels

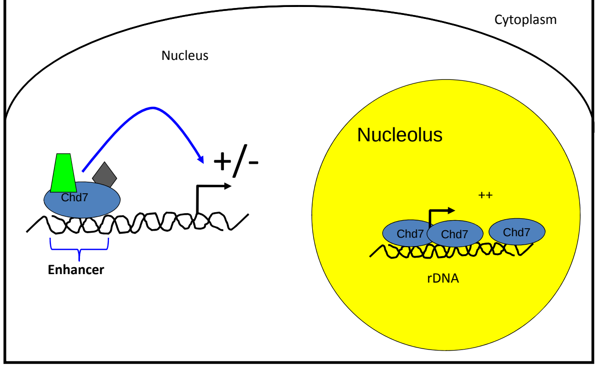
Problems with rRNA production can cause genetic diseases

Diamond-Blackfan anemia
- Mutations in RPS19, and other ribosomal genes
- Mostly sporadic dominant
- Likely haploinsufficiency
- Red blood cell aplasia, craniofacial, thumb, cardiac and urogenital abnormalities

Treacher Collins syndrome
- Mutations in TCOF1, encoding nucleolar treacle
- Autosomal Dominant
- Haploinsufficiency
- craniofacial abnormalities, including coloboma of the lid, micrognathia, microtia and other ear deformities. Conductive hearing loss, cleft palate

CHARGE Syndrome

Summary: Two functions for CHD7



Outline

- Overview of the cellular functions of CHD7
  - Regulator of genes that orchestrate development
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- Where we are headed

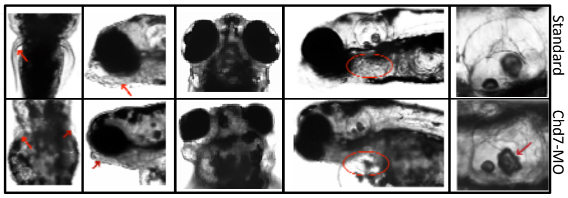
Zebrafish: A powerful model system

- Zebrafish is a vertebrate that shares the majority of genes with mammals, including CHD7
- Developmental processes are highly conserved between zebrafish and mammals
- Transparent embryogenesis that is also very rapid
  - Egg to embryo within 24 hours
- Targeted gene knockdown is feasible and straightforward (Morpholino (MO) technology)
- Low cost

Stephanie Balow

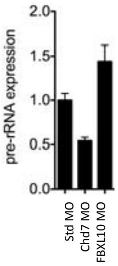
Chd7-MO phenotypes

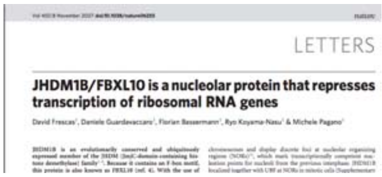
(Highly Dose Dependent)



Pectoral Fin Defects | Jawless | Ocular Edema, Lens Defects | Cardiac edema, Weak heartbeat | Deformed otoliths

Effects on rRNA biogenesis in zebrafish



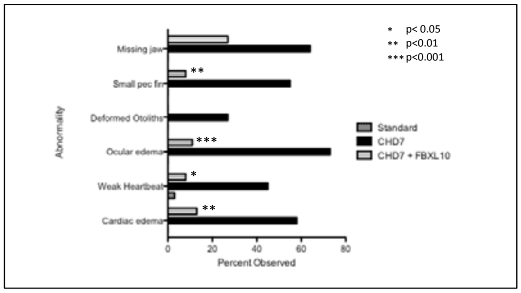


LETTERS

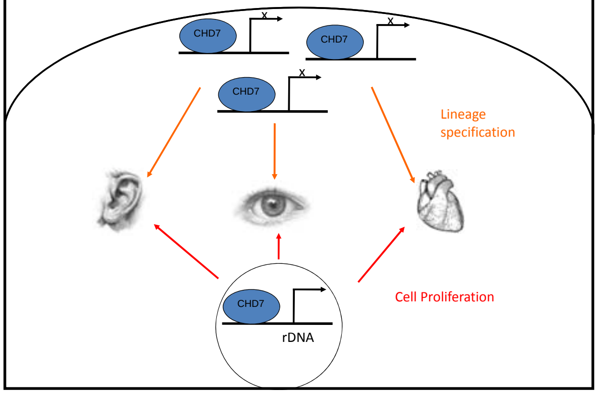
JHDM1B/FBXL10 is a nucleolar protein that represses transcription of ribosomal RNA genes

David Frescas[1], Daniele Guardavaccaro[1], Florian Bassermann[1], Ryo Koyama-Nasu[1] & Michele Pagano[1]

Rescue of CHARGE phenotype



Summary & Model for how haploinsufficency of CHD7 leads to CHARGE syndrome



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CHD7 gene sequence analysis that is fast and cheap


Illumina Genome Analyzer

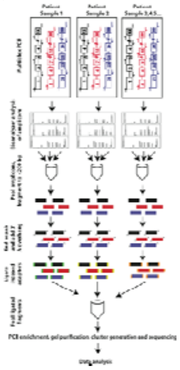
•CHD7 gene is big, mutation analysis is expensive through commercial labs.

•CHARGE syndrome shares clinical overlap with Kallmann syndrome, T-cell immunodeficiency, idiopathic scoliosis, and DiGeorge syndrome.

•Allows for testing in large patient cohorts

Cindy Bartels

Strategy



It works!

•We pooled ~50 patients with known mutations in CHD7 and ran them through our analysis. We were "blinded" to the location of the CHD7 mutation in all patients. Virtually all CHD7 mutations were detected!

•Also tested 80 patients with isolated coloboma.

•Major reduction in costs.

Summary

- CHD7 binds to thousands of gene enhancer elements
  - These enhancers regulate the genes that specify the tissues & organs that are affected in CHARGE syndrome.
- CHD7 controls the genes that are responsible for synthesizing proteins.
- The gene targets of CHD7 are dysregulated in other congenital disorders that show clinical overlap with CHARGE syndrome.
- CHD7 mutations can be identified in large patient cohorts relatively quickly and at low cost

Outline

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- Where we are headed

Where we are headed & what we need

- Clinical Samples (blood and skin biopsies)
  - – CHD7 mutation screening
  - – Generation of induced pluripotent stem cells (iPS cells).
    - Further molecular understanding of human CHD7.
- Additional Government funding for research

Acknowledgments

Scacheri Lab
Cindy Bartels, MS
Batool Akhtar-Zaidi
Michael Schnetz*
Gabe Zentner
Stephanie Balow
Olivia Corradin
Deb Schelling
Alina Saiakhova
Pavel Manaenkov*
Dheepa Balasubramanian, PhD
Lain Pierce, PhD*
*former member

CWRU
Paul Tesar, PhD
Tom LaFramboise, PhD
John Wang, PhD
Xiaodong Zhang, PhD
Maria Hatzoglou, PhD
Peter Harte, PhD

University of Michigan
Donna Martin, MD, PhD

Funding:
NICHD & NHGRI

Publications/Resources

- Review Article on CHARGE syndrome
  - – http://www.ncbi.nlm.nih.gov/pubmed/20186815
- CHD7 as an enhancer binding protein
  - – http://www.ncbi.nlm.nih.gov/pubmed/20657823
- CHD7 as a regulator of ribosomal genes
  - – http://www.ncbi.nlm.nih.gov/pubmed/21355038

THE CHARGE SYNDROME FOUNDATION

Toilet Training and CHARGE Syndrome

Saturday, 07/30/11
Breakout Session #6: 1:00-2:00
Panzacola F1 & F2

Laurie S. Denno, Ph.D. Candidate
Perkins School for the Blind
Simmons College

Presenter Information:

Laurie is a Ph.D. Candidate in Applied Behavior Analysis at Simmons College. She has more than 20 years experience working as a Behavior Analyst with children who have CHARGE syndrome at the Perkins School for the Blind, Deafblind Program. During her master's degree training, at Southern Illinois University in the Behavior Analysis and Therapy program, Laurie worked with Dr. Nathan Azrin, one of the foremost authorities on toilet training for typical children, children with developmental delays and adults with intellectual disabilities. Since then, Laurie has been involved in dozens of toilet training programs for children, adolescents and adults of many ability levels.

Presentation Abstract:

Toilet training is a rite of passage for many children and their families. It is a milestone in development and independence as well as a significant accomplishment for children and parents. While this process may be delayed in children with CHARGE syndrome due to sensory, physical and communication issues, many children can be and are toilet trained. This presentation will outline when children are ready to be toilet trained, what parents need to do to get ready for toilet training, including logistics, equipment and scheduling. A description of the basic toilet training procedure will be provided as well as special considerations for children with CHARGE syndrome. Parental responses to accidents and troubleshooting specific problems will be reviewed. This is an educational/behavioral teaching approach to toilet training. This presentation will provide a template for success, but individual children may require adjustments to the stand procedure.

Toilet Training for Children with CHARGE Syndrome

Laurie S. Denno, Ph.D. Candidate, BCBA
Simmons College
Boston, MA
Perkins School for the Blind
Watertown, MA
lauriedenno@yahoo.com

Approaches to Toilet Training

- There are two main approaches to toilet training: “wait until the child is ready”(Brazelton, 1962) and “rapid toilet training” approach (Foxx and Azrin, 1973).
- I am a proponent of Foxx and Azrin program because it is backed up by data in published research studies, it is a teaching approach, and I have used it myself many times.
- It is “rapid” because it is the primary focus of learning for a short but intensive time and other educational objectives are set aside.

Before You Begin

- Approximately 20-40% of children with CHARGE syndrome have urinary tract abnormalities (Issekutz, Graham, Prasad, Smith and Blake, 2005)
- Both structural and functional anomalies
- Need renal ultrasound to ID structural anomalies. Identify any physiological abnormalities.
- Need a voiding Cystourethrogram (VCUG) to evaluate if there is a functional anomaly such as reflux. (This is a voiding problem.)
- ***This is an absolute must before you begin!***

Some Basic Facts

- Most children regulate bowel movements before urination
- Children achieve daytime continence before overnight continence
- Toilet training to 100% success can take from 3-6 months
- Girls by 29 months. Boys by 31months.
- It is not unusual for children to be incontinent overnight until they are 6 years old. However, overnight continence is usually achieved within 12 months of day time continence.

Pre-requisite Skills

- Child should be 24-27 months old, with DD, 30 months
- Child should have periods of dryness of from 1-2 hours.
- The child should be able to sit for 5 minutes.
- The child should be able to follow one-step directions.
- The child needs to have some identified reinforcers.
- The child should have some communication skills around around toileting. (Pee, poop, T, etc.)

Helpful but Not Essential Skills

- The child can dress and undress.
- Bowel movements at a predictable time.
- The child displays some behavioral indications of having a urination or bowel movement in the diaper.
- Child does not like a wet or soiled diaper.
- The child displays imitation skills or wants to do things “by myself”
- The child can get to the bathroom independently and transfer to the toilet (if necessary).

Other Factors

- Select a good time to start toilet training-a low stress time when you can really attend to the project
- Be sure your child is healthy when you start
- Enlist support from all caretakers
- Coordinate between school, daycare and home

Plan In Advance

- What equipment do you need: potty chair, potty seat, seat back, urine guard? Your child needs to feel safe and secure on the toilet used with their feet firmly planted on a stable surface.
- Consider a seat belt.
- Ask an OT.
- Check: www.pottytrainingsolutions.com
- Decide when training will start

Get Ready

1. Take baseline data.
2. ID a frequently available bathroom
3. Purchase any special equipment.
4. Put all of your supplies in the bathroom you will be using (potty chair, extra clothes, wipes, books, toys, reinforcers, data sheet, etc.)

Get Set

- Talk to your child about the new and exciting toilet training program
- Reiterate “big boy” behavior, like dad and Johnny,
- Big boy underpants, earning treats and rewards
- Read (sign) the child social stories, www.pottytrainingsolutions has many good books or use drawings or M-J pictures
- You also can use dolls to model the correct toilet training behavior

GO

- It is best if you can give toilet training your undivided attention. Review your own schedule.
- Check the baseline data and determine how long the child can be dry.
- Once toilet training commences, take the child out of diapers for all waking hours.
- Introduce a toileting schedule that is slightly less than the dry interval.
- Have the child sit for about 5 minutes based on the schedule you have devised. It may be as often as 30 minutes or as little as 90 minutes.

- If your child is cooperative with sitting on the toilet, increase the time to 10 minutes.
- If the child is fidgety, read to the child, have the child look at a book or play with a sensory toy.
- Do not “make” the child sit against resistance. This will make toileting aversive and prevent success. This process needs to be pleasant, fun and an educational time, not a power struggle.
- Continue to take data on successes and accidents. This information will help you make informed decisions about scheduling, etc.

The First Success

- Your child may sit on the toilet many, many times before there is a success.
- However, when there is a success, reinforcement is critical in increasing the frequency of urination in the toilet.
- Be prepared! You may need hugs, praise, or other items (food, activities) to encourage further successes.
- Reinforcement needs to be immediate (.5 seconds after the behavior), dramatic and special. <u>Effective reinforcement is the most critical variable in toilet training.</u>

- Whether or not the child is successful on the toilet, have him practice the whole toileting sequence: going to the bathroom, pulling down pants, sitting, wiping, pulling up pants, flushing, washing hands, etc.
- Offer positive feedback for following the sequence on every occasion.
- Cleanliness after a bowel movement is important. Parents usually wipe young children but children need to be taught to wipe. Use hand over hand assistance, modeling and verbal prompts until the child masters the skill. Flushable wipes can be helpful during the initial training

I

- If your child asks to go to the toilet at any time, please take him. It is not the schedule we are going for but urinating in the toilet.
- If the child displays any behavioral indications that he needs to urinate (little dance, squatting, leaning over furniture), take him to the bathroom
- When you child is having no more than one accident per day, increase the intervals between trips by brief (15 minutes) increases

- Also, you can start taking your child to other bathrooms, at the library, at rest stops, at the Dr.'s office. This promotes generalization.
- Attempt to fade reinforcement to praise only or use a token system, stars, checks, smiley faces, etc.

Accidents

- Accidents are inevitable. State the facts. "You are wet. You need to change."
- Have the child go to the bathroom and sit on the toilet for a brief time. (Probably does not need to go because he just went.)
- Have the child follow the entire bathroom procedure. However, have the child change on a separate chair or stool, not the toilet. The toilet is where you urinate, not where you change.
- The child should place the wet clothing in the designated place (diaper pail, dirty clothes basket, etc.)

- There should be no reprimands, no negative feedback.
- <u>Remain neutral. State the facts. Be calm.</u>
- Remember this is an "accident". Telling your child about toilet training is not the same as teaching the child to urinate in the toilet.
- Bowel and bladder control is regulated by a complex set of muscles, some that must be developed over time to ensure successful holding and releasing

Special Considerations for CHARGE Syndrome

- Micropenis
- Boys should sit
- Vision impairments require more "hands on" assistance because they do not learn by watching. Use regular O and M strategies. Fade "hands on" ASAP.
- Light, sound and shiny surface can be over-stimulating leading to flapping, finger flicking or light gazing.
- Prompt to use hands-on activities

- Modify the environment by dimming lights, removing shiny items, or adding towels or rugs to muffle bathroom echo
- Cover the toilet with a blanket before flushing if the child is afraid of the sound
- Balance and low tone can interfere with sitting and producing
  - Use a seat belt, seat back, potty chair with arms, regular sized toilet arms, foot stool, foot rest
  - Integration of responses can take longer than in typically developing children, therefore, practice, be patient, be positive

Troubleshooting

- <u>Child is afraid of the toilet or potty chair</u>: approach gradually, sit for short periods, hang out in the bathroom
- <u>Child is afraid of flushing</u>: have the child "fade into" listening, from afar, the doorway, the sink, standing next to, etc.
- <u>No urination on the toilet after 20 hours of training</u>: consider increasing fluids to increase need to urinate. Discuss how much and what with your pediatrician.

- <u>Child masturbates while seated</u>: direct hands to thighs, toilet grab bar or offer a hands-on object
- <u>Child absolutely refuses to participate:</u> stop training immediately and wait a month or two. Evaluate your procedures and start slow with a lot of praise, fun and reinforcers.
- <u>Child "withholds" urine or bowel movements, there is constipation, loose stools or accidents "on purpose"</u>: first consult your pediatrician. Second, consult a behavior analyst (www.BACB.com) to assist you in doing a functional assessment of the "on purpose" accidents. Change your response to these accidents

Summary

- Take baseline
- Gather supplies
- Start with a plan
- Implement the plan
- Adjust the plan
- Increase intervals
- Fade reinforcement
- Get help if you need it

GOOD LUCK!!

Resources

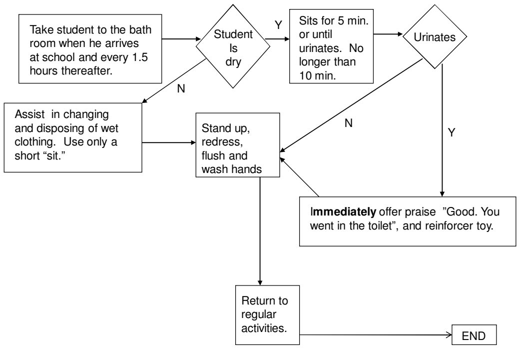
Azrin, N. H., & Foxx, R. M., (1974). *Toilet training in less than a day*. New York, NY: Simon and Shuster.

Brazelton, T. B., (1962). A child-oriented approach to toilet training. *Pediatrics, 29*, 121-129.

www.bacb.com for a behavior analyst

www.pottytrainingsolutions.com for books, equipment supplies and underwear

Instructional Goal: Student will urinate/defecate in the toilet



Take student to the bath room when he arrives at school and every 1.5 hours thereafter.

Student Is dry

Y

Sits for 5 min. or until urinates. No longer than 10 min.

Urinates

N

Assist in changing and disposing of wet clothing. Use only a short “sit.”

N

Stand up, redress, flush and wash hands

Y

Immediately offer praise ”Good. You went in the toilet”, and reinforcer toy.

Return to regular activities.

END

5/12/11 Laurie S. Denno©

THE CHARGE SYNDROME FOUNDATION

Language Acquisition through Motor Planning (LAMP) – Part 2

Saturday 07/30/11
Breakout Session #6: 10:45 – 11:45
Panzacola F3 & F4

John Halloran M.S.CCC-SLP
Center for AAC and 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. He also finds much reward in exploring ways to best implement assistive technology with children who have autism. John Halloran graduated from the University of Arkansas at Little Rock in 1990 with a bachelor's degree in Communication Disorders. He 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 is the primary developer of Language Acquisition through Motor Planning(LAMP) and presents internationally. John maintains membership in several professional organizations, including the American Speech Language Hearing Association and the Arkansas Speech Language Hearing Association.

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 developmental disabilities a method to develop independent and spontaneous communication. Strategies to teach language/communication skills within this framework will be discussed.

Growth/Puberty, Part 1

Smell and puberty in CHARGE syndrome; a smell test is able to predict whether puberty will occur spontaneously

Saturday, 07/30/11
Breakout Session #6: 1:00-2:00
Panzacola G1

Prof. Conny van Ravenswaaij-Arts
Dept. of Genetics, University Medical Centre Groningen, Groningen, The Netherlands

Presenter Information:

Conny van Ravenswaaij studied medicine at the University of Leiden. In 1997 she was registered as a clinical geneticist. Her main interest has always been children with multiple congenital anomalies. Her group discovered the CHD7 gene as major cause of CHARGE syndrome in 2004. In 2006 she changed affiliation to the University Medical Centre Groningen, where she continued her multi-disciplinary outpatient clinic for CHARGE syndrome. She supervises studies in CHARGE syndrome, focusing on clinical variability and phenotype-genotype correlations, puberty development and smell, the role of CHD7 in heart development, Cochlear Implants and other aspects of CHARGE syndrome.

Presentation Abstract:

Smell deficiency and delayed or absent puberty often occur in CHARGE syndrome, but few studies have looked at these features in adolescent patients. Therefore, we studied smell and puberty development in 35 adolescent patients with CHARGE and showed that puberty and smell problems always co-occur. Because sense of smell and pubertal development are always correlated, a smell test can predict pubertal development. When a patient with CHARGE has absent sense of smell, he/she will probably not enter puberty spontaneously. This makes it possible to start with hormone replacement therapy earlier, leading to age-appropriate puberty. This will reduce social problems and osteoporosis risk.

Puberty and smell in CHARGE syndrome

Conny van Ravenswaaij-Arts, Jorieke van Kammen-Bergman

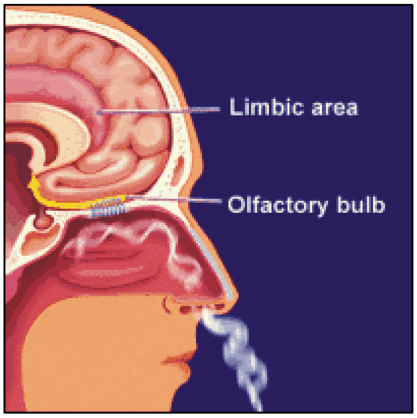
In my presentation I will focus on two aspects of CHARGE syndrome: sense of smell and puberty. First, I will talk about sense of smell and puberty in general and then I will show the results of my study in which I wanted to answer 3 questions:

- How often does a smell deficit occur in CHARGE syndrome?
- How often do individuals with CHARGE syndrome have delayed or absent puberty?
- Are smell and puberty always correlated in CHARGE syndrome?

The third question is very relevant, because if a smell deficit is always seen in association with delayed puberty, one can use a smell test to predict whether spontaneous puberty will occur.

How do we smell?

We smell with our nose. Volatile odour molecules can enter the nostrils and will go to the top of the nasal passages to a patch of special neurons about as big as a postage stamp. These neurons have hair-like projections that can bind to specific odour molecules. When an odour molecule is bound, the neuron will be trigged and sends a signal to the brain via the olfactory nerve. The olfactory bulb, located in the frontal part of the brain, is the brain region that is in involved in olfaction.



How does our body know when it is time to enter puberty?

Our brain will direct the timing of puberty. A specialized area of the brain, called the hypothalamus, will start to produce gonadotropin releasing hormone (GnRH). GnRH will in turn activate another region of the brain, called the pituitary gland, to secrete luteinizing hormone (LH) and follicle stimulating hormone (FSH) into the central blood circulation. In response to LH and FSH, the ovaries will start to make estrogens in girls and the testicles will make testosterone in boys. The rising levels of estrogens in girls and testosterone in boys will start pubertal development. Girls enter puberty around the age of 8 – 13 years, whereas boys enter puberty around 9 – 14 years of age. The fist sign of puberty is breast development in girls and growth of the testicles in boys.

Why do we think smell and puberty could be related?

The areas in the brain that are involved in smell and puberty develop together during embryonic development. Therefore, a smell deficit is often seen in association with a delay in pubertal development, for example in persons with Kallmann syndrome.

Study on smell and puberty in CHARGE syndrome

Smell deficiency and delayed or absent puberty often occur in CHARGE syndrome, but few studies have looked if these features are associated in adolescents with CHARGE syndrome. Therefore, we studied smell and pubertal development in 35 individuals with CHARGE syndrome from the Netherlands. In this study, we included 19 boys and 16 girls aged 10 years or older who all had a mutation in the *CHD7*-gene. We performed a smell test (the University of Pennsylvania Smell Identification Test, see the picture below) in all persons without mental retardation, bilateral choanal atresia and/or severe feeding difficulties (26/35). Also, we re-analyzed MRI brain scans (whenever available) for abnormalities of the olfactory bulbs (the area in the brain involved in olfaction). Pubertal development was evaluated by a paediatric endocrinologist who did a physical exam and measured hormone levels in blood.

5. This odor smells most like	5	4	3	2	1
a. skunk	a	a	a	a	a
b. coconut	b	b	b	b	b
c. cedar	c	c	c	c	c
d. honey	d	d	d	d	d

How often does a smell deficit occur in CHARGE syndrome?

Smell testing showed absent sense of smell in 21/26 (81%) individuals and normal or slightly decreased sense of smell in 5/26 (19%) individuals. History taking was not reliable for determining sense of smell. MRI brain scans were available in 10 persons, but could be analysed for olfactory bulb abnormalities in only three persons. These three persons all had abnormal olfactory bulbs.

How often do individuals with CHARGE syndrome have delayed or absent puberty?

23 Individuals were old enough to distinguish between delayed or normal puberty. In 17 persons puberty was delayed or absent (74%), whereas 6 persons had experienced normal puberty (26%).

Was there a correlation between sense of smell and pubertal development?

From 15 individuals complete data on both smell and puberty were available: 11 persons had both a smell deficit and delayed puberty and 4 persons had normal sense of smell in combination with normal pubertal development. Seven boys were too young to know if they would enter puberty at a normal age, but they all had cryptorchidism or a micropenis, which is suggestive for delayed puberty. These seven boys had no sense of smell. Therefore, a total of 22 persons showed concordance between smell and (suspected) pubertal development. We conclude that smell and pubertal development are 100% correlated in this study.

Can a smell test predict whether spontaneous puberty will occur?

Because of the correlation between sense of smell and pubertal development, a smell test can probably predict whether spontaneous puberty will occur. When a patient with CHARGE syndrome is unable to smell, he/she will probably need hormone replacement therapy to enter puberty. We recommend timely start of hormone replacement therapy in children with CHARGE syndrome who have no sense of smell to make sure they enter puberty simultaneously with their peers. This will reduce social problems and risk of osteoporosis (brittle bone disease).

Reference

Bergman JEH, Bocca G, Hoefsloot LH, Meiners LC, van Ravenswaaij-Arts CMA. Anosmia predicts hypogonadotropic hypogonadism in CHARGE syndrome. *J Pediatr* 2011;158(3):474-9.

Acknowledgements

We thank all the persons with CHARGE syndrome and their parents for participating in this study, and we are grateful to the Dutch control subjects who took the olfaction test. In addition, we thank A. Schoenmaker and E. Tiems, Kentalis, Sint Michielsgestel; S.L.S. Drop, Dept. Pediatric Endocrinology, Erasmus Medical Center, Rotterdam; M.C.J. Jongmans, Dept. Human Genetics, and H. Claahsen-van der Grinten, Dept. Pediatric Endocrinology, Radboud University Nijmegen Medical Center, for referring patients to the CHARGE outpatient clinic. Finally, we thank the Netherlands Organisation for Health Research (ZonMW 92003460) for financial support.

THE CHARGE SYNDROME FOUNDATION

Using Person-Centered Planning to Enhance Transition from School to Adult Life.

Saturday 07/30/11
Breakout Session #6 1:00 – 2:00
Panzacola G2

David Wiley, Transition Specialist,
Texas Deafblind Project, Austin, TX

Presenter Information:

Since 1989 David Wiley has worked with the Texas Deafblind Project, providing statewide technical assistance to families, schools, and community service agencies around issues of deafblindness and transition planning. Before joining the Project, he worked with students in the Deafblind Program at the Texas School for the Blind and Visually Impaired. He serves as co-chair of the Texas Interagency Task Force on Deafblindness, and was formerly on the Steering Committee of the Texas Transition Task Force. His work includes providing consultation on person-centered planning for individual students, writing training materials, serving as editor of the Project publication, *TX SenseAbilities*, and organizing workshops and conferences. He has co-authored a training curriculum for direct-care workers providing services through the Texas Medicaid Waiver for Persons who are Deafblind with Multiple Disabilities. For over 20 years David has made numerous workshop presentations to families and professionals throughout Texas, as well as around the U.S. and in Canada.

Presentation Abstract:

Planning satisfying and meaningful lives for students with sensory impairments, including those with additional disabilities, must start with concrete future goals. Person-centered planning strategies help students, families, educators, and community service providers discover and clarify these goals. Developing concrete goals for the future, learning skills for adult life, and arranging appropriate support after the school years will be discussed.

Using Person-Centered Planning to Enhance Transition from School to Adult Life.

David C. Wiley
Texas Deafblind Project
110 W. 45th Street
Austin, TX 78757
davidwiley@tsbvi.edu

During the teen years…

…students with deafblindness should be preparing for adult life by discovering skills and interests, becoming more independent, and making plans for the future.

However, many have led…

…sheltered lives with limited expectations. Many students have few opportunities to take on responsibilities and make meaningful decisions. This presentation will cover how to prepare students for transition to adult life by creating opportunities for independence and self-direction.

During transition planning…

…students are expected to answer difficult questions about what they want for their future lives.

IDEA 2004

Transition Services. Beginning not later than the first IEP to be in effect when the student turns 16, or younger if determined appropriate by the IEP Team, and updated annually, thereafter the IEP must include appropriate measurable postsecondary goals based upon age appropriate transition assessments related to training/education, employment, and where appropriate independent living skills (§ 300.320(b)(1).

State Performance Plan – Indicator 13
(Transition services for students age 16 – 21)

Percent of youth aged 16 and above with an IEP that includes coordinated, measurable, annual IEP goals and transition services that will reasonably enable the child to meet the post-secondary goals. [20 U.S.C. 1416(a)(3)(B)]

From NICHCY:
http://nichcy.org/laws/idea/partb/indicators-partb/indicator13

Life Design: Planning and Goal Setting

This class will focus on developing a long-range "vision" of the future and then work "backwards" from this vision to the present—planning specific events and long-range, short-range, and micro-range goals to get "from here to there"…

"Classes to Improve Yourself" in *Wellness*, a newsletter from Seton hospital in Austin

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Career, Life Planning

Do You want to "have it all" but are unsure what "it" is or how to get it? This two-part class will help you determine your focus and design an action plan to reach your goals.

"Wellness Classes" in *Health Source* from Brackenridge Hospital in Austin

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Dream, Achieve, Excel: Realize Your Potential

Basic goal setting from wants and needs is presented and then expanded to include dreaming and visualization techniques that will move goal-setters to their greatest potential.

"Classes to Improve Yourself" in *Wellness*, a newsletter from Seton hospital in Austin

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State Performance Plan – Indicator **14**

Post-school outcomes

Percent of youth who had IEPs, are no longer in secondary school and who have been competitively employed, enrolled in some type of postsecondary school, or both, within one year of leaving high school. [20 U.S.C. 1416(a)(3)(B)]

From NICHCY:
http://nichcy.org/laws/idea/partb/indicators-partb/indicator14

Your Definition of Success:

- There is more to life than work.
- Purpose, Satisfaction, and Joy.
- If your child took control of his or her life, how would he or she describe a successful life?
- Is the student ready to make this determination?
- How do we know what the student chooses as success?

Your Definition of Success

?

Person-Centered Planning

… replaces, supplements, or enhances traditional System-Centered Approaches

Opportunities for Decision-making and Risk-taking

- Avoid “learned helplessness.”
- Accept that growth involves risk.
- Consider that decision making means more than making simple choices.

Decision-making includes:

1. Accepting responsibility
2. Understanding options based on experience
3. Taking a long-term perspective
4. Considering resources
5. Considering how decisions affect others
6. Being a self-advocate

1. Accepting Responsibility

- A first step toward self-determination is taking on responsibilities.
- A responsibility is something more than a chore.
- Responsibility involves understanding and investment.

2. Understanding options based on experience

- Information deficits related to deafblindness may prevent students from fully understanding even common events and situations.
- This leads to mysteries for students.

To the students, many things in life are a mystery…

…but students need to know what happens behind the curtain!

Where would you rather live?

- Uganda or Madagascar?
- Central City or Suburbs?
- House or Apartment?

Do you have enough information to decide?
Have you made a decision like that before?

---

3. Taking a long-term perspective

4. Considering resources

5. Considering how decisions affect others

---

6. Being a self-advocate

- Knowledge
- Communication
- Structure and routine

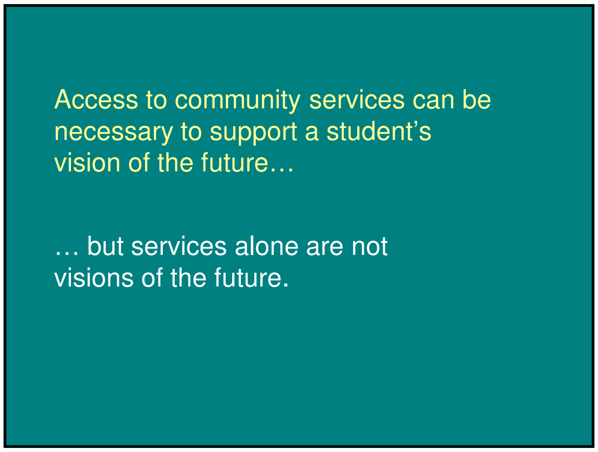
A clear, shared vision of the future is often easier said than done for students with deafblindness, their families, and service providers…

… but without it, transition planning is isn't meaningful.

If there isn't a clear vision of the future for the student, the priority becomes finding a vision that everyone involved can share.

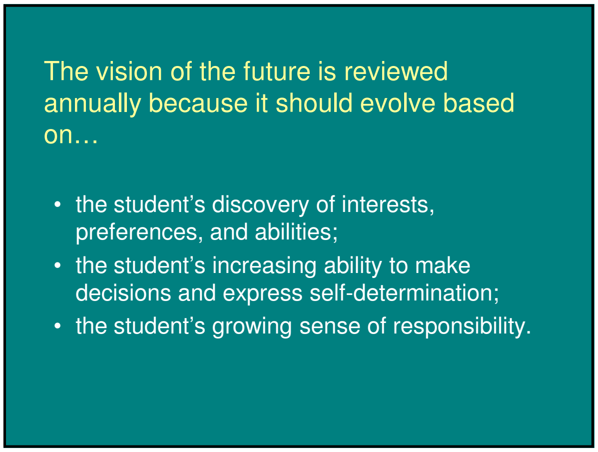
What a student learns at school can help the student develop a vision of the future, and prepare the student to take steps toward bringing the vision to reality…

… but courses and curricula are not visions of the future.



Access to community services can be necessary to support a student’s vision of the future…

… but services alone are not visions of the future.



The vision of the future is reviewed annually because it should evolve based on…

- the student’s discovery of interests, preferences, and abilities;
- the student’s increasing ability to make decisions and express self-determination;
- the student’s growing sense of responsibility.

National Resources on Transition Planning

- *National Secondary Transition Technical Assistance Center* (NSTTAC) www.nsttac.org/
- *National Center on Secondary Education and Transition (NCSET)* www.ncset.org
- *Helen Keller National Center (HKNC)* www.hknc.org
- *Postsecondary Education Programs Network (PEPNet)* www.pepnet.org
- *National Consortium on Deaf-Blindness (NCDB)* www.nationaldb.org

THE
CHARGE
SYNDROME
FOUNDATION

Feeling Good?

Saturday, 07/30/11
Breakout Session #6: 1:00-2:00
Wekiwa 3 & 4

Gail Deuce M.Ed
Principal MSI Consultant
Children's Specialist Services
Sense UK

Presenter Information:

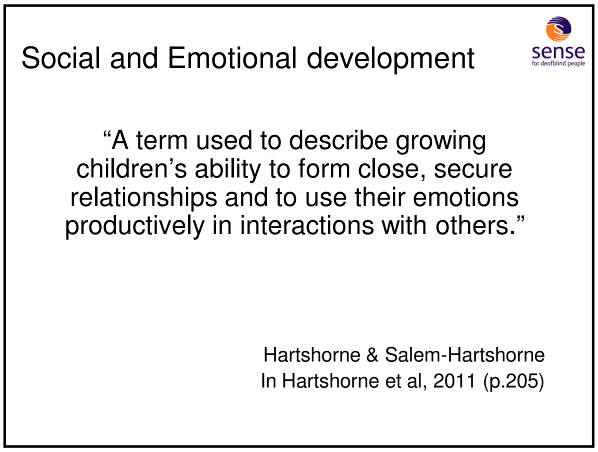
Gail has a B.Ed in Special Education, is a qualified teacher of the deaf and also has a M.Ed. in Multi-sensory Impairment. Gail works in the UK and has over twenty-five years experience in the field of special education, working initially in schools for children with severe learning difficulties and then a school for the deaf before moving into peripatetic work focusing on learners who are deafblind. Gail worked for different local authorities before joining Sense in December 2001. She works in the Children's Specialist Services as a Principal MSI Consultant. Gail has a particular interest in CHARGE and is on the committee for the CHARGE Family Support Group in the UK. She is currently undertaking a PhD, focusing on the educational environment for children with CHARGE syndrome. Email: gail.deuce@sense.org.uk

Presentation Abstract:

It is recognized that many young people with CHARGE experience social and emotional difficulties for a number of possible reasons. This presentation will provide a summary of the social and emotional difficulties observed in young people with CHARGE with illustrations from the presenter's own experience of working with young people with CHARGE. This will be supported by a case study of a young person with CHARGE syndrome, outlining the difficulties she encountered and strategies employed to help support her emotional development and social interactional skills.

Feeling Good?
Social and Emotional Development in Young People with CHARGE

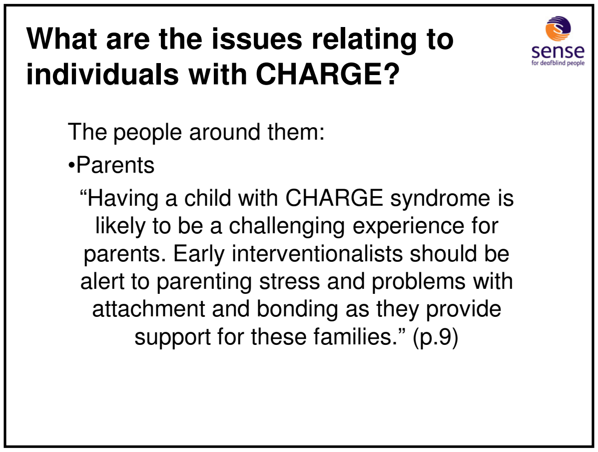
Gail Deuce
Principal MSI Consultant
Children's Specialist Services
Sense, UK



Social and Emotional development

"A term used to describe growing children's ability to form close, secure relationships and to use their emotions productively in interactions with others."

Hartshorne & Salem-Hartshorne
In Hartshorne et al, 2011 (p.205)

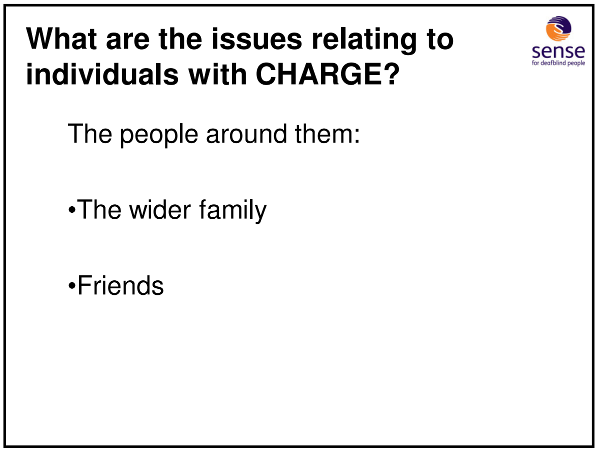


What are the issues relating to individuals with CHARGE?

The people around them:

•Parents

"Having a child with CHARGE syndrome is likely to be a challenging experience for parents. Early interventionalists should be alert to parenting stress and problems with attachment and bonding as they provide support for these families." (p.9)

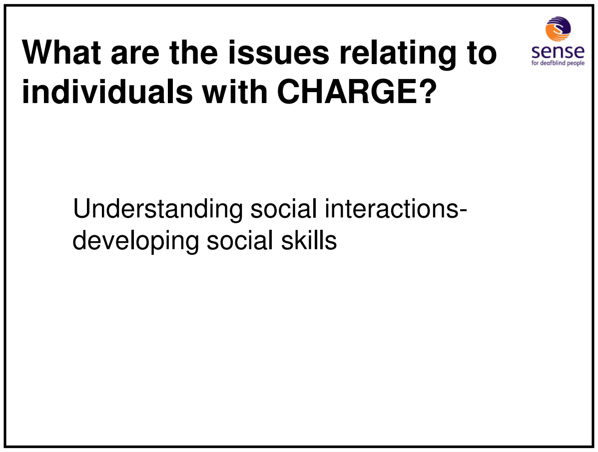


What are the issues relating to individuals with CHARGE?

The people around them:

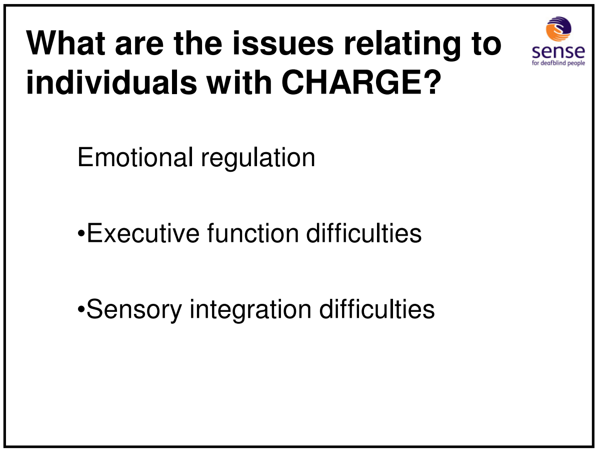
•The wider family

•Friends



What are the issues relating to individuals with CHARGE?

Understanding social interactions- developing social skills



What are the issues relating to individuals with CHARGE?

Emotional regulation

•Executive function difficulties

•Sensory integration difficulties

Case study- Jenny

THE CHARGE SYNDROME FOUNDATION

Balance Group: Why Does It Work?

Saturday, 07/30/11
Breakout Session #6: 1:00-2:00
Wekiwa 5

Maryann M. Girardi, PT, DPT, ATP
Physical Therapist
Perkins School for the Blind
Deafblind Program

Presenter Information:

Ms. Girardi is a physical therapist in the Deafblind Program at Perkins School for the Blind. She has evaluated and worked with children with CHARGE syndrome for over 10 years. During this time she and her colleague have developed a program of activities to facilitate the development of static and dynamic balance.

Presentation Abstract:

In this presentation the mechanisms of balance and motor learning will be discussed in relation to the acquisition of static and dynamic balance for children with CHARGE syndrome. This will include a basic review of human balance system and the challenges these students can have within that system. Theories of motor learning will be applied to the activities used in Balance Group to explain how the group is able to facilitate the development of balance and gross motor skills.

Balance Group: Why Does It Work?

Maryann M. Girardi, PT, DPT, ATP
Perkins School for the Blind
Watertown,MA

Components of Balance

- Sensory System
- Visual System
- Vestibular System
- Motor System
- Central Nervous System Integration

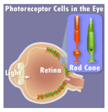
Sensory System

- Somatosensory
  - The system that detects experiences labeled as touch or pressure, temperature, pain and proprioception

Somatosensory Receptors

- Skin Receptors
  - Touch
  - Pressure
- Proprioceptive Receptors
  - Muscle Spindles-located in the muscle belly to detect muscle length
  - Golgi Tendon Organs-located in the tendons to detect muscle tension
- Joint Receptors- located in the joint capsule to detect joint pressure and position

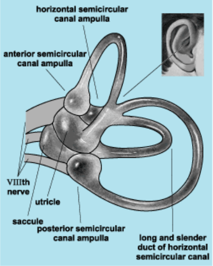
Visual System



- Receptors
  - Retina
    - Cones- central vision, acuity, high light
    - Rods- peripheral vision, low light, low threshold
- Detects orientation in space
- Provides environmental information

www.learn.genetics.utah.edu

Vestibular System



- Semicircular canals (angular acceleration)
  - Positioned in 3 different planes to detect up/down, side to side and tilting movement of the head
- Otoliths (linear acceleration)
  - Utricle- horizontal plane (back and forth)
  - Saccule- vertical plane (up and down)
  - Maculae (located inside the otoliths) - head position in space

www.encyclopeida2.thefreedictionary.com/vestibular+apparatus

Vestibular Reflexes

- Ventibulo-ocular reflex
  - Ensures best vision during head movement by moving eyes contrary to the head to maintain line of sight
- Vestibulo-spinal reflex
  - Stabilizes the body
- Vestibulo-collic refles
  - Stabilizes the head

Righting Reactions

(Midbrain level reflexes)

- Head on Body/Body on Body
- Labyrinthine Righting on Head
- Optical Righting
- Parachute

Equilibrium Reactions

(Cortical Level Reflexes)

- Tilting- Prone,Supine, Quadruped,Sitting and Standing
- Protective Extension- Forwards, Backwards, and Sidewards
- Postural Fixation- Prone, Supine, Quadruped, Sitting and Standing
- Protective Stepping

Motor System

- Range of Motion
  - Amount of movement at each joint
    - Excessive range = lack of stability
    - Limited range = decreased movement, misalignment
- Motor Function
  - Adequate motor skill development
  - Ability of muscles to work -tone, strength, paralysis
  - Ability to perform controled/coordinated movement

Central Nervous System

- Brain Stem
  - Visual reflex center
  - Houses motor pathways from the brain to the body
- Cerebellum
  - Processes information from sensory and motor pathways to provide coordinated muscle movement
  - May contribute information about automatic movements that have been learned through practice
- Cerebral Cortex
  - Contributes previously learned information (ice is slippery)

Central Nervous System

- Integrates the information from other systems
- Processes the information from the systems and determines what is the most accurate
- Sends directions to the body for motor output to maintain balance

Balance Reactions

- Ankle Strategy
  - Movement to maintain balance occurs at the ankle joint in the opposite direction of the challenge
- Hip Strategy
  - Movement to maintain balance occurs at the hip joint in the opposite direction of the challenge
- Step Strategy
  - To maintain balance a step is taken in the direction of the challenge

Definitions

Motor Control- The ability to regulate or direct the mechanisms essential to movement (Shumway-Cook, Woolacott 2001)

- Motor Learning -A set of processes associated with the practice or experience leading to relatively permanent changes in the capacity for producing skilled action (Shumway-Cook &Woolacot 2001)
- Motor Program- An abstract representation of movement that centrally organizes and controls the many degrees of freedom involved in performing an action

Theories

- Open Loop
  - Attention required only to initiate movement
- Closed Loop
  - Uses feedback to control movement
- Schema Theory
  - General rules that can be applied to various situations
- Ecological Theory (Task Oriented Approach)
  - All movement and actions are influenced or constrained by the environment.

- Systems Theory
  - Includes consideration of client,occupation and environmental factors
- Multiple paired forward & inverse models
  - A computational process of selecting a motor command to achieve a desired sensory feedback
- Non-Associative Learning
  - Habituation sentization
- Associative Learning
  - Classical and operant conditioning

Accommodations

- Communication
- Clear expectations
- Enhanced Visual information
- Maximal Assistance for success
- Activity based
- Provide Feedback

Why does it work?

- Activity used are enjoyable and appropriate
- Task Oriented Approach
- Assistance is given at the level which the child will be able to successfully complete the task
- Appropriate movement is learned through repetition
- Practiced with variations including environment, assistance and equipment
- Feedback is provided both with and without language
- Use of all systems
- Fun

Activities

References

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www.dizziness-and-balance.com
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THE
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Children with CHARGE A Longitudinal Study 1993 -2010 – A Film

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Panzacola F1 & F2

Rob Last
Early Childhood Educator CHARGE Syndrome Association of Australasia

Presenter Information:

Rob Last is a teacher who has a long history of working with children who have sensory disabilities, particularly in the areas of hearing and vision.
Since the mid 1980's he has been particularly involved with children who have CHARGE syndrome and their families. He has been a presenter at the CHARGE Syndrome International Conferences since 1993 and at the Australian and New Zealand Conferences since 1994.
Rob is a director of the CHARGE Syndrome Association of Australasia parent support group.

Presentation Abstract:

In 1993 I filmed a number of children who had been diagnosed with CHARGE syndrome. At that time I focused on the communication systems they were using and their gross motor development. I then followed their progress by revisiting them in 2000 and 2007, continuing to focus on communication and gross motor development, as well as broadening the focus to health, growth, education, development and social issues. In the film we see them as children, teenagers and adults. The result is a film depicting ten young people living with CHARGE syndrome and their progress. Excerpts from the film will be shown as part of this presentation.

Children with CHARGE
A longitudinal study
1988 – 2010
A film by Rob Last

In 1993 I filmed a number of children who had been diagnosed with CHARGE Syndrome. At that time I focussed on the communication systems they were using and their gross motor development.

I then followed their progress by revisiting them in 2000 and 2007, continuing to focus on communication and gross motor development, as well as broadening the focus to health, growth, education, development and social issues.

Further follow up information was also obtained in 2010.

In this presentation excerpts from the film will be shown.

A complete copy of the film is available from Rob Last.
Email: roblast@iinet.net.au

Excerpts from the film may also be viewed on the
CHARGE Syndrome Association of Australasia website:
www.chargesyndrome.org.au

Rob Last

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Can Person Centered Planning Work with Younger Individuals with CHARGE?

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Panzacola F3 & F4

Nancy Salem-Hartshorne, Ph.D.
Delta College

Gail Deuce M.Ed
Children's Specialist Services
Sense UK

Presenter Information:

Gail is a principal MSI Consultant with the Children's Specialist Services, Sense UK. Gail has worked in the field of special education for 25 years, working in different local authorities before joining Sense in December 2001. Gail has a particular interest in CHARGE and is on the committee for the CHARGE Family Support Group in the UK. She is currently undertaking a PhD, focusing on the educational environment for children with CHARGE syndrome. Email: gail.deuce@sense.org.uk

Dr. Nancy Salem-Hartshorne is an instructor at Delta College located in central Michigan. Her research has focused on developmental outcomes for individuals with CHARGE syndrome. Her published work has focused on cognitive and adaptive behavior skills in CHARGE, as well as on adolescent and adult medical and life issues. She has a son, Jacob, aged 22, who has CHARGE syndrome

Presentation Abstract:

The use of Person Centred Planning is becoming widely recognised as a tool to support effective planning for transition to adulthood and life-long learning. This presentation will consider how this approach can also be used with younger individuals with CHARGE (especially those with more complex needs) to gather information and plan an effective educational programme. The Person Centred Planning Approach can support clear identification of the child's strengths and needs and can lead to an appropriate individually tailored programme and package of support by discovering and learning who the person really is, drawing on the knowledge of family members and involved professionals. Examples will be provided to show how this approach may be used to provide positive outcomes for these individuals.

“Can Person Centered Planning Work with Younger Individuals with CHARGE?”

Gail Deuce M.Ed
Children’s Specialist Services, Sense UK

Nancy Salem-Hartshorne, PhD
Department of Psychology,
Central Michigan University.

What is Person Centred Planning (PCP)?

- A process of planning around the individual
- A tool to support effective planning for transition to adulthood
- A replacement for more traditional assessment approaches

Why use PCP with a younger person with CHARGE?

- To gather information and plan an effective educational programme
- To support clear identification of strengths and needs
- To take a longer term view

Why use PCP with a younger person with CHARGE?

- It encourages a focus on the question:

“What can be done to support greater independence, ownership of learning and self-determination?”

What information should PCP attempt to gain for the younger person?

- To identify strengths and needs
- To identify their communication preferences and needs
- Use of vision, hearing and other senses and identification of support needed to enable optimal use of sensory information
- What the young person’s likes/dislikes are at home and at school
- Things that can be achieved independently
- Things that help is needed with, and the type of help required
- Hopes for the future

How to obtain this information

A range of tools:

- Photographs
- Drawing
- Writing on large sheets of paper
- Talking and words being scribed
- Mind maps
- PATH

What is vital:

- That the young person remains at the centre
- It reflects what is important to the young person
- It reflects what they are capable of

Case study

Growth/Puberty, Part 2
CHARGE Syndrome:
The R and the G

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Panzacola G1

Dr. Jeremy Kirk
Consultant pediatric endocrinologist
(Honorary Reader) at the Diana
Princess Of Wales Children's Hospital
in Birmingham UK

Presenter Information:

Dr. Jeremy Kirk is a Medical Advisor and Committee member UK CHARGE Family Support group. Regular lecturer at UK and Australasian CHARGE groups; currently attending his 7th USA Family Group meeting. Consultant pediatric endocrinologist (Honorary Reader) at the Diana Princess Of Wales Children's Hospital in Birmingham UK, with a special interest in hormonal problems including growth, growth hormone therapy and puberty, and has published on these, including recent chapters in both the German and also American books on CHARGE syndrome. Dr. Kirk also set up UK screening for the CHARGE gene CHD7 in Birmingham. He is currently setting up a national UK survey of CHARGE syndrome through the Royal College of Paediatrics and Child Health (RCPCH) and is also working with Great Ormond Street and Manchester Children's Hospital to set up a network of national specialist CHARGE clinics.

Presentation Abstract:

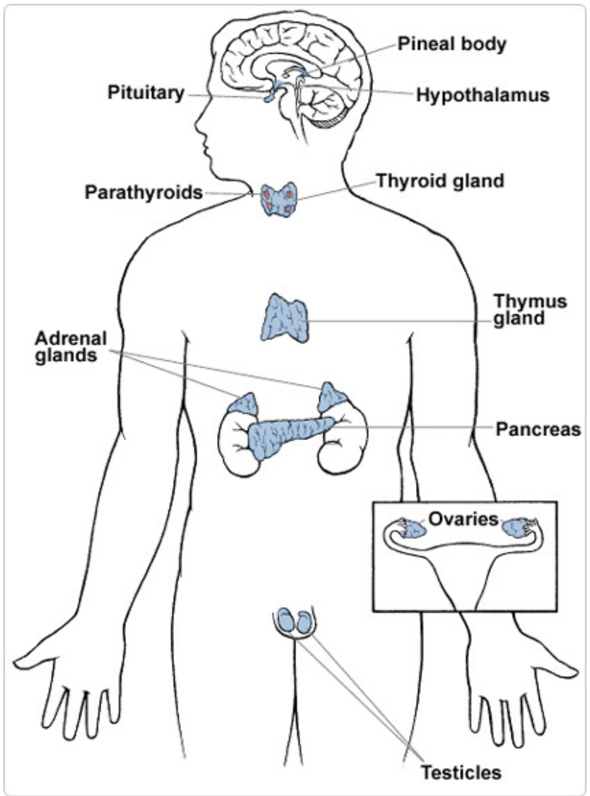
A number of different hormones (chemicals secreted into the bloodstream by specialized glands) are known to be affected in CHARGE syndrome, including those not only involved in growth but genital development and also puberty. It is now well recognized that puberty can be absent, delayed or arrested part way through in adolescents with CHARGE, more so in boys than girls. Although the presence at birth in boys of undescended testicles and also a micropenis (less than 1") may point to future problems in puberty, recent research indicates that a simple smell test may predict those children of both sexes who will need assistance in puberty. This may well help to ensure that teenagers with CHARGE syndrome receive timely and appropriate hormone replacement therapy, reducing the long-term risks of osteoporosis.

CHARGE syndrome; The R and the G

Jeremy Kirk

That growth and genital problems are an important part of CHARGE syndrome was recognised in their inclusion in the original description. In CHARGE the **R** stands for retarded growth and (development), and **G** for genital abnormalities.

These processes are not only under the control of nutrition (food) but also hormones, chemicals produced from specialised glands, which are released into the bloodstream and carried by it to other parts of the body to produce their effect. There are several different hormones involved in growth and puberty, and unfortunately many of them are affected in CHARGE.



The Endocrine system

GROWTH

There are 3 growing phases, all of which are under different controls. These phases are:

Infantile: This occurs during the first 2-3 years of life, and is almost completely dependent on nutrition.

Childhood: This occurs from ~2 years of age until puberty, and is dependent on both nutrition and also hormones including growth hormone.

Puberty: From puberty onward, this phase is under the control of growth hormone and sex hormones acting together.

All of these phases are affected in CHARGE, which results in around ¾ of children having heights and weights below the normal range, with reduced final height in many.
Infantile: many babies with CHARGE have low birthweight and/or length, and failure to thrive after birth is common as around 90% have feeding problems, which can be either mechanical (choanal atresia/stenosis, cleft lip and/or palate, tracheo-esophageal fistula) or abnormalities in nerves involved in swallowing (cranial nerves VII, IX, X.)
Childhood: children can grow normally during childhood, although they are often small. Poor growth, especially if they are getting adequate calories, should raise the question of an underlying hormone problem.
Puberty: either failure to enter puberty (delayed/absent), or getting only part way through (arrested) is very common in CHARGE, especially in boys, few of whom enter puberty on their own, whilst up to ½ of girls do.

As a result we would recommend that all children with CHARGE are seen by a multidisciplinary team including a pediatrician with expertise in growth and hormonal problems (pediatric endocrinologist), ideally throughout childhood, and into adulthood.
At each clinic the following should be measured and plotted: height, weight, weight for height (body mass index), head circumference in younger children, and puberty assessment in older patients.
Additional information on height potential also can be obtained by measuring parents heights to produce a genetic target height, and assessing remaining growth by performing a "bone age," looking at the growing ends of the bones in a left wrist X-ray.
Apart from assessing nutrition eg. calcium and Vitamin D levels, blood levels of hormones may also be checked, either on a single blood test eg. thyroid, growth factors (IGF-1/IGF-BP3), although as some hormones are not produced in a constant fashion then a day case admission for stimulation testing may be required. This is commonly done to assess growth hormone, stress hormones (cortisol), and sex/puberty hormones.

Growth hormone therapy

Some children with CHARGE syndrome have been treated with injections of growth hormone in order to try and improve not only short term growth but also final height. Many (~75%) of these children were growth hormone deficient, but this may reflect the fact that they are more likely to be treated than children who are not. Data on several dozen growth hormone treated children with CHARGE from Europe and also the USA has shown initial good response, although there is little long-term data.

Genital abnormalities

These are present at birth, and as the hormones involved in genital development and puberty are the same may give a pointer to possible future problems in puberty. Indicators are:

Boys

- Micropenis (penis smaller than 1 inch at birth): Present in 85%.
- Undescended testicles (one or both): Present in 60%.

Girls

- Small inner lips (labia) probably very common.

Hormone testing can involve single blood tests (testosterone in boys, oestrogen in girls) but also commonly includes testing of the hormone drive of the hypothalamus and pituitary using the LHRH (GnRH) test, as this is often deficient in CHARGE (hypogonadotrophic hypogonadism), and in boys the hCG test which tests the ability of the testicles to produce testosterone. Girls may also have an ultrasound scan looking at the uterus (womb) and ovaries. These tests are often done shortly after birth when there is a “mini-puberty” and also from 10-11 years of age.

Our experience in Birmingham is that many girls show normal hormone responses, whilst these are significantly reduced in boys both at birth and also puberty.

Micropenis is treated with the male hormone (testosterone) either given by monthly injection or daily cream. Undescended testicles usually need to be brought down surgically, and if the child is having repeated operations this is usually done at the same time to avoid extra anesthetics. Hypoplastic labia don’t need treatment.

Delayed puberty

Is it delayed, absent or arrested? The definition of delayed puberty is absence by 13 years in a girl, 14 in a boy. As these pubertal hormones are involved not only in the physical changes of puberty but also the puberty growth spurt, patients with delayed puberty will not only show absence of puberty signs but also poor growth and short stature in teenage years.

NB: Absent sense of smell (anosmia) now appears to be a pointer to pituitary puberty hormone problems.

NB: Remember that some sex hormones arise from the adrenal glands which lie above the kidneys, so pubic or armpit hair alone, spots, greasy hair and skin, adult body odour and adolescent mood swings may not necessarily be indicators of true puberty!

Testing is the same as in genital abnormalities ie.

Baseline "one-off" bloods are often not useful: LH, FSH, oestrogen (oestradiol) in girls, testosterone in boys.

Stimulation tests:

- LHRH tests "top end": hypothalamus and pituitary.
- hCG (in boys) tests "bottom end": testicles.

Treatment involves sex hormone replacement in gradually increasing doses to mimic normal puberty:

- In boys testosterone given by injection, tablet or gel.
- In girls oestrogen given by mouth, and then usually the oral contraceptive pill which contains both oestrogen and the other female hormone progesterone. To avoid frequent and distressing periods, the packs can be run into each other with withdrawal bleeds only 2–3 times per year rather than monthly.

Whilst many parents are concerned about giving sex hormones to children with CHARGE because of concerns about worsening behaviour, inappropriate sexual behaviour, menstrual bleeding (in girls), persistent erections (priapism) in boys, this must be balanced against the risks of long-term osteoporosis (brittle bones), as most bone strength is laid down in the mid to late teens under the influence of sex hormones, and then steadily lost after that. Every 10% increase in bone mass is equivalent to a 50% reduction in fracture rate.

Although other factors such as calcium and vitamin D are important neither on their own will prevent osteoporosis, and so we do give sex steroid therapy to all adolescents with CHARGE, which is usually well tolerated, and if problems arise with gel or tablets the dose can be rapidly cut back. The bone scans (DEXA) do require specialist interpretation to take into account other factors such as short stature which may overestimate osteoporosis, although studies on adults with CHARGE do indicate that bone strength is significantly reduced especially if sex hormone therapy has been delayed or not given.

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1. Blake, K., Kirk, J. M., & Ur, E. (1993). Growth in CHARGE association. *Archives of Disease in Childhood*. *68*, 508–509.
2. Blake, K. D., Salem-Hartshorne, N., Daoud, M. A., & Gradstein, J. (2005). Adolescent and adult issues in CHARGE syndrome. *Clinical Pediatrics, 44*, 151–159.
3. Forward, K., Cummings, E. A., & Blake, K. D. (2007). Bone health in adolescents and adults with CHARGE syndrome. *American Journal of Medical Genetics, 143A*, 839–845.

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No More Confusion about the Transition to Adult Services

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Panzacola G2

Beth Jordan, M.Ed., CRC
Regional Representative
Helen Keller National Center

Presenter Information:

Beth Jordan is the Great Plains Regional Representative for the Helen Keller National Center, serving deaf-blind consumers and their families. She also provides training and technical assistance to agencies/organizations who work with deaf-blind people. The Great Plains Region includes Iowa, Kansas, Missouri and Nebraska. Prior to her work at HKNC, she worked four years as a VR counselor with a deaf caseload in Pasadena, Texas. Ms. Jordan received her Master of Education degree from the University of Arkansas in 1988, majoring in rehabilitation counseling and independent living with an emphasis in deafness.

Presentation Abstract:

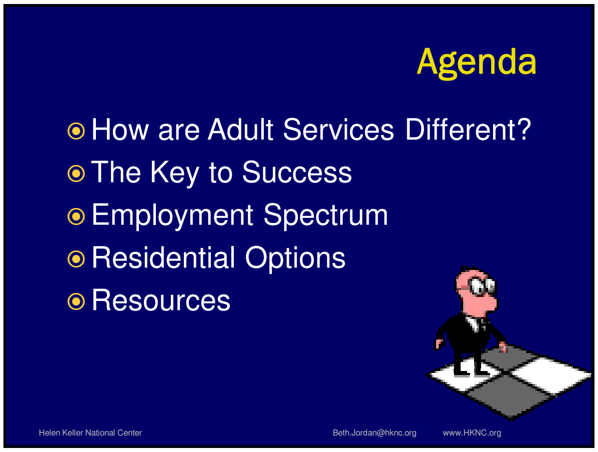
Often, the transition after high school can be confusing, overwhelming and stressful. Avoidance doesn't help. We will discuss the key differences between high school and adult services, and reveal the secret to success for your young adult. We will discuss the broad spectrum of options for work and home, from the least to the most supports. Walk away with an action plan of next steps for/with your young adult.

July 30, 2011

10th International CHARGE Syndrome Conference

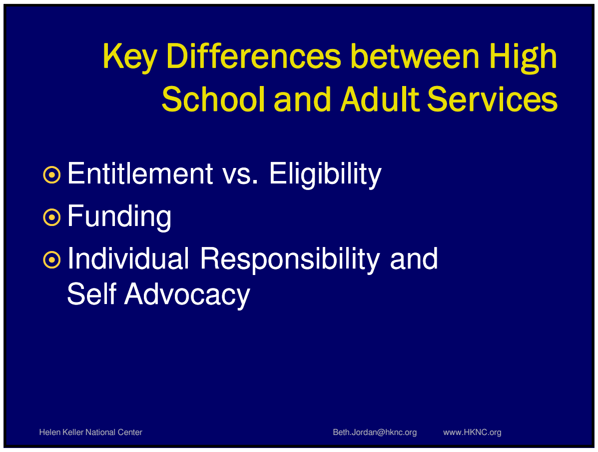
No More Confusion about the Transition to Adult Services

Presented by Beth Jordan,
Helen Keller National Center



Agenda

- How are Adult Services Different?
- The Key to Success
- Employment Spectrum
- Residential Options
- Resources



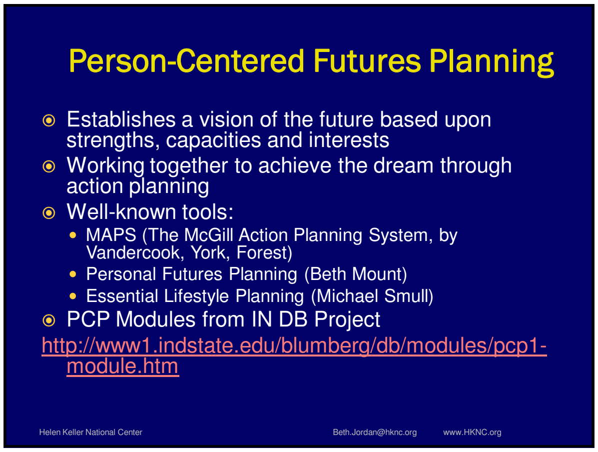
Key Differences between High School and Adult Services

- Entitlement vs. Eligibility
- Funding
- Individual Responsibility and Self Advocacy

The KEY

Identify goals and then look for supports

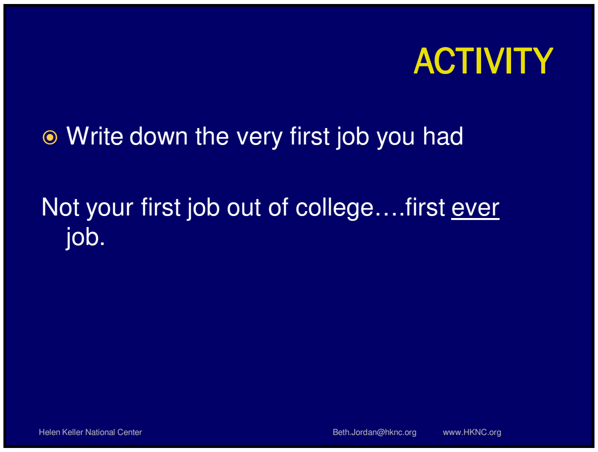
DO NOT identify programs and then select from your choices



Person-Centered Futures Planning

- Establishes a vision of the future based upon strengths, capacities and interests
- Working together to achieve the dream through action planning
- Well-known tools:
  - MAPS (The McGill Action Planning System, by Vandercook, York, Forest)
  - Personal Futures Planning (Beth Mount)
  - Essential Lifestyle Planning (Michael Smull)
- PCP Modules from IN DB Project

http://www1.indstate.edu/blumberg/db/modules/pcp1-module.htm



ACTIVITY

- Write down the very first job you had

Not your first job out of college….first ever job.

Services and Supports

- Vocational Rehabilitation
- Ticket to Work
- Medicaid
- Social Security Administration (SSA) Work Incentives
  - PASS (Plan for Achieving Self-Support)
  - IRWE (Impairment-Related Work Expenses)

www.familiestogetherinc.com
http://www.beachcenter.org/families/family_employment_awareness_training.aspx

Vocational Rehabilitation Services

Key Points

- Vocational Rehabilitation can provide support/services with the end goal of *employment*.
- No one can be denied an application.
- Even during Order of Selection, persons with the most severe disabilities get first priority.
- An IPE (plan) will be developed with the VR counselor once eligibility is determined.
- Services are time-limited.

Employment Options

Most Restrictive to Least Restrictive

- Day Program
- Sheltered Workshop (social enterprise)
- Enclave
- Supported Employment
- Competitive Employment
- Self-Employment

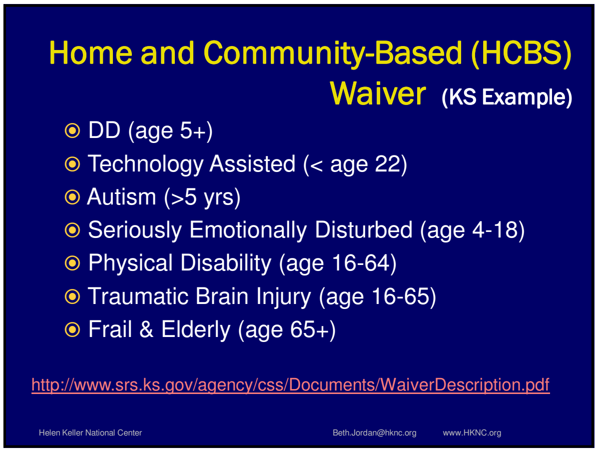
Customized Employment (CE)

- Person-Centered approach to work
- Time-intensive; consumer focused
- Use of "discovery" to develop direction
- Michael Callahan of Marc Gold & Associates is national leader in the field
- Office of Disability & Employment Policy published *Customized Employment: Practical Solutions for Employment Success part 1 & 2*
  http://www.dol.gov/odep/pubs/custom/index.htm
  http://www.onestops.info/article.php?article_id=420

Residential Options

Most Restrictive to Least Restrictive

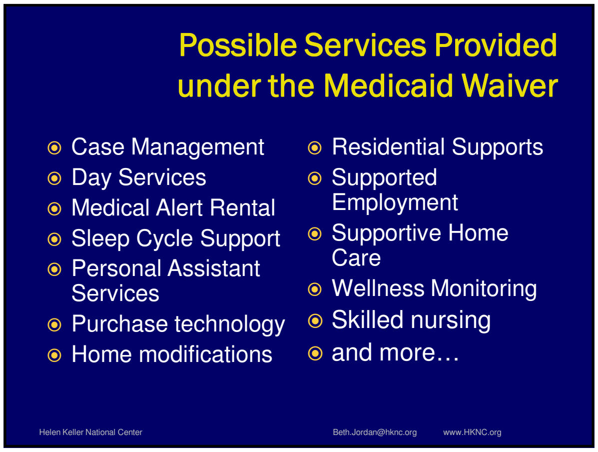
- ICFMR (Skilled Nursing Care)
- At Home with Parents/Family
- Group Home
- Apt/House with Paid Staff
- Independent
  - College dorm
  - Apartment
  - House



Home and Community-Based (HCBS) Waiver (KS Example)

- DD (age 5+)
- Technology Assisted (< age 22)
- Autism (>5 yrs)
- Seriously Emotionally Disturbed (age 4-18)
- Physical Disability (age 16-64)
- Traumatic Brain Injury (age 16-65)
- Frail & Elderly (age 65+)

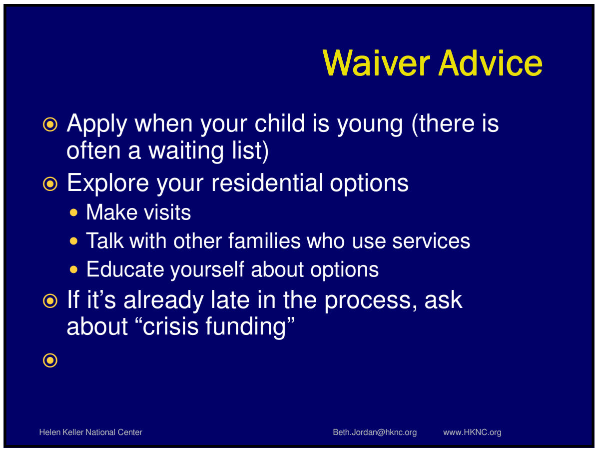
http://www.srs.ks.gov/agency/css/Documents/WaiverDescription.pdf



Possible Services Provided under the Medicaid Waiver

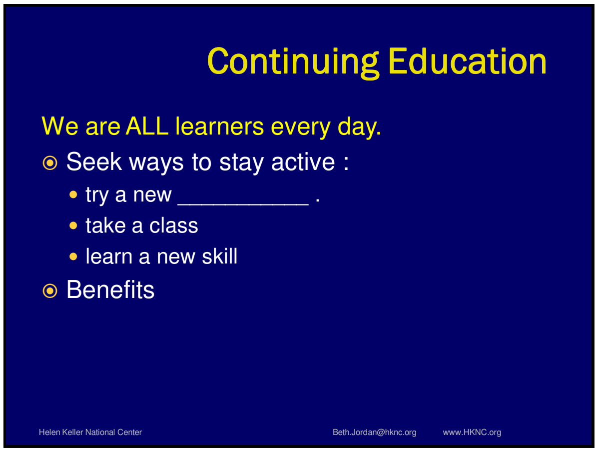
- Case Management
- Day Services
- Medical Alert Rental
- Sleep Cycle Support
- Personal Assistant Services
- Purchase technology
- Home modifications
- Residential Supports
- Supported Employment
- Supportive Home Care
- Wellness Monitoring
- Skilled nursing
- and more…

Where is the entry point(s) for Residential Supports in your state?



Waiver Advice

- Apply when your child is young (there is often a waiting list)
- Explore your residential options
  - Make visits
  - Talk with other families who use services
  - Educate yourself about options
- If it's already late in the process, ask about "crisis funding"
-



Continuing Education

We are ALL learners every day.

- Seek ways to stay active :
  - try a new ___________ .
  - take a class
  - learn a new skill
- Benefits

Resources

Helen Keller National Center (HKNC)

Mission: to enable each person who is Deaf-Blind to live and work in his or her community of choice.

- National Training Center on Long Island, NY
- National Registry
- Lifelong point of contact for information
- www.hknc.org or Beth.Jordan@hknc.org
- 913-677-4562 voice/TTY
- 866-327-6621 VideoPhone

HKNC Summer Programs for Young Adults

- 2-wk Program
  - 15th annual event; ages 16-22
  - Focus on Empowerment, Advocacy, Transition
  - August 1-12, 2011
- 8-wk Program
  - Individualized eval/training; ages 16-22
  - Assist with final education goals/transition
- Deaf-Blind Young Adults in Action (DBYAA)
  - 3rd annual event; ages 18-30
  - Mentoring; Learning the policy process
  - June 5-11, 2011; Washington, DC

HKNC Young Adult Summer Program

DBYAA – Washington, DC

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Adult Services Referral Checklist

For Youth and Adults with Vision and Hearing Loss

(see handout)

PFP Resources

It's Never Too Early, It's Never Too Late
A Booklet about Personal Futures Planning *by Beth Mount*

Available from the MN Governor's Council on Developmental Disabilities
http://www.mnddc.org/learning/document/GT075.PDF

PFP Process

Making Futures Happen
A Manual for Facilitators of Personal Futures Planning
by Beth Mount

Available from the MN Governor's Council on Developmental Disabilities
http://www.mnddc.org/learning/document/GT124.PDF

For Facilitators

Other Helpful Resources

Opening Doors to Post Secondary Education
by the WI Dept of Public Instruction

http://dpi.wi.gov/sped/pdf/tranopndrs.pdf

A Lifetime of Learning and Earning: VR Services FAQ for Parents of Deaf and Hard of Hearing High School Students
by Laurent Clerc National Deaf Education Center

http://www.gallaudet.edu/documents/clerc/vr_galluadet_stringer.pdf

Questions

Adult Services Referral Checklist for Transition-Age Young Adults Who are Deaf-Blind

Employment Services	Postsecondary Ed. Services	Housing Services	Independent Living Services	Legal/Advocacy/ Medical Services
Vocational Rehabilitation (includes Blind Services)	State/Private Universities	Dept of Mental Health & MR (CDDOs)	Centers for Independent Living (CILs)	Dept of Social Services (e.g. food stamps)
Workforce Investment Act (One-Stop Sites)	Community Colleges	Centers for Independent Living (CILs)	Deaf Service Centers (Commission f/t D&HH)	Deaf Service Centers (Commissions f/t D&HH)
Helen Keller National Center (HKNC) (for training)	Adult Education Programs	Deaf Service Centers (Commissions f/t Deaf & Hard of Hearing)	Services for the Blind (rehabilitation teaching, mobility instruction)	Community Mental Health Services
Private Employment Agencies (job development and placement)	Vocational/Technical Schools	State & Local Housing Authorities	Dept of Social Services (e.g. food stamps)	Community Medical Referral Systems
	Correspondence Schools (e.g. Hadley School f/t Blind)	Department of Aging	Assistive Tech Projects (Telecommunications Equipment Distribution Program)	Information & Referral Agencies (e.g. United Way)
	Internet-Based Education (on-line courses and webinars)	Private Agencies & Organizations (Columbus Colony, Ohio; Chestnut Lane, Gresham, Oregon)	State & National Consumer Organizations (AADB, NFADB, NFB, ACB, NAD, SHHH)	Protection & Advocacy (including the Client Assistance Program, Disability Rights Center)
				Centers for Independent Living (CILs)
				Social Security Administration (SSI, Medicaid)

Adapted from Everson & Killiam, *Using Adult Services to Achieve Transition Goals & Objectives,* in Everson's Supporting Young Adults Who Are Deaf-Blind in Their Communities, 1995. Presented 07/2011 by Beth Jordan, Helen Keller National Center, Beth.Jordan@hknc.org.

Today's date: ________________________

Action Plan			
Tasks	**Person(s) Responsible**	**By When**	**Accomplished?**

THE
CHARGE
SYNDROME
FOUNDATION

Issues in School Transitions: Preschool to Kindergarten

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Wekiwa 3 & 4

Linda L. Gerra, Ed.D.
The Jewish Guild for the Blind

Presenter Information:

Dr. Gerra has been involved with children of all ages with visual impairments and other disabilities in the administration of children’s programs as well as in providing direct services. She has extensive experience educating young children in Early Intervention and Preschool Programs. For the past 11 years, she has directed a program at The Jewish Guild for the Blind, and in this capacity has partnered with parents and families of children with visual impairment, blindness, and other disabilities, including those with CHARGE Syndrome. Helping families navigate through the second major transition of their child’s educational career has been an important aspect of the program. Dr. Gerra has also taught courses at Teacher’s College, Hunter College, and Dominican College

Presentation Abstract:

The transition process for a child with a visual impairment and his/her family from Preschool to Kindergarten can be a stressful time. This workshop will help decrease the stress by preparing parents for the transition. Resources for families will be discussed, and parents are encouraged to take advantage of local resources. Selecting a program for your child can be challenging. The benefits of an inclusion program vs. a self-contained program will be discussed. The Individualized Education Plan or IEP process can be daunting, but becoming familiar with the process will make it less so. Being an active participant in this process will ensure that your child has an appropriate education plan. Communication and collaboration between the Early Childhood Program and the Kindergarten program will also create a smooth transition for your child. This presentation reflects a process that can be used with preschoolers for the specific purpose of facilitating positive transitions to Kindergarten programs.

Issues in School Transitions: Pre-School to Kindergarten

Linda Gerra, Ed.D.
Children's Vision Consultant
The Jewish Guild for the Blind

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Family Support During Transition

- Resources for Families
  - NAPVI
  - JGB Teleconference
  - Family Connect
  - Commission for the Blind
  - Local workshops
  - Network with other parents
  - National Organization of Parents of Blind Children
  - National Federation of the Blind

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Evaluation

- Social history
- Educational evaluation in 5 domains
- Classroom observation
- Other assessments as necessary
- Psychological evaluation

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Selecting a School

- Community School vs. Private School
  - Related Services
  - Socialization and friends
  - Transportation
  - Other considerations: Adaptive Physical Education

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Your Job as a Parent during IEP Process

- Learn and understand the process
- Share information
- Ask questions
- Offer suggestions
- Speak up on your child's behalf
- Come prepared with a list of what goals you have for your child.

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Developing your Child’s IEP

IEP Process involves 2 main things:

- IEP Meetings
  - Discuss your child’s strengths
  - Discuss current level of functioning
  - What are your concerns for your child’s education
  - Review all evaluations or assessments
  - Review any unique needs related to your child’s disability (communication, behavioral, health, etc.)

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Developing your Child’s IEP

- IEP Document
  - Puts the decisions from the meeting in writing.
  - Outlines all of the components of where your child will attend school, including all services and supports.
  - Lists goals and objectives for your child
  - States that parents are equal participants
  - Decisions based on least restrictive environment
  - Lists special equipment your child might need
  - Identify related services and other supports
  - Highlight transportation needs

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Job as Parent during IEP Process

- Learn and understand process
- Share information
- Ask questions
- Offer suggestions
- Speak up on your child’s behalf
- Come prepared with a list of what goals you have for your child

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Communication Between Early Childhood Program and Kindergarten

- Collaboration
  - Working together
  - Building trust
  - Sharing information
  - Sharing a common vision
  - Ability to come to agreement
  - Make time for both teams to meet

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Orientation to the Kindergarten Staff

- Who Should do It?
  - Staff from previous program
  - TVI from previous program
  - New TVI
  - Parent

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Orientation to the Kindergarten Staff

- Orientation Includes
  - Approach to child with visual impairment
  - Modifying environment
  - Special equipment
  - Orientation to facility
  - Specific O&M needs of child with visual impairment

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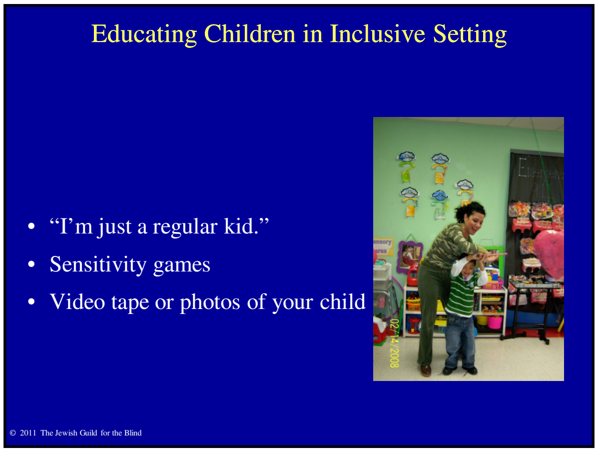

Educating Children in Inclusive Setting

- "I'm just a regular kid."
- Sensitivity games
- Video tape or photos of your child

02/14/2008

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Tips for successful Transitions

- Choose a specific date and stick to it
- Collect all personal materials – including communication systems and make sure they move with student
- Change is stressful for the student
- Change is good
- Schedule visits to the former program

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Role of Teacher of the Visually Impaired

- Teach the child skills
- Modify materials and environment.
- Support classroom teacher

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Orienting the Child to the New School

- Visit school in the spring and summer with O&M instructor.
- Visit school with parents

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Websites

- Special Education Guidelines
  http://www.education.com/topic/special-education-guide/
- Special Education Help
  http://www.education.com/topic/special-education-accommodations/
- Children with Special Needs: Your Rights as a Parent
  http://www.education.com/reference/article/special-needs-children-rights-parent/
- IEP Pop-Up for Parents of Children with Visual Impairment
  http://www.unco.edu/ncssd/resources/popup.shtml

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The End

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THE CHARGE SYNDROME FOUNDATION

My Intervener is Deaf!

Saturday, 07/30/11
Breakout Session #7: 2:15-3:15
Wekiwa 5

Dottie Snyder, M.S., M.Ed. NC Department of Public Instruction

Djenne-amal Morris, B.A. Family Specialist, NC Deaf-Blind Project

Presenter Information:

Dottie is an Educational Consultant with the NC DPI in Raleigh, NC. She has more than 35 years experience in the field of Special Education, initially working with developmentally individuals in Intermediate Care Facilities (ICF-MR) before becoming the teacher in a classroom for students who were deaf-blind in 2003. Contact information: dsnyder@dpi.state.nc.us

Djenne is a Family Specialist with the NC Deaf-Blind Project. She is a nationally recognized motivational speaker and is a Parent Educator/Training Coordinator at BEGINNINGS for Parents of Children Who Are Deaf or Hard of Hearing in Raleigh, NC. As the parent of a young man with CHARGE syndrome she strives to build a better world that sees the true beauty of the special needs world. Djenne lives in Knightdale, NC with her husband, son, and two daughters. Contact information: djennemorris@gmail.com/todaywithdjenne.com

Presentation Abstract:

Interveners have traditionally been considered the “eyes and ears” to assist a person who is deaf-blind to fully interact in the environment. But what if the “eyes and ears” happens to be deaf? At ENCSD in Wilson, NC, two such interveners have had success in assisting students who are deaf-blind to fully participate in school and community activities. The session and video will show the students and interveners participating in routine activities. A brief history and the recent standardization of training will also be discussed.

My Intervener is Deaf!

CHARGE CONFERENCE
Orlando, Florida
July 29, 2011
Dottie Snyder - Department of Public Instruction – North Carolina
Djenne Morris – Parent of a wild child with CHARGE Syndrome

Deaf-Blindness

- Creates unique needs for the child in the home, school, and community
- Ability to access visual and auditory information is affected
- Connection to the environment
- Learning, communication, and social development - Isolation

---

- Concept Development
- Cause-Effect
- Miss information and feedback/consequences
- Limited incidental learning
- Confused by multiple staff

What is an Intervener?

- Initially – the eyes and ears for a student who was deaf-blind
- Not an interpreter, not an EDA
- Purpose is to assist the student with a dual sensory impairment to access the environment
- Most famous intervener of all time 1887

An Intervener

- Is specially trained in relating to the issues with deaf-blindness – impact on learning and development
- Works CONSISTENTLY on a 1 to 1 basis with the child who is deaf-blind
- Role model and facilitator of independence
- Knows strategies and interventions

Communication

- Behavior is communication – need control
- Others miss or don't recognize communication attempts by the child
- Child doesn't recognize or understand communication attempts
- Don't realize that back and forth communication exists
- Don't understand that their actions lead to communication attempts deaf burglar

What an Intervener Isn’t

- EDA, bus duty, lunch duty, hall monitor, interpreter, gofer . . . . . .
- Comparison of Interveners and Paraprofessionals – Alsop, L., et.al. Interveners in the classroom: Guidelines for teams working with students who are deafblind

History

- Role of the intervener since the 1970’s
- 2002 National Intervener Task Force
- 2004 National Intervener Competencies
- National Intervener Task Force, National Personnel Preparation Consortium in Deaf-Blindness, and International Council for Exceptional Children
- Academy for Certification of Vision Rehabilitation and Education Professionals

Back in the Day

- Training informal (done by parent, teacher, or other)
- Training was not standardized (if any formal training at all)

What’s Happening Now *

- Task force met in 2002 Input from representatives from 30 states, NCDB, HKNC, NFADB, and DB-Link
- Five focus areas: Data and Information on Child I , Training, Awareness and Advocacy, Systems Issues and Change, and Parent Education and Involvement

- Role of an Intervener
- Establish Competencies
- Training standards
- Consistency in terminology and definitions
- Guidelines to establish a credible ongoing practice nationally

Determining Need

- Never too early
- No “level” or prerequisites needed
- Child needs a link to the rest of the world
- Consider vision, hearing, and communication level
- What about the physical environment - classroom, other students, staff training?

- What are the lesson like? Is your child expected to be on grade level?
- What is the pace in the classroom?
- Does your child have the concept development to keep up?
- Will your child have access to all information?
- Is the child psychologically and emotionally comparable to peers?

Interveners who are Deaf

At Eastern NC School for the Deaf

- 2 of 4 Interveners are deaf
- Adjustments in class minimal
- Work great in a school for the deaf
- Life experiences in terms of community and hearing loss beneficial
- They NEVER forget to use sign language

ENCSD

- One has completed training through the program at NC Central University (now at ECU)
- One is attending ECU online courses now and will finish soon.
- Other 2 are in limboland (the hearing ones)

Education

- Utah State University – Utah (Ski Hi Institute) - Certificate of Completion or Associate Degree
- East Carolina University – NC Certificate of Completion

QUESTIONS?

Contact:

- dksnyder@dpi.state.nc.us
- djennemorris@gmail.com

REFERENCES

- Alsop, L; Blaha, R; Kloos,E The Intervener in Early Intervention and Educational Settings for Children and Youth with Deafblinness
- Brown, D. Things to Remember When Requesting an Intervener for Your Child
- Zambone, A. and Alsop, L. Ensuring Access to Highly Qualified Interveners and Teachers : Establishing Intervener and Teacher Specialized Professional Associations in Council for Exceptional Children DVI Quarterly, Vol 54 #3, Spring 2009

- Webinar "Interveners: What's Happening" 2009 PowerPoint Located at NCDB site Topics Linda Alsop, Cyral Miller, Maurice Belote, and Alana Zambone
- Alsop, L., Goehl, k., Lace, J., Belote, M., & Rodriguez-Gil, G. (2007) Interveners in the Classroom: Guidelines for Teams Working With Students Who Are Deafblind

Resources

- California Deaf-Blind Services http://www.cadbs.org (fact sheets, Ask the Expert, links, publications)
- CHARGE Sydrome Foundation http://www.chargesyndrome.org
- DB link – National Consortium on deaf-blindness (articles, information on state deaf-blind grants, training,workshops, etc.)

- Helen Keller National Center Deaf-Blind Youth and Adult 141 Middle Neck Road, Sands point, NY www.hknc.org
- National Family Association for Deaf-Blind www.nfadb.org
- SKI-HI Institute

THE
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Ways & Means: Educating Children with CHARGE Syndrome in Local Schools

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Panzacola F1 & F2

David Brown
Education Specialist
California Deaf-Blind Services

Presenter Information:

David Brown is an itinerant teacher of children with deaf-blindness, and has worked in the field of special education since 1976. He first met a child with CHARGE syndrome when he joined the Sense organisation in the UK in October 1983. Since that time David has worked with a great many people with this syndrome. He has given presentations at the CHARGE Syndrome Foundation conferences in the US since 1995, at the Australasian CHARGE Association conferences since 1996, at the German CHARGE Syndrome conferences since 2008, and at the French CHARGE conference in 2004. He has also lectured at CHARGE meetings in 15 different states of the US, and in Canada, the UK, and India. David has published articles on various aspects of CHARGE (many translated into French, German, Italian, Dutch, Danish, Greek, Japanese, Hebrew, Norwegian, Finnish, Portuguese, Russian, Swedish, and Spanish) in '*Child: Care, Health & Development', 'Deafblind Education', 'Talking Sense', 'reSources', 'Deaf-Blind Perspectives', 'Fruhorderung Interdisziplinar', 'the American Journal of Medical Genetics', 'DbI Review',* and *'CHARGE Accounts'.* He has also contributed chapters to the first two books about CHARGE, published in Germany in 2009 and in the USA in 2010.

Presentation Abstract:

There is a widespread and growing myth that children with CHARGE cannot be educated successfully in the local school system. While it is true that these students might need some very particular adaptations and approaches, there are no obstacles that cannot be overcome if the district is prepared to be flexible, creative, open to innovative ways of evaluating and teaching, and prepared to collaborate fully with everyone involved with the student.

Ways & Means Case Study

David Brown
California Deaf-Blind Services
San Francisco State University

The student

- 8 years old girl
- Multiple health issues with many early surgeries & hospitalizations
- Suppressed immune system - frequent infections
- Severe balance issues, with delayed age of independent walking
- Fed completely by G-tube
- Some hearing – she has some speech perception with amplification
- Lip reader
- Primarily an American Sign Language user
- Visual acuities 20/400 in both eyes + visual field loss

School

- Program for deaf students with additional disabilities
- The Director of the service was flexible, imaginative, persistent, and supportive of students and staff
- Large support team (e.g. Deaf Teacher, Behaviour Specialist , Physiotherapist, Occupational Therapist, Speech Therapist, Adaptive Physical Education Teacher, Nurse, Psychologist, Sign Language Interpreter, Social Worker)
- High ratio of adults to students in the classroom
- Teachers & aides were all very highly qualified and experienced

School (continued)

- Strong emphasis on communication and language
- Total Communication approach, plus some ideas from the literature on CHARGE (eg. daily schedule of symbolic objects & pictures)
- Student had her own aide due to behavioral issues
- High level of academic expectation in the class
- Student spending increasing proportion of the day in individual activity with her aide because group sessions were very challenging for her
- Challenging behaviors were increasing in severity & frequency

The issues

- Often ***impulsive*** in behavior - e.g. escaping and running away, sweeping tables clear of objects, throwing objects, physical attacks on adults and (especially) other students
- All the above behaviors could also be used with intention in a planned way
- Alternation between extreme ***passive compliance*** and extreme ***non-compliance*** - described as "wanting to please people and wanting to upset and hurt the same people"
- Clearly an ***intelligent*** student yet also with perceived ***cognitive issues*** - e.g. unable to attend for long, unable to retain or generalize concepts learned, huge ***variability in skill level*** (from moment to moment and from day to day)
- Strong dependence on familiar, ***predictable routines***, with behavioral outbursts if her expectations were not met
- ***Ritualistic behavior*** in familiar activities
- Frequent insistence on ***getting horizontal*** on her back on the floor and being left alone for short periods of time

Before my visit - what had worked?

- Providing a '*safe area*' next to the window for his 'horizontal' periods
- Increase in the number of Adapted Physical Education sessions per week because of the beneficial after-effects of these sessions
- Daily calendar of objects & pictures
- Sessions with her favorite aide in the classroom (who was not her official 1-on-1 aide)
- Increasing the time spent in individual sessions with less time working in a group
- Being taken for a walk around campus after more severe behavioral events or outbursts

Before my visit - what had ***not*** worked?

- Physical restraint in a chair
- Being taken for a walk around campus after her more severe behavioral events or outbursts
- Extending the duration of a lesson to complete the task if it had been disrupted by her behavior
- Limiting the availability of time with her favorite aide to reduce the risk of her developing 'over-dependence'
- Limiting & scheduling the availability of the '*safe* area' next to the window for 'horizontal' periods
- Behavior plans (from 2 different specialists) that depended primarily on the use of punishers & on a token system
- Intensifying homework to facilitate better academic achievement & attention in class

My perception of what was wrong (1)

- There was no recognition that this is a student with deafblindness
- There was too much pressure to '*perform*' like her peers who are 'only' deaf, because she is a clever girl with very good adaptive skills, so her challenges were easy to miss or easy to under-estimate
- Failure to consider her individual motivators, and then use them within appropriate areas of the curriculum
- Failure to recognize her extended processing time and processing strategies (e.g. standing up to pay attention and learn, walking around the room briefly to process)
- Insistence that the student sit upright on a regular chair at a regular table for working on many areas of the curriculum

My perception of what was wrong (2)

- Not understanding the need for her to move and vary her position, and to perform tasks that provide strong proprioceptive input, to assist with her poor attention and poor self-regulation (& postural insecurities)
- Expecting the student to work at the thresholds of her sensory abilities most of the time
- The deliberate involvement of a variety of adults to prevent over-dependence on one adult in the school setting
- Team anxiety about using the "*follow the child*" idea in case this would spoil her, or increase her non-compliance
- Persistent use of behaviorist methods to control non-compliance and impulsivity, using a succession of practitioners, none of whom had familiarity with CHARGE nor with deafblindness, "….which just made the problems much worse"

What improved the situation? (1)

- The label of '***Deafblind***' - opening access to appropriate expectations, assessment approaches, teaching strategies, and resources
- Provision of an Intervener (who was already her favorite adult in the school)
- The use of the student's individualized motivators in as many curriculum areas as possible
- "*Follow the child*" as a guiding philosophy
- More careful '*reading*' of the student by staff, to identify and notice the precursors of stress, tiredness, and over-arousal (and ideas about what to do in response to these problems)

What improved the situation? (2)

- Involving the student in making a Personal Passport so that other people (adults and peers) from outside the class could '*tune in*' to her quickly and appropriately
- More precise, carefully chosen, & carefully presented, choices so that the student could feel more empowered and in control, & not so threatened and confused
- Provision of information and ideas about the vestibular and proprioceptive senses, and their impact on behavior (especially on attention and arousal), then putting this knowledge to use in both structured and opportunistic ways (involving Adapted Physical Education, Occupational Therapy, and Physiotherapy specialists)

What improved the situation? (3)

- Regular movement was encouraged, tolerated, and observed for its impact, once the relationship with the Intervener had developed and non-compliance had reduced accordingly

- The existing '*safe place*' for getting horizontal and staring at light was made accessible throughout the day rather than only for short periods at times pre-determined by a fixed daily schedule

- Determined efforts were made to link home & school effectively through daily contact

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Identifying the ‘P’ in CHARGE: Pain & the Relationship of Pain to Challenging Behavior

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Panzacola F3 & F4

Kasee Stratton, M.A.
Kennedy Krieger Institute: Pediatric Developmental Disorders Clinic at John Hopkins University School of Medicine
Central Michigan University

Presenter Information:

Kasee Stratton is a doctoral student at Central Michigan University. She received her Master of Arts in School Psychology in December of 2010. Currently she is completing her pre-doctoral internship in the Pediatric Developmental Disorders Clinic at the Kennedy Krieger Institute, a part of the John Hopkins University School of Medicine. She has been researching CHARGE syndrome, pain, and challenging behaviors for six years and has presented previously in Australia, New Zealand, Denmark, and at the 9th International CHARGE Syndrome Conference.

Presentation Abstract:

The research on pain in children with developmental disabilities is limited, including individuals with CHARGE. It has long been suspected that individuals with CHARGE have a high-threshold for pain. Our research, however, found that individuals with CHARGE experience considerable pain, including long term (chronic) pain. A relationship was found between challenging behavior (e.g. self injury) and an increase in pain intensity. A non-vocal pain measure will be discussed that was designed specifically for individuals with CHARGE to identify pain. Further, the relationship between pain and challenging behaviors will be described and intervention strategies will be suggested.

Identifying the ‘P’ in CHARGE: Pain and the Relationship of Pain to Challenging Behavior

Kasee K. Stratton, M.A.
Kennedy Krieger Institute at John Hopkins University School of Medicine
Central Michigan University

CHARGE FAST PASS

- Pain in developmental disabilities
- CHARGE syndrome and pain
- Are we identifying pain?
- Challenging behaviors in CHARGE
- How pain is related to challenging behavior
- Reducing the pain experience

Pain in Developmental Disabilities

- “Higher” threshold for pain
  - Has been suggested in CHARGE (Davenport, 2002)
- No evidence
- Higher risk for experiencing more frequent pain
- Identifying Pain in CHARGE
  - Poor communication strategies

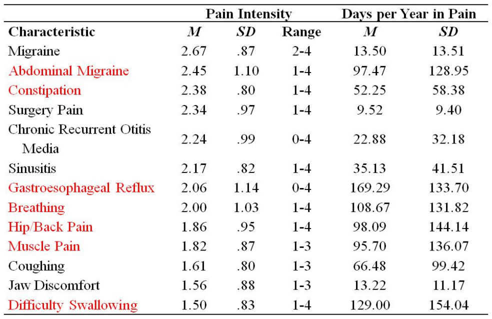
The ‘P’ in CHARGE

- Surgery (Stratton & Hartshorne, 2011)
  - 1 to 63 procedures
  - Average 13
  - Rated as painful experience
  - Average age of study 15 yrs. (Range of 7 months to 41.5 years)
- Procedures
- Doctor visits
- CHARGE related characteristics

Common Pain Experiences

Pain Experience	(N= 58*), n=	Percentage of Participants
Ear Infections	39	67.2
Sinus Infections	24	41.4
Gastroesophageal Reflux	24	41.4
Constipation	24	41.4
Surgery	23	39.7
Tactile Defensiveness	20	34.5
Migraine	14	24.1
Stoma Pain	12	20.7
Abdominal Migraine	11	19.0
Muscle Pain	11	19.0
Back Pain	8	13.8
Hip Pain	6	10.3
Jaw Pain	5	8.6
Pain During Sleep	5	8.6

Most Intense Pain and Average Duration



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

Migraines

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

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

Abdominal Migraine

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

Constipation

- Painful bowel movements
- Dry or hard stool
- Nausea
- Cramps, abdominal pain

- Fecal impaction
  - Abdominal cramping
  - Rectum discomfort

Gastroesophageal Reflux

- Average of 170 days a year
- Heartburn
  - Involves a burning pain in the chest (under the breastbone)
  - Increased by bending, stooping, lying down, or eating
  - More frequent or worse at night
  - Relieved by antacids
- Nausea and vomiting
- Regurgitation of food
- Sore throat

Tactile Defensiveness

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

Are we identifying pain?

- Are you able to determine when your child is experiencing pain?
  - 75% -Yes
  - Did not vary significantly by age of child

- Zero parents could identify chronic pain and no child could indicate chronic pain

Behaviors that Indicate Pain

- Vocal
  - Crying, moaning
- Social
  - Withdrawn, obstinate, difficult to distract, hard to console
- Facial
  - Frowns, mouth turned down, grinding teeth
- Activity
  - Less active, restless, disturbed sleep
- Body and Limb Movement
  - Rubbing area of pain, stiffens/spasms/seizures
- Physiological
  - Change in color, sharp intake of breath

Challenging Behaviors Indicate Pain

- Behavioral Challenges
- Self-Injurious Behavior (SIB)
- Dangerous Behaviors

- Aggressive, bites, hits head, throws objects, punches, pulls out g-tube

Why is it difficult to identify and measure pain in CHARGE?

- Limited or no communication strategies
  - Cannot use the gold-standard

- Possible social-communicative deficits
  - (Craig, 2006)

- Possible social referencing deficit
  - (Recchia 1997)

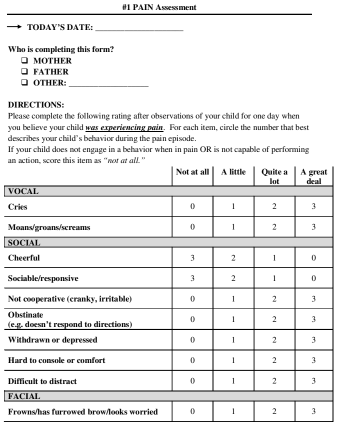
Measuring Pain

- Facial Reactions to Pain
  - Limited research
  - Facial palsy in CHARGE

- Rating Pain
  - Numerical ratings with pictures
  - Multidimensional pain tools

Measuring Pain

- Created a non-vocal, multidimensional pain scale
  - CHARGE Non-Vocal Pain Assessment (CNVPA)
- Developed from:
  - NCCPC-R (Breau et al., 1998)
  - PPP (Hunt, 2003)
  - Parent/caregiver input



#1 PAIN Assessment

→ **TODAY'S DATE:** ____________________

Who is completing this form?
☐ **MOTHER**
☐ **FATHER**
☐ **OTHER:** __________________

DIRECTIONS:
Please complete the following rating after observations of your child for one day when you believe your child ***was experiencing pain***. For each item, circle the number that best describes your child's behavior during the pain episode.
If your child does not engage in a behavior when in pain OR is not capable of performing an action, score this item as "*not at all.*"

	Not at all	A little	Quite a lot	A great deal
VOCAL				
Cries	0	1	2	3
Moans/groans/screams	0	1	2	3
SOCIAL				
Cheerful	3	2	1	0
Sociable/responsive	3	2	1	0
Not cooperative (cranky, irritable)	0	1	2	3
Obstinate (e.g. doesn't respond to directions)	0	1	2	3
Withdrawn or depressed	0	1	2	3
Hard to console or comfort	0	1	2	3
Difficult to distract	0	1	2	3
FACIAL				
Frowns/has furrowed brow/looks worried	0	1	2	3

What do we know about the CNVPA?

- Mean differences between no-pain and pain assessments were significantly different

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

Relevance	*n*	Percentage of Participants
Extremely Relevant	14	24.6
Relevant	17	29.8
Somewhat Relevant	18	31.6
Not Relevant	8	14.0

Instrument may not be relevant because:

- Child can verbalize pain vocally (12)
- Never complains of pain and seems to tolerate it well
- I've already developed ways to identify pain for my child (3)
  - "After 24 years, I am in tune to my child's health"

PAIN AND BEHAVIOR

Does Pain Impact Behavior?

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

Does Pain Impact Behavior?

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

Understanding Pain

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

All Behavior is Communication!

Reducing the pain experience

- Mitigation
  - Analgesics
  - Bed rest
  - Dietary change
- Redesigning the environment
  - Reducing the demands
- Teaching coping skills
  - Self advocacy
  - Functional communication alternatives

Caution with Medications

Analgesic failure may be due to …

- Inappropriate drug or dose selection for type of pain
- Genetic factors inherent to capacity to metabolize medications
- Impact of use of multiple drugs with competition for metabolic and excretory pathways
- Neurological substrate underlying CHARGE

Presenter Information:
Kasee Stratton, M.A.
Central Michigan University
Doctoral Student in School Psychology

Kennedy Krieger Institute at John Hopkins University School of Medicine
Behavioral Psychology Intern

Contact Information:
strat1kk@cmich.edu

THE
CHARGE
SYNDROME
FOUNDATION

Otolaryngologic Considerations in CHARGE: A New Understanding

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Panzacola G1

Dr. Sharon L. Cushing MD MSc. FRCSC
Department of Otolaryngology
Head & Neck Surgery
Hospital for Sick Children

Presenter Information:

Dr. Sharon Cushing is an Assistant Professor at the University of Toronto and a Pediatric Otolaryngologist at the Hospital for Sick Children in Toronto, Canada. She completed her fellowship training at Seattle Children's Hospital. Her clinical and research interests include disorders of the external, middle and inner ear with a specific focus on hearing loss, vestibular and balance function in children. Dr. Cushing is fortunate to be involved in the care of a number of children with CHARGE syndrome and their families.

Presentation Abstract:

Caring for a child with CHARGE syndrome provides the Pediatric Otolaryngologist with a unique and rewarding opportunity to build a long term relationship with the child and their family. The Otolaryngologic considerations in CHARGE syndrome are varied and range from early management of the airway to addressing multisensory issues throughout childhood. The current presentation will provide an overview of the diagnostic and therapeutic considerations for the management of the airway, hearing, taste and in particular balance function in children with CHARGE syndrome. Future therapeutic trends will also be discussed.

THE CHARGE SYNDROME FOUNDATION

Parent Panel Discussion

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Panzacola G2

Sandy Mack
Jillana Holt-Reuter
Mary Hancock

Presenter Information:

Presenters Sandy Mack, Jillana Holt-Reuter and Mary Hancock are parents of young adults with CHARGE syndrome. All are strong advocates who possess knowledge and skills on how to effectively navigate the transition planning process. Each panelist brings a unique perspective to this discussion. Sandy Mack is from Bristol, RI. Her son John is 19 and attends Perkins School for the Blind in Watertown, MA. Jillana Holt-Reuter is from Marshall, TX and her daughter Caitlyn, 22, graduated from high school last year. Mary Hancock lives in Lithia, FL and her son Philip, who lives in his own home near his parents, is 27.

Presentation Abstract:

The purpose of this parent panel discussion on transition will be to highlight the positive transition outcomes that were accomplished by the panelists, no matter how small, and to share with the audience how they achieved these outcomes.

THE CHARGE SYNDROME FOUNDATION

The Heart and CHARGE Syndrome

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Wekiwa 3 & 4

Nicole Janssen, MD
Department of Genetics
University Medical Center Groningen
P.O. Box 30.001 Groningen, the Netherlands
EMAIL: n.janssen@medgen.umcg.nl

Presenter Information:

Nicole Janssen studied medicine at the University of Groningen in the Netherlands and started working as a medical doctor in clinical genetics in Groningen in 2008. She recently began her training in clinical genetics. In 2009 she started her PhD project on CHARGE syndrome, CHD7 and heart defects that is still ongoing. As a part of this PhD project she studied heart defects in patients with CHARGE syndrome and a *CHD7*-mutation. In addition she is currently searching for a new genetic cause of CHARGE syndrome using next generation sequencing. She also participated in the Dutch multidisciplinary CHARGE outpatient clinic and is actively involved in the Dutch CHARGE parent support group.

Presentation Abstract:

I will focus on the H in CHARGE syndrome, the heart. The majority of individuals with CHARGE syndrome have a heart defect. Most individuals with CHARGE syndrome have a defect in a hereditary factor, the CHD7 gene. We studied heart defects in almost 300 individuals with CHARGE syndrome and a mutation (defect) in the CHD7-gene. We determined how often congenital heart defects occur in these individuals and what type of defects these are. We also looked if heart defects occur more often in combination with certain other symptoms of CHARGE syndrome, for example coloboma.

I will start my presentation by showing the anatomy and function of the heart and its development from embryo to baby. Thereafter, I will present the results of our study on CHARGE syndrome and the heart. We estimated how often heart defects occur in CHARGE syndrome, what type of heart defects these are and also if heart defects occur more often in combination with other CHARGE features. This information helped us to learn more about heart defects in CHARGE syndrome and the function of the CHD7-gene.

The heart in CHARGE syndrome

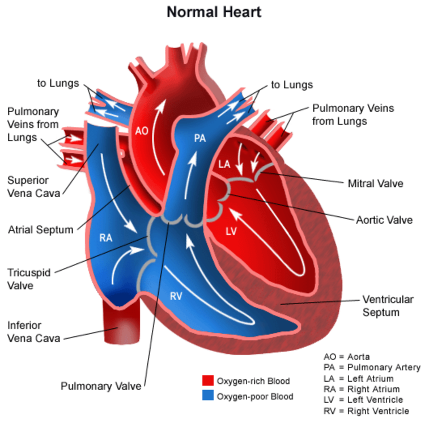
Nicole Janssen, MD
Clinical Geneticist in training
Department of Genetics
University Medical Center Groningen
The Netherlands
Email: n.janssen@medgen.umcg.nl

The heart in CHARGE syndrome

I will focus on the H in CHARGE syndrome, the heart. The majority of individuals with CHARGE syndrome have a heart defect. In addition most individuals with CHARGE syndrome have a defect (=mutation) in a hereditary factor, the *CHD7* gene.
We studied heart defects in almost 300 individuals with CHARGE syndrome and a mutation in the *CHD7*-gene. We determined how often congenital heart defects occur in these individuals and what type of defects are present. We also looked if heart defects were related to other features of CHARGE syndrome, the type of mutation and survival.

The heart, in general

The heart is an important organ that pumps blood through our body. The heart is located in the middle of our chest behind the sternum. It consists of a hollow muscle, which is divided in four chambers: two atria, a left and a right one, and two ventricles, a left and a right one. The atria first receive the blood, the ventricles pump it further. The right ventricle pumps blood to the lungs and the left to the rest of the body. Between the atria and ventricles are the mitral (left) and tricuspid (right) valve, which make sure that the blood does not flow backwards during pumping. Between the ventricles and aorta and pulmonary artery are valves as well.



What is already known about heart defects in CHARGE syndrome?

Large studies on the prevalence of heart defects in individuals with CHARGE syndrome have not been done before. According to current literature, heart defects occur in 66-92% of individuals with CHARGE syndrome due to a mutation in the CHD7 gene. These figures are based on a total of 178 individuals. Thus, not every person who has CHARGE syndrome has a heart defect. If a person does have a heart defect this is usually caused by an error in the embryonic development of the heart. There are many different types of heart defects related to the development of the heart. Exactly which types of heart defects occur in individuals with CHARGE syndrome and a proven CHD7 mutation has not been studied previously.

What did we study?

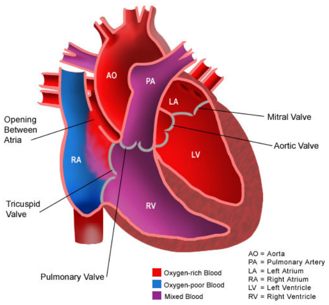
We tried to collect the medical data of as many individuals with CHARGE syndrome due to a CHD7 mutation as possible. We first looked if the person did or did not have a heart defect. If someone had a heart defect we accurately recorded the type of heart defect. All heart defects were classified based on what might have gone wrong during embryonic development. We also recorded all other features that may occur in CHARGE syndrome, such as coloboma (eye defect) and choanal atresia (blockage between nose and throat), as well as the specific mutation in the CHD7 gene and survival.

<u>What did we find in our study?</u>

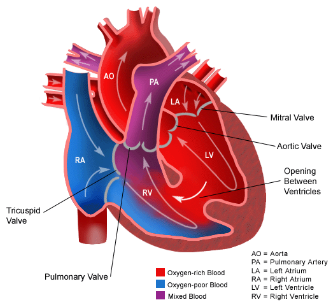
How often and which types of heart defects occur?

Our study revealed that heart defects are present in 74% of individuals with a *CHD7* mutation. Just like individuals with a heart defect without a syndrome, individuals with a CHD7 mutation often have atrial septal defects, in which an opening occurs between the two atria, and ventricular septal defects, in which an opening occurs between the two ventricles (see figures).

Atrial Septal Defect (ASD)

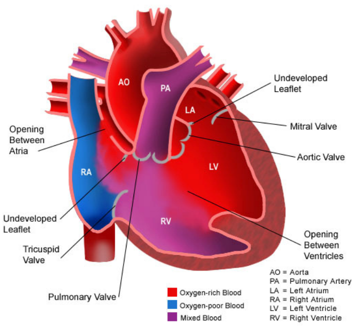


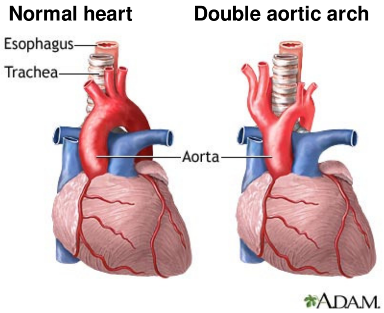
Ventricular Septal Defect (VSD)



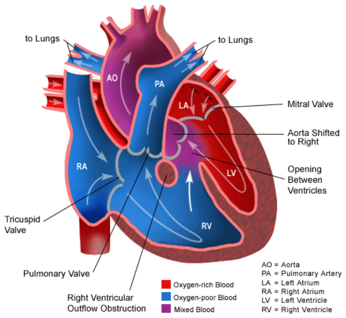
Remarkably, two types of heart defects occurred more often in the group of individuals with CHARGE syndrome than in individuals with non-syndromic heart defects. The first type is an atrioventricular septal defect (AVSD) or atrioventricular canal defect, in which an opening in the middle of the heart exists. The second type is an conotruncal or outflow tract abnormality. Examples of this type are a tetralogy of Fallot and double aortic arch.

Atrioventricular Canal Defect





Tetralogy of Fallot (TOF or "Tet")



Which features occurred more often in combination with heart defects?

We compared different features of CHARGE syndrome between the group of individuals with a CHD7 mutation and a heart defect and those without a heart defect.

It is important to realise that the relation between heart defects and other features is studied in groups of people. The outcome of a group can be different from the outcome of an individual person!

We discovered that individuals with CHARGE syndrome and a heart defect more often have tracheo-oesophageal abnormalities and a choanal atresia (blocked passage from nose to throat) than individuals without a heart defect. The trachea and oesophagus are located close to the heart and previous studies have already shown that tracheo-oesophageal abnormalities occur more often in combination with heart defects. That choanal atresia occurs more often in combination with a heart defect is a new finding. We don't know exactly why choanal atresia is associated with a heart defect in our group of individuals with a CHD7 mutation.

Furthermore, children with CHARGE syndrome who died at a young age, more often had a heart defect than the children who survived the neonatal period. This relation has been

described previously and is not surprising. Heart defects can be very severe and lead to death at young age, also in patients without CHARGE syndrome.
We also found a relationship between the type of CHD7 mutation and the chance of having a heart defect. We found heart defects more often in individuals with mutations that resulted in a complete loss of the CHD7 protein than in mutations that resulted in an altered CHD7 protein.

Acknowledgements

We thank all parents of patients with CHARGE syndrome and their doctors who helped in collecting the clinical information. Especially I would like to thank our collaborators from Radboud University Nijmegen Medical Centre in Nijmegen The Netherlands, the Great Ormand Street Hospital in London UK, Copenhagen University Hospital at Rigshospitalet, Copenhagen in Denmark and Oslo University Hospital Rikshospitalet in Oslo Norway.
We thank the NutsOhra foundation (number 0901-80) for financial support.

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THE CHARGE SYNDROME FOUNDATION

National Cochlear Implantation Studies with Children Who Experience Deaf-Blindness: Results for Participants with CHARGE Syndrome

Saturday, 07/30/11
Breakout Session #8: 3:45-4:45
Wekiwa 5

Susan M. Bashinski, Ed.D.
East Carolina University

Presenter Information:

Dr. Susan M. Bashinski has been working in the field of special education for more than 35 years, teaching in public school Pre-K through high school programs, as well as at the university level. She has been recognized with several teaching awards. She is the author / co-author of numerous published research articles, chapters, and manuals associated with topics relevant to learners who experience low-incidence disabilities and / or deaf-blindness (DB). For the past six years, Dr. Bashinski has served as a Site Principal Investigator for two national research projects conducted with learners who experience DB and have received a cochlear implant. Dr. Bashinski has extensive experience providing professional development and technical assistance across the US and internationally, particularly in the areas of communication development, AAC, and nonsymbolic communication intervention strategies for learners with low-incidence disabilities, including CHARGE syndrome

Presentation Abstract:

Since October 2005, back-to-back national research studies have been underway with children who experience deaf-blindness and have received a cochlear implant. The second largest participant subgroup in these studies, by etiology, is children with CHARGE syndrome. This session will highlight results obtained with this subgroup of participants. Data for the entire subgroup of participants with CHARGE, as well as a comparison of skill gains and communication development for the CHARGE participant subgroup and the overall study group will be presented. Specific data regarding any relationship of age at implant, as well as duration of time in sound since implantation, to communication development for children with CHARGE will be discussed. Finally, the session will introduce the Language Environmental Analysis (LENA)—an emerging technology that measures a child's auditory environment (i.e., meaningful talk, distant talk, television, general noise, and silence) across a 16-hour period. LENA software, used to analyze the frequency of adult words spoken to a child, a child's vocalizations, and conversational turns will be described. One case study with a participant with CHARGE syndrome, including LENA data, will be presented.

National Cochlear Implant Studies with Children with Deaf-Blindness: Results for Participants with CHARGE Syndrome

10th International CHARGE Syndrome Conference

July 2011

Susan M. Bashinski, Ed.D.

Funded by the U.S. Department of Education, Office of Special Education Technology and Media Services for Individuals with Disabilities (CFDA 84.327A).
Grant #H327A080045
Project Officer, Maryann McDermott

Opinions expressed within are those of the project / authors and do not represent the position of the U.S. Department of Education.

Project Staff

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Western Oregon University
Kat Stremel Thomas & Mark Schalock

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Susan M. Bashinski, Ed.D.

Cincinnati Children's Hospital Medical Center
Susan Wiley, MD & Charlotte Ruder

Acknowledgements

- We wish to extend a special "thank you" to all of the children and their parents who are participating in the study.
- We also wish to thank the manystate Deaf-Blind projects and private consultants who have assisted with the research.
- We couldn't have accomplished these tasks without you!

States Represented (26 + DC)

(States in blue include children with CHARGE)

Arizona	Maryland	Oklahoma
California	Massachusetts (Perkins)	Oregon
Delaware	Mississippi	Pennsylvania
Florida	Missouri	South Carolina
Georgia	Nebraska	Tennessee
Illinois	New Jersey	Texas
Indiana	New York	Virginia
Kansas	North Carolina	Washington
Kentucky	Ohio (CCHMC)	Washington DC

Three Major Project Phases

- Research - *Today's focus*
- Research to Practice - *Intervention Strategies*
- Practice to Technical Assistance & Training – *Methods used to teach care providers*

Outcomes
Participants will:

- learn aspects of cochlear implantation that appear to be positively correlated with communication skill gains by children with CHARGE
- gain knowledge regarding the range of outcomes achieved by children who have CHARGE Syndrome, following CI surgery
- learn about LENA technology and ways in which this data collection system can be used to guide intervention and language stimulation in the home

2009 National Child Count for Children Who Are Deaf-Blind

- Overall 4,313 children have a moderate-severe, severe, or profound sensori-neural hearing loss
- States increased their identification of children with implants from 251 in 2005, to 581 in 2009
- An increased number of children are receiving bilateral implants
- 747 children have been identified as having CHARGE Syndrome, of which 72 have confirmed implants

Outcomes of Project

- To collect data on the outcomes and related factors for children so parents / guardians can make more informed decisions about implantation, services, types of therapy for their children
- To identify factors correlated with more positive child outcomes, with the long-term objective of improved intervention and access to opportunities for language growth

Research Studies

- **Study A** – What effect does age at implant and "time in sound" (hearing age) have on child outcomes?
- **Study B** – What are the differences in the care provider's verbal interactions before and after implant?
- **Study C** – What are the effects of individualized interventions carried out by care providers, post-implant, in natural environments? (In Progress)

Research: Children Who Are Deaf-Blind With Cochlear Implants

- Participants' Status: How many children are participating?
- Demographics: Who are these children?

Participant Demographics: Children with CHARGE

Status	Number of Assessments					Total
	0	1	2	3	4+	
Post CI Only	1	9	4	3	1	**18**
Pre CI Only		6		--	--	**6**
Pre-Post CI		--	2	2	1	**5**
TOTAL	**1**	**15**	**6**	**5**	**2**	**29**

- 3 children with bilateral implants

Participant Demographics: Children with CHARGE

(n = 29)

Vision Impairment	Participants
Low Vision (<20/200)	38%
Legally Blind	28%
Light perception only	3%
Totally Blind	3%
Other	13%

Participant Demographics: Children with CHARGE

(n = 29)

Additional Challenges

- 58.6% have physical challenges
- 58.6% have cognitive challenges
- 20.7% have behavior challenges
- 93.1% have complex health care needs

Participant Demographics: Children with CHARGE

(n = 29)

Race/Ethnicity

- Black (6.9%)
- Latino (6.9%)
- White (82.8%)
- Mixed Race (3.4%)

Participant Demographics: Children with CHARGE

Participants' Age at Implant (n = 23)

(Range = 11 months to 5 years 2 months)

12 months or younger = 6
13 - 24 months = 7
25 – 36 months = 8
37 – 48 months = 1
over 48 months = 1

Participant Demographics: Children with CHARGE

Participants' "Time in Sound" / Hearing Age(as of most recent assessment) (n = 22)

(Range = 3 months to 6 years, 11 months)

12 months or less = 8
13 - 24 months = 4
25 – 36 months = 2
37 – 48 months = 1
over 48 months = 7

* A large number of young participants have little "time in sound."

Participant Demographics: Children with CHARGE

Participants' Age
(as of most recent assessment) (n = 22)
(Range = 19 month to 8 years 3 months)

12 months or less = 0
13 - 24 months = 4
25 – 36 months = 4
37 – 48 months = 2
over 48 months = 12

Research Studies

- Study A: What effect does age at implant and "time in sound" (hearing age) have on outcomes for children with CHARGE Syndrome?
- n = 22
- Longitudinal design
- Outcomes: Taken from a battery of assessments

Assessments

- A battery of assessments was selected that examined child behaviors across a variety of domains (birth to 60 months) & included small increments across items
- The Reynell-Zinkin Scales have been validated for children with low vision and blindness
- Assessments were repeated across time (depending on post-implant or pre-implant status; at least annually for post)

Assessments Used In the Research/Intervention Project

- *Communication & Symbolic Behavior Scales Developmental Profile*
- *MacArthur-Bates Communicative Developmental Inventory (W&G or W&S)*
- *Reynell-Zinkin Scales-* 7 sub-scales
- *Infant-Toddler Meaningful Auditory Integration Scale or Meaningful Auditory Integration Scale*
- *Speech Intelligibility Measures*

STUDY A: Example Data Analyses

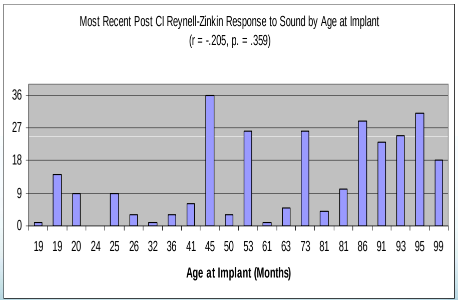
Reynell – Zinkin Scales:

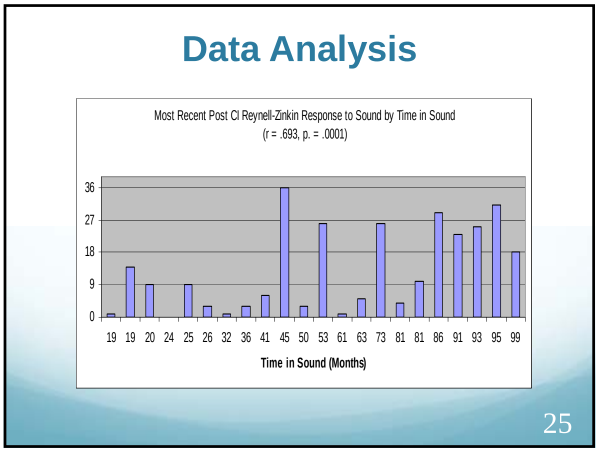
- Response to Sound
- Vocalization and Expressive Language

Other:

- Age at Implant
- "Time in Sound"
- Age at Assessment

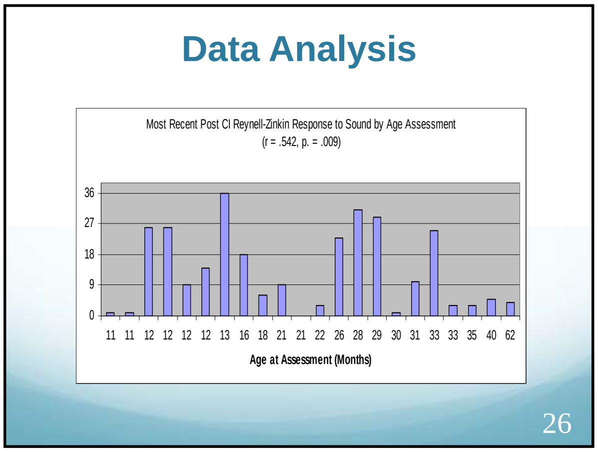
Data Analysis





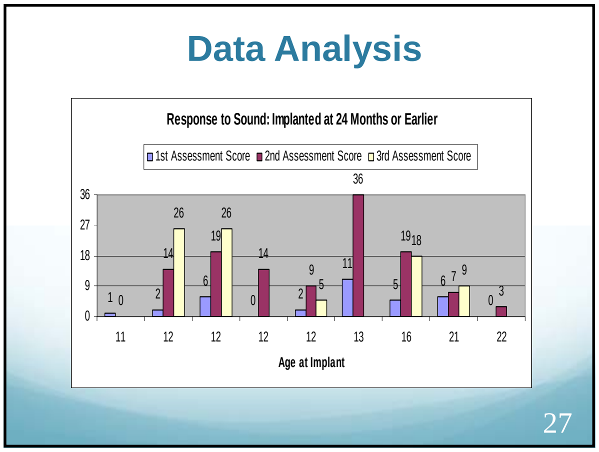
Data Analysis

Most Recent Post CI Reynell-Zinkin Response to Sound by Time in Sound
(r = .693, p. = .0001)



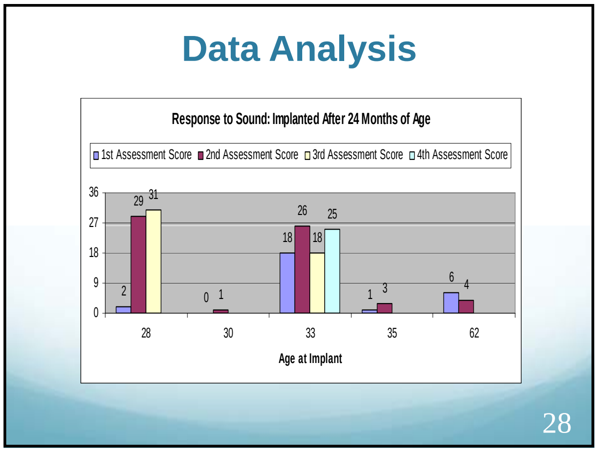
Data Analysis

Most Recent Post CI Reynell-Zinkin Response to Sound by Age Assessment
(r = .542, p. = .009)



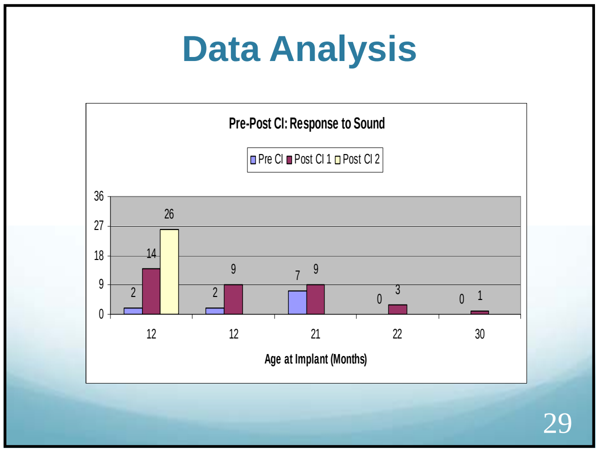
Data Analysis

Response to Sound: Implanted at 24 Months or Earlier



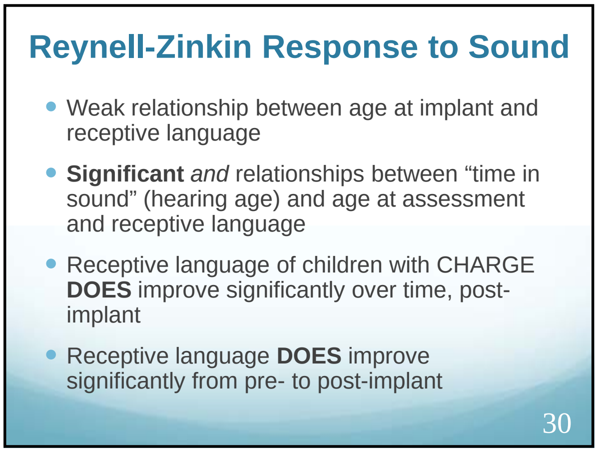
Data Analysis

Response to Sound: Implanted After 24 Months of Age



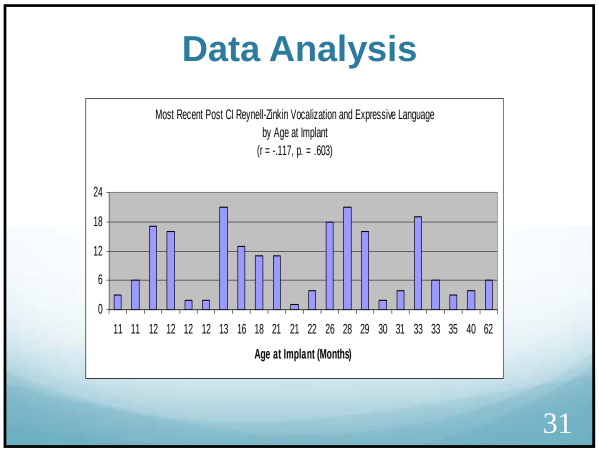
Data Analysis

Pre-Post CI: Response to Sound



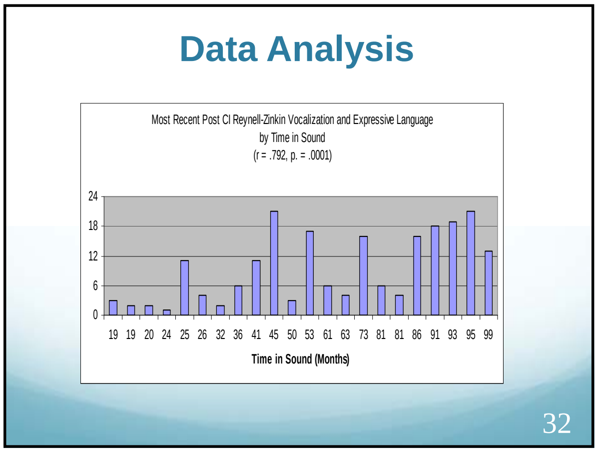
Reynell-Zinkin Response to Sound

- Weak relationship between age at implant and receptive language
- **Significant** *and* relationships between "time in sound" (hearing age) and age at assessment and receptive language
- Receptive language of children with CHARGE **DOES** improve significantly over time, post-implant
- Receptive language **DOES** improve significantly from pre- to post-implant



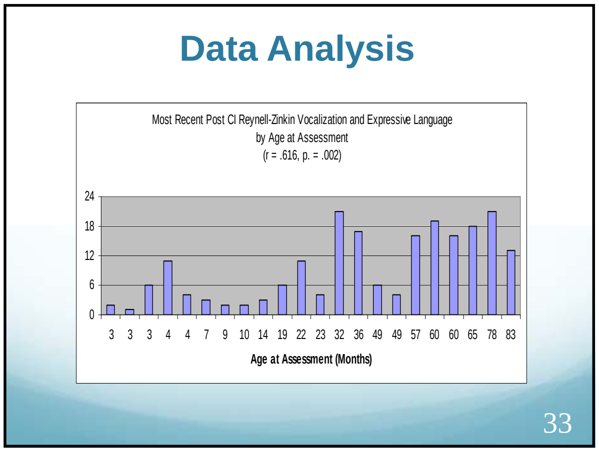
Data Analysis

Most Recent Post CI Reynell-Zinkin Vocalization and Expressive Language
by Age at Implant
(r = -.117, p. = .603)



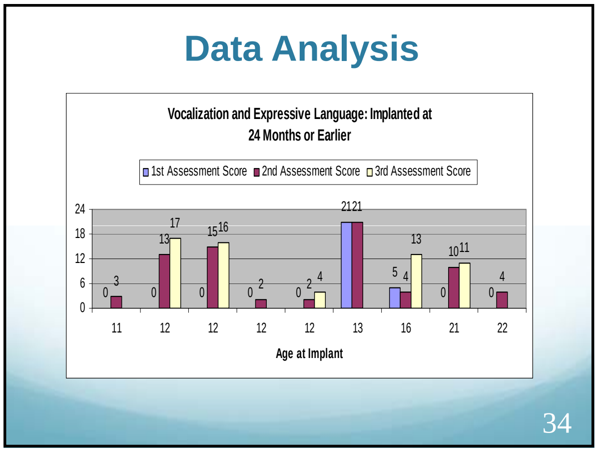
Data Analysis

Most Recent Post CI Reynell-Zinkin Vocalization and Expressive Language
by Time in Sound
(r = .792, p. = .0001)



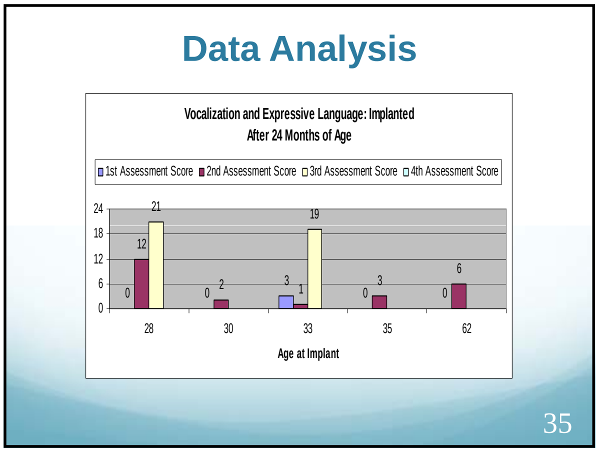
Data Analysis

Most Recent Post CI Reynell-Zinkin Vocalization and Expressive Language
by Age at Assessment
(r = .616, p. = .002)



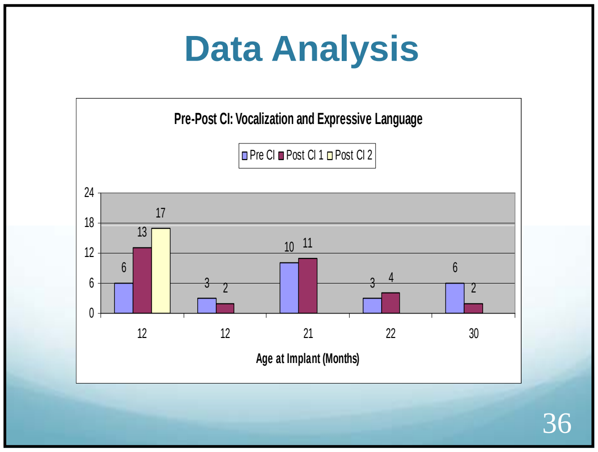
Data Analysis

Vocalization and Expressive Language: Implanted at
24 Months or Earlier



Data Analysis

Vocalization and Expressive Language: Implanted
After 24 Months of Age



Data Analysis

Pre-Post CI: Vocalization and Expressive Language

Reynell-Zinkin Vocalization and Expressive Language

- Little relationship between age at implant and expressive language
- **Significant** and strong relationships between “time in sound” (hearing age) and age at assessment and expressive language
- Expressive language of children with CHARGE **DOES** improve significantly over time, post-implant
- Expressive language **DOES** improve significantly from pre- to post-implant for *some but not all* children [to date]

Data for Post-Implant Children

(n = 22)

RECEPTIVE LANGUAGE		EXPRESSIVE LANGUAGE	
Response to sound	94.4%	Sound production	100%
Response to words and phrases	53.5%	One-word production/jargon	45.4%
Word identification (out of context)	45.4%	Meaningful words	45.4%
Simple directives	36.3%	Simple sentences	31.8%
Complex directives	31.8%	Complex sentences	18.2%

Data for Pre-Post Implant Children

(n = 5)

RECEPTIVE LANGUAGE			EXPRESSIVE LANGUAGE		
	Pre-	Post-		Pre-	Post-
Response to sound	60%	100%	Sound production	11%	100%
Response to words and phrases	20%	60%	One-word production/jargon	0%	40%
Word identification (out of context)	0%	20%	Meaningful words	0%	40%
Simple directives	0%	20%	Simple sentences	0%	20%
Complex directives	0%	20%	Complex sentences	0%	0%

Overall Findings to Date: Study A

- Participants in the study are a very diverse group
- With this diversity come complex relationships (rather than simple relationships between such variables as age and outcomes)

Overall Findings to Date: Study A

- The participants (as a group) ***do*** experience improvements in receptive and expressive language over time, after receiving an implant
- Individual outcomes vary considerably

Study B – Care providers’ talk to the child, after implantation (i.e., compared to pre-implant)

Use of the Language Environmental Analysis (LENA) to record:

- the auditory environment
- the adult’s verbalizations
- the child’s vocalizations
- the turns taken in conversation

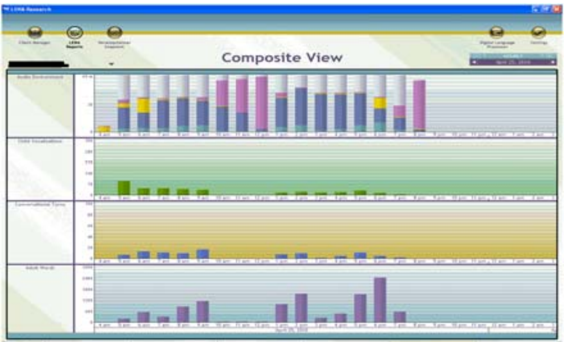
LENA Data

- Auditory Environment:
  - meaningful talk
  - distant talk
  - TV
  - noise
  - silence
- Adult Words
- Child Vocalizations

LENA Data

- Conversational Turns
- Estimated Mean Length of Utterance
- Estimated Developmental Age (in months)
- Standard Score
- Percentile

LENA Data



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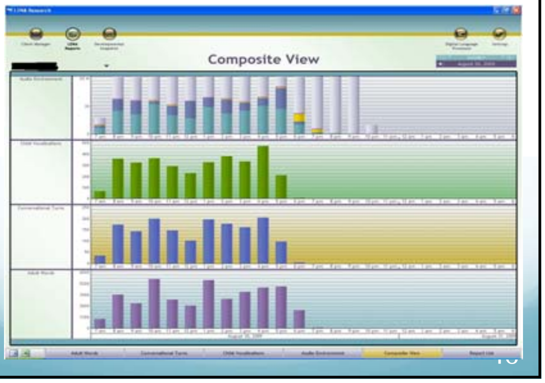

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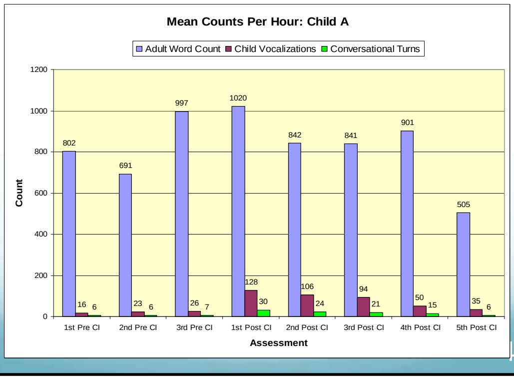

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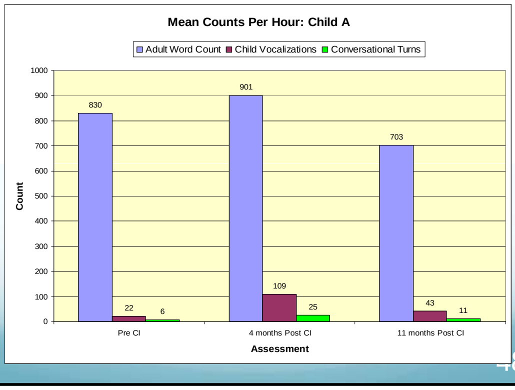

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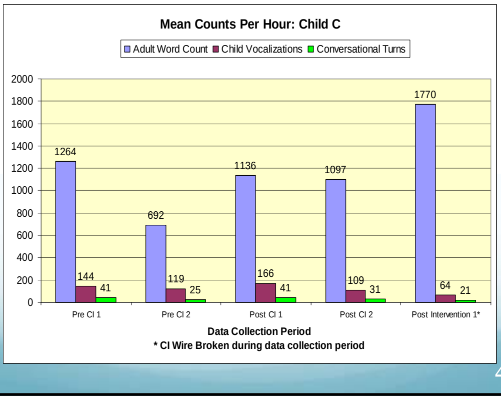

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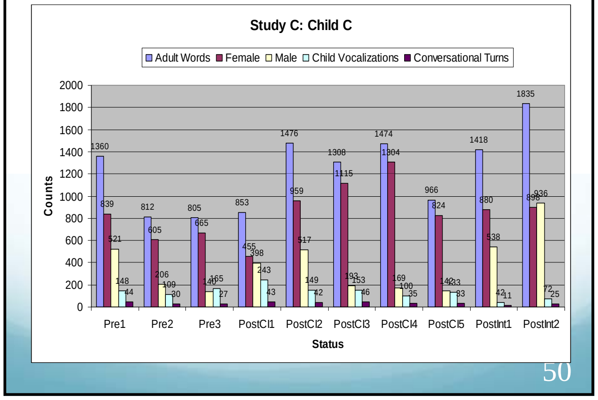


Overall Findings to Date: Study B

- Small numbers of pre- / post-implant children and their parents have participated [to date]
- Significant variability seen in parents' interactions with their children
- Some initial increase in verbal interactions, by both parents, has been observed after CI surgery
- Parents' verbal interactions vary considerably over time

Study C –Effects of individualized interventions, implemented by the care providers in natural environments, *after* CI surgery (In Progress)

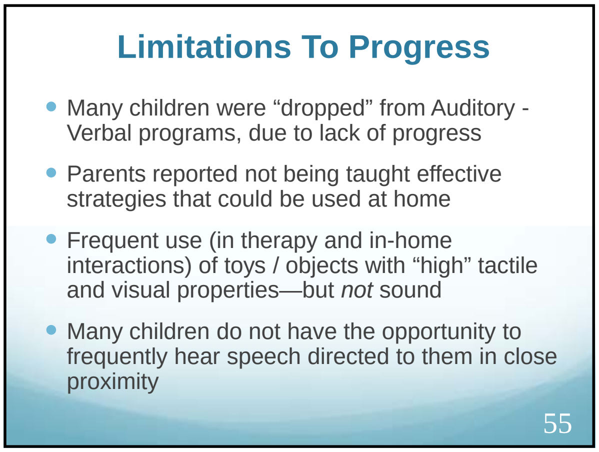
[One participant with CHARGE]

Overall Findings to Date: Study C

- Multiple child behaviors and care provider strategies are targeted in 12-16 sessions
- Repeated sessions, across time, are necessary for parent implementation [implications for TA]
- Three participants & their care providers have completed the intervention; four others are in progress
- Observed parent and child outcomes in maintenance and generalization conditions are encouraging

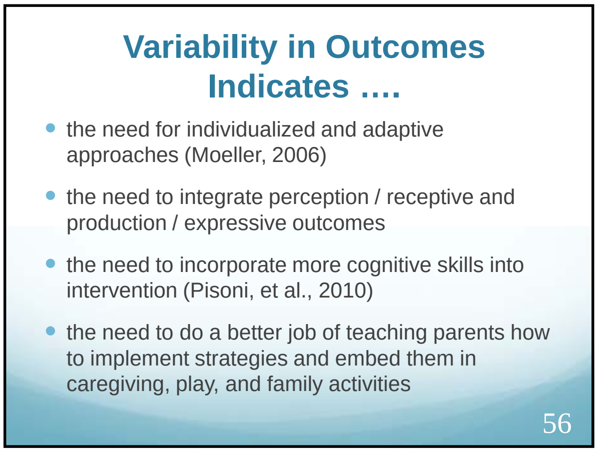
Limitations To Progress

- Many participants did not have prelinguistic communication skills
- Many participants did not have skills of functional object use
- Auditory - Verbal programs were not individualized
- Many participants did not wear their implants consistently
- Many participants were not mapped frequently (and, possibly, accurately)



Limitations To Progress

- Many children were “dropped” from Auditory - Verbal programs, due to lack of progress
- Parents reported not being taught effective strategies that could be used at home
- Frequent use (in therapy and in-home interactions) of toys / objects with “high” tactile and visual properties—but *not* sound
- Many children do not have the opportunity to frequently hear speech directed to them in close proximity



Variability in Outcomes Indicates ….

- the need for individualized and adaptive approaches (Moeller, 2006)
- the need to integrate perception / receptive and production / expressive outcomes
- the need to incorporate more cognitive skills into intervention (Pisoni, et al., 2010)
- the need to do a better job of teaching parents how to implement strategies and embed them in caregiving, play, and family activities

PLEASE visit our website:
www.kidsdbci.org

Family stories
Resources
Links

Thanks so much for your attention!

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THE
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It takes a community to build services for children with CHARGE

Sunday 07/31/11
General Session 9:15-9:45
Panzacola F&G

Marianne Riggio, M.Ed
Regional Coordinator
Africa and Caribbean
International training coordinator
Perkins International
Perkins School for the Blind

Presenter Information:

Marianne Riggio, M.Ed., is Program Coordinators for the Africa and Caribbean Regions and Training Coordinator for Perkins International at Perkins School for the Blind. She has worked extensively throughout the Untied States and the developing world toward the development of services to children with visual impairment with additional disabilities or deafblindness. She is a widely published author in the area of educating children with multiple disabilities and deafblindness

Presentation Abstract:

In this presentation we will reflect upon the ingredients of the success of the CHARGE foundation. It will present a brief overview of the evolution of the CHARGE community; the collaboration of professionals and families; the importance of the connections within the field of deafblindness and the lessons to be shared with other groups of children with low incidence disabilities.

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Young Adult Panel

Sunday, 07/31/11
General Session: 9:45-10:45
Panzacola F & G

Celie Harris, Parent
Ellen Howe, Young Adult
Kristin Ogan, Young Adult
Sal Schifano, Parent
Christopher Sence, Young Adult

Presenter Information:

Celie Harris and Sal Schifano are both parents of an individual with CHARGE. Ellen Howe from Australia, Kristin Ogan (a junior in high school at Indiana School for the Deaf), and Chris Sence (a Senior at Sam Houston University in Texas) are young adults with CHARGE syndrome.

Presentation Abstract:

A recurring theme among families is to know how individuals with CHARGE are going on in the world. What are they doing after school? Where do they live? What supports do they receive? Past conferences have had an interview/discussion session with young adults who are comfortable sharing their experiences. These presenters will share their experiences: Joys, celebrations, and possibly some of the frustrations in their lives. We have included parents of individuals who are more involved medically, in an attempt to give insight into the world in which they now live.

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Deep Thoughts

Sunday, 07/31/11
General Session: 11:00-11:45
Panzacola F & G

David Brown
Education Specialist
California Deaf-Blind Services

Presenter Information:

David Brown is an itinerant teacher of children with deaf-blindness, and has worked in the field of special education since 1976. He first met a child with CHARGE syndrome when he joined the Sense organization in the UK in October 1983. Since that time David has worked with a great many people with this syndrome. He has given presentations at the CHARGE Syndrome Foundation conferences in the US since 1995, at the Australasian CHARGE Association conferences since 1996, at the German CHARGE Syndrome Conferences since 2008, and at the French CHARGE conference in 2004. He has also lectured at CHARGE meetings in 15 different states of the US, and in Canada, the UK, Germany, and India. David has published articles on various aspects of CHARGE (many translated into French, German, Italian, Dutch, Danish, Greek, Japanese, Hebrew, Norwegian, Finnish, Portuguese, Russian, Swedish, and Spanish) in '*Child: Care, Health & Development', 'Deafblind Education', 'Talking Sense', 'reSources', 'Deaf-Blind Perspectives', 'Fruhorderung Interdisziplinar', 'the American Journal of Medical Genetics', 'Dbl Review',* and *'CHARGE Accounts'.* He has also contributed chapters to the first two books about CHARGE, published in Germany in 2009 and in the USA in 2010.

Presentation Abstract:

An event like the CHARGE Foundation Conference serves many different functions, and means many different things to the people who attend; I once wrote that each of these conferences was "…a seminar, a talking-shop, a party, a vacation, a networking paradise, a battery recharger, a morale booster, and a school reunion". By the end of the meeting emotions are always running high, and a huge amount of energy has been built up and focused on the future. In this session I will try to reflect on what seem to be the outstanding themes of the conference, mention some of the most significant events and personalities, and think about the most significant goals on the road ahead.