

Saturday, August 3, 2019

Breakout Session A5 • 10:45-11:45am • Chantilly Ballroom West

## CHARGE 101: Diagnosis, Features, and the Importance of Sensory Deficits

Meg Hefner, MS, Saint Louis University Kate Beals, Southland Pediatric Therapy

## **Presenter Information**

Meg is a genetic counselor with more than 35 years' experience with CHARGE syndrome. She was a founding member of the CHARGE Syndrome Foundation, Inc. and has written extensively on CHARGE for families (editor of the Management Manual for Parents), geneticists (American Journal of Medical Genetics special issues on CHARGE syndrome), and professionals working with children with CHARGE (CHARGE Syndrome, Plural Publishing). Recognition for her CHARGE syndrome work include the Stars in CHARGE award from the CHARGE Syndrome Foundation and the Diane Baker Alumni Award from the University of Michigan Genetic Counseling graduate program. Meg has presented at every CHARGE Syndrome Conference and actively answers genetic and medical questions via email and on several CHARGE Facebook pages.

Kate Beals is pediatric occupational therapist with 21 years of experience. She has worked with children who have CHARGE Syndrome at the South Carolina School for the Deaf and the Blind, in the South Carolina Interagency Deafblind Project, and in the Deafblind Program at Perkins School for the Blind in Massachusetts. Kate has served as a Consultant for Perkins International in China, and has provided training for international participants in the Perkins Educational Leadership Program. Currently, Kate is contracted with Southland Pediatric Therapy to provide OT services for children from birth to three years of age throughout the Upstate area of South Carolina.

## **Presentation Abstract**

The 101, 102, 103 series is designed for families with young children or new to conference and for professionals looking for a comprehensive overview of the medical and developmental features of CHARGE, In the 101 presentation, Meg will cover the diagnostic criteria and other medical features of CHARGE, including how various features affect early infant development. Why various labels are used (CHARGE, CHARGE-like, CHD7-related). How every child is unique and outcomes vary. Kate will elaborate on the differences in the sensory systems (especially hearing, vision, balance, proprioception) and sensory processing in CHARGE affect the way children experience themselves and the world around them. By understanding how the seven sensory systems operate differently in children with CHARGE, families, caregivers, therapists, and teachers can learn to recognize behaviors that suggest specific sensory processing issues and needs. Tips for dealing with these issues and needs will be presented. Kate's presentation will continue in the CHARGE 102 presentation.

## **Learning Objectives**

- Identify the most important features in distinguishing CHARGE from other syndromes.
- List three of the sensory deficits that can be part of CHARGE
- Illustrate three ways sensory deficits change typical behavior or delay milestones

### CHARGE Syndrome 101 Series for New Families

Meg Hefner, M.S. Genetic Counselor Adjunct Associate Professor of Pediatrics Saint Louis University School of Medicine meg@chargesyndrome.org

## Where does the name come from? \*\*1981 Pagon, et al.

- **C** = Coloboma of the eye
- H = Heart anomalies
- A = Atresia of the choanae
- R = Retardation of growth or development
- **G** = Genitourinary anomalies
- E = Ear anomalies and/or deafness

### Name features should NOT be used for diagnosis

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### Epidemiology of CHARGE Syndrome

- ~1 in 10,000 births worldwide
- Most often a new dominant variant (mutation)
  - Copy error in sperm (or egg) production
  - Increased but low (1-2%) recurrence risk
  - Major gene identified in 2004 (CHD7)
- · Mild end of spectrum is still being investigated

## Major CHARGE gene: *CHD7* on chromosome 8q12

- Very large gene with many unique variants - Gene sequencing required to detect
- What the gene does is being worked out
  If CHD7 pathogenic variant (mutation) is identified
  - It confirms diagnosis in questionable cases
    - Help define the mild end of the spectrum
    - Not finding a mutation does NOT rule out CHARGE
  - Possible to test other people in the family
  - Prenatal diagnosis /Pre-implantation genetic diagnosis – First identify a CHD7 variant in the person with CHARGE
  - Prost identify a CPD' value in the person with CHARGE
     Possible to detect the presence or absence of a mutation but not the severity of the features
- See presentations on Genetics of CHARGE

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## Chance of Recurrence

- Parents who have one child with CHARGE: 1-2%
- Children of individuals with CHARGE: 50%
- Extended family members:
- <<1%



Two children who have CHARGE, three (two pictured) without



## 2019 Diagnostic Criteria and Medical Features

- Which features help us know it is CHARGE
  - Major features
  - Supporting features
- What are some of the other features
   Not as important for diagnosis, but often medically important
- Clinical diagnosis is still the gold standard

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## Major diagnostic features

- Common in CHARGE but rare in other syndromes - Coloboma of the eye
  - Cranial nerve anomalies
  - Choanal atresia/stenosis
  - Characteristic CHARGE ears
    - Outer
    - MiddleInner
  - CHD7 pathogenic variants
- Need at least 2 Major features + supporting features

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## Macular coloboma: central vision loss

- The child will look above the point of interest and not appear to be making eye contact
- Note also facial palsy in this child
- This combination can make child appear inattentive



### Cranial nerves 9 & 10: Swallowing problems

- Many require G-tube feeding
- May last for years
- Some adults avoid certain textures and continue to have choking episodes

### Swallowing complication: Aspiration

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

## Cranial Nerve #7 Facial palsy





Unilateral – lopsided face

Bilateral - no facial expression

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### Cranial Nerve #1 Sense of smell

### Most have decreased or absent sense of smell

- Difficult to evaluate, but EXTREMELY common (>90%)
- Changes "taste"
- Implications for feeding
- Social implications
  - · smelly feet
  - body odor
- passing gas

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## **Choanal atresia/stenosis**

- Two sides (BL) or one side (UL)
- Bony or membranous
- Blockage (atresia) or narrowing (stenosis)
- · UL stenosis may be difficult to diagnose:
  - goopy nose all the time
  - lots of infections, fluctuating hearing loss
- · Often requires several surgeries to remain open



Baby with stents

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## Characteristic CHARGE Ear: OUTER EAR

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





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### Characteristic CHARGE Ear: MIDDLE AND INNER EAR

- Middle ear ossicular (bony) malformations (stapes, incus), other abnormal structures - Conductive hearing loss - not cured by tubes
- Inner ear (by MRI or CT scan) - Mondini defect: 1-1/2 turns to the cochlea - Small or absent semicircular canals\*\*
- Hearing Loss
  - Test early, test often
  - Know your child's hearing status
- \*\* most common feature of CHARGE



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CHARGE Syndrome Supporting/minor Diagnostic Features

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

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CHARGE Syndrome Diagnosis Supporting features that are common in many syndromes: **Heart defects** 

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



Heart surgery at four years old

### CHARGE Syndrome Supporting Feature: **Facial Clefts**

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



Repaired cleft lip

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## CHARGE syndrome supporting feature: Esophagus/Trachea

- Esophageal atresia (EA)

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

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### CHARGE syndrome supporting feature: Renal (**kidney**) anomalies

- Hydronephrosis (retained fluid)
- Reflux
- Horseshoe kidney
- · Small or absent kidney
- Cystic kidneys
- 40% have renal anomalies

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### CHARGE Syndrome Supporting Features

### Short Stature

Due to:

- Medical problems

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

### **Genital hypoplasia**

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

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## Variability in CHARGE

- EVERY feature can vary from absent to severe in different children
- NO ONE FEATURE is seen in every person with CHARGE
- Definite CHARGE is 3 Majors or 2 Majors and 3 Minors
- Probable/Possible/CHARGE-like with fewer
- CHD7 testing is especially helpful in less clear cases, but should always be done

## Other features of CHARGE

- Not necessarily helpful with diagnosis, but could be important medically or developmentally
- Differences have been noted in pretty much every organ system

## Other features of CHARGE

- Brain anomalies seen on MRI or CT
- Seizures can develop at any age
- Apnea
- Laryngomalacia
- Floppy cartilage
- Thymic or parathyroid abnormalities
- Immune system abnormalities
- Abdominal wall defects
  - Umbilical hernia
  - Omphalocele

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## Other features - continued

- Webbed neck
- Sloping shoulders
  - Absent trapezius muscle
- Nipple anomalies
- Limb/skeletal anomalies
  - Limbs: missing or extra fingers or bones
  - Vertebral anomalies
  - Scoliosis
- Craniosynostosis
- Others (any organ system)

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### Additional CHARGE management issues

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

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## Intelligence in CHARGE

- Assume normal intelligence until proven otherwise - long-term prognosis may be very good
- "Input impairment" due to combined vision & hearing loss – see CHARGE 102
- A few are "output impaired" due to bilateral facial palsy
- Delayed motor milestones due to vestibular dysfunction, upper body hypotonia and impaired vision.

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### How well do they do? The typical 2 year old with CHARGE:

- has spent 6 months in the hospital
- has had at least 6 surgeries
- is followed by 10 medical specialists
- is fed by G-tube
- is not walking or talking
- has some unusual behaviors
- Delayed? Yes. But may have a lot of potential to catch up you can't know yet.

## Senses in CHARGE

- \*Vision loss field and acuity
- \*Hearing loss mixed, often severe
- Balance abnormalities
- Touch
  - Sensitive to certain textures
  - Light touch??
- Altered pain sensation
  \*Smell absent or decreased
- Taste possibly normal
- \* distance senses

# Distance Senses and the Communication Bubble

- How far out and how well can the child see?
  - Coloboma (visual field)
  - Acuity
  - Other
- How far out and how well can the child hear ?
   Sensorineural loss
  - Conductive loss
  - Fluid. infections
  - Amplification
- Can the child *smell*?
- Anything outside the bubble is "off the radar"
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### **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"

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## Sensory loss and development

- Early motor development vision is important:
  - Head control
  - Sitting
  - Cruising, walking
- Communication: vision and hearing
  - Communication is learned by seeing and hearing what if you don't see and hear?

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- seeinghearing
- breathingbalancing
- smelling
- multitasking
- feeling
- empathizing
- swallowing

· spacial awareness

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- Hearing status (bubble)
- Balance
- Overall health, moods
- Observe carefully (learn from David Brown), communicate with caregivers
- Work on communication, communication, communication

## Walk a mile in her shoes

- It is impossible to understand hearing loss and vision loss at a purely intellectual level
- Do a simulation yourself try out your child's world
- Get the therapists and IEP team to participate in a simulation
- Find and use (read) the resources on the CHARGE Syndrome Foundation website



Saturday, August 3, 2019

Breakout Session B11 • 1:30-2:30 pm • Chantilly Ballroom West

## CHARGE 102: Sensory Deficits - How They Affect Development and Behavior and How To Help

Kate Beals, OTR/L , Southland Pediatric Therapy Laurie Denno, PhD, Behavior and Learning Consultant

## **Presenter Information**

Kate Beals is pediatric occupational therapist with 21 years of experience. She has worked with children who have CHARGE Syndrome at the South Carolina School for the Deaf and the Blind, in the South Carolina Interagency Deafblind Project, and in the Deafblind Program at Perkins School for the Blind in Massachusetts. Kate has served as a Consultant for Perkins International in China, and has provided training for international participants in Perkins' Educational Leadership Program. Currently, Kate is contracted with Southland Pediatric Therapy to provide OT services for children from birth to three years of age throughout the Upstate area of South Carolina.

Laurie S. Denno, Ph.D., is a board certified behavior analyst (BCBA-D) and a licensed applied behavior analyst in Massachusetts with over 40 years of experience working with individuals who have developmental disabilities. Laurie has worked in public schools, private schools, vocational programs, day habilitation programs and residential programs. Laurie has testified in educational hearings and conducted functional behavioral assessments for schools, parents and vocational programs. Laurie worked for 26 years at the Perkins School for the Blind with deafblind students, many of whom had CHARGE syndrome. She has been a faculty member at Simmons University (College) and Antioch University New England. Laurie continues to work as a behavior consultant in private practice. Her primary interests are CHARGE syndrome, deafblind education and behavior analytic applications for individuals with developmental disabilities and mental illness. She is a dedicated proponent of Positive Behavior Supports in education and adult services. Laurie has been a frequent presenter at the CHARGE Syndrome Foundation International Conferences on behavior analytic topics.

## **Presentation Abstract**

Most children with CHARGE are not just deafblind (hearing and vision deficits) but have multiple sensory deficits: there are also issues with balance, smell, and even touch. This creates differences in the way they receive and process sensory information, which in turn affect how they experience themselves and the world around them. All of this has a significant impact on early motor development, later motor development and behavior. In this presentation, Kate will continue her 101 presentation: review the relationship between the sensory and motor systems of the body and explore how sensory processing differences are reflected in motor function (sitting, walking, moving, etc). Then Laurie will outline common behavioral challenges seen in children with CHARGE and point out the interaction of behavior and the environment. She will then present possible ways to mold behaviors and teach socially acceptable behavior through behavior analytic interventions and outline additional avenues for behavior improvement.

## Learning Objectives

- Be able to explain how differences in sensory processing influence early motor development and balance in individuals with CHARGE Syndrome.
- Know how to set up an activity in a way that enables the child to derive maximum physical benefits from their participation.
- Identify common behavior challenges in CHARGE Syndrome and discuss recommended methods of assessment and intervention.

## Sensory and Motor Issues in Charge Syndrome CHARGE 101-102

14<sup>th</sup> International CHARGE Syndrome Conference Kate Beals, OTR/L, including material contributed by Danielle Bushey, DPT

### \*\*\*\*\*

### Sensory Systems and How they Operate Differently in Individuals with CHARGE Syndrome

1. The only way our brains get information about the world around us, and about what our bodies are doing, is through our senses.

2. Sensory information only goes INTO the brain, not out. It is structurally impossible for sensory information to go out away from the brain. It only goes IN.

3. Each sensory system has specialized receptors that react to different kinds of stimuli, including light, sound waves (vibration), chemicals, stretch, and others. That's why we can't see with our ears or hear with our eyes.

4. Each specialized type of receptor converts the stimulus into electrical impulses that travel along nerves TO the brain.

| TYPICAL   |          |  | IN CHARGE SYNDROME   |
|-----------|----------|--|--|
| SENSE     | FUNCTION | NOTES  |  |
|           |          |  |  |
| Vision    | Seeing   | 80-90% of what we<br>know about the<br>world around us<br>comes through the<br>sense of vision. It is a<br>"distance sense." | Colobomas cause visual field cuts. Usually<br>these are upper field cuts, but not always.<br>Think of an upper field cut as wearing a<br>baseball cap pulled down low. With a visual<br>field cut, a lot of information about the<br>environment is missing. <b>Reduced vision has</b><br><b>an effect on Balance.</b> |
|           |          |  |  |
| Auditory  | Hearing  | Also a "distance<br>sense," hearing<br>informs us about<br>what is going on in<br>our environment.                           | Individuals with CHARGE Syndrome are<br>often deaf or hard of hearing. Hearing loss<br>reduces access to information about the<br>environment.   |
|           |          |  |  |
| Olfactory | Smell    | Sort of a<br>combination of<br>"distance" and<br>"near" sense.   | For individuals with CHARGE Syndrome, the<br>sense of smell may be reduced or<br>altogether absent. This reduces access to<br>information about the environment, and<br>can cause safety concerns such as not<br>noticing the smell of smoke.  |
|           |          |  |  |
| Gustatory | Taste    | "Near" sense, greatly<br>influenced by the<br>sense of smell.  | Taste is reduced by the lack of a sense of<br>smell. Think about how food has no taste<br>when you have a cold. Also, many<br>individuals with CHARGE Syndrome are fed<br>through a G-tube or other type of tube,, so<br>they have reduced experience with the<br>sense of taste.                                      |

| Light Touch                          | Protection  | Receptors in the skin,<br>close to the surface<br>of the body.<br>Near/Body sense<br>that is alerting and<br>arousing. Think<br>about how it feels<br>when a bug is<br>crawling on you.<br>Yikes!  | Individuals with CHARGE Syndrome spend a<br>lot of time in the hospital early on. They are<br>poked and prodded a lot. Sometimes they<br>become defensive to being touched.<br>Sometimes they don't like to touch certain<br>textures, or sometimes they have an<br>excessive need to touch things. This is<br>something that needs a lot more research.   |
|--------------------------------------|---|--|--|
|                                      |   |  |  |
| Proprioception<br>(Deep<br>Pressure) | Provides<br>information<br>about body<br>position, body<br>movement,<br>and the effect<br>of gravity. | Receptors deep in<br>the muscles,<br>tendons, and joints.<br>Think massage.<br>Calming and<br>organizing. When<br>driving, you don't<br>have to look at your<br>ankle to know that<br>you moved your foot<br>from the gas pedal to<br>the brake, because<br>you can FEEL it. | Individuals who have CHARGE Syndrome<br>tend to have reduced proprioceptive<br>awareness. It is harder for them to "feel"<br>their joints moving and the position of their<br>bodies. Poor proprioception means you<br>need MORE OF IT in order to understand<br>how your body is moving. Lack of body<br>awareness in itself makes it harder to<br>understand your body's relationship to the<br>objects, people, obstacles, and open space<br>in the environment around it. <b>Poor</b><br><b>proprioception has an effect on Balance.</b> |
|                                      |   |  |  |
| Vestibular                           | Position of the<br>head in space.   | Tells brain:<br>*if you are right side<br>up, upside down,<br>moving forward or<br>backwards, side to<br>side, or up and down<br>*rotating/spinning<br>*what direction<br>*how fast<br>*if you are<br>accelerating or<br>decelerating.                                       | Individuals who have CHARGE Syndrome<br>frequently LACK THE STRUCTURES in the<br>inner ear that receive vestibular<br>information. These are called the<br>"Semicircular Canals" and they are located<br>close to the "Cochlea" which is the structure<br>that receives sound. If the person with<br>CHARGE Syndrome does have semicircular<br>canals, they may be malformed and<br>therefore inefficient in receiving<br>information about head movement. <b>Poor</b><br><b>vestibular processing has an effect on</b><br><b>Balance.</b>   |

5. The brain is constantly processing information from all of out sensory systems simultaneously, and figuring out what it all means, what is important, and what isn't.

6. SENSORY INTEGRATION: None of the sensory systems operates in isolation. Rather, the sensory systems work TOGETHER in a COMPLEX, DYNAMIC PROCESS. Various senses work together to either verify or refute information from other senses, and to inform other senses with additional information about what is happening inside and outside of the body. They also work IN CONJUNCTION with the MOTOR SYSTEM.

### How Sensory Input Relates to Motor Output

1. Motor instructions go OUT from the brain to the muscles. It is structurally impossible for them to travel in toward the brain. Motor instructions only go OUT.

2. "Motor output" just means moving our bodies. It is WHAT WE DO, and what we DO is BEHAVIOR. Behavior is motor output.

3. The body has a SKELETON made up of BONES. Our bodies can BEND at JOINTS, where certain bones meet.

4. A joint bends when the MUSCLES on one side of the bone CONTRACT (get shorter and tighter) and the muscles on the other side RELAX (get longer and less tight).

5. Muscles that move joints attach to the bone on the opposite side of the joint. When the muscle contracts, it pulls on the bone of body segment that the muscle is attached to, so the joint goes into flexion. When you bend your elbow, the biceps muscle contracts, while the triceps muscle relaxes, causing your forearm to move closer to your upper arm.

6. MUSCLE TONE: At rest, muscles have a certain state of readiness to move – not too tight and not too loose, but just right. This is normal muscle tone. Some conditions cause muscle tone to be too tight (high, such as in cerebral palsy) or too loose (low, such as in Down Syndrome). People with CHARGE Syndrome tend to have muscle tone on the low side.

| FROM      | то        | MEANING  |
|-----------|-----------|--|
| Cephalo   | Caudal    | "From head to tail"                                  |
| Proximal  | Distal    | "From inner to outer" (from the center of the        |
|           |           | body out to the extremities)                         |
| Flexion   | Extension | "From bent to straight"                              |
|           |           | A person has to feel stable in a position (like they |
| Stability | Mobility  | won't fall over) before they can use that position   |
|           |           | as a base from which to move. A child has to         |
|           |           | stand first, then he can learn to walk.              |

### 7. GROSS MOTOR DEVELOPMENT occurs:



| TYPICAL                                | IN CHARGE SYNDROME  |
|--|---|
|  |   |
| Healthy child                          | Many surgeries and hospitalizations                             |
| Fed by mouth, so okay to engage in     | Fed via G-tube or other type of system, so it may not be        |
| tummy time without discomfort          | comfortable to spend time on the tummy                          |
| Normal muscle tone                     | Low tone – have to work harder to activate muscle               |
|  | movement  |
| Normal joints                          | Joint laxity due to low muscle tone                             |
| Normal strength                        | Reduced strength due to low tone and lack of opportunities      |
|  | to move and practice during hospitalization and recovery        |
|  | times   |
| Normal amount of energy to devote to   | Lots of energy is used up during recovery from various          |
| growth and motor development           | surgeries, plus coping with cardiac, respiratory, and digestive |
|  | issues  |
| Intact sensory systems, including      | Absent and/or inefficient registration, processing, and         |
| vision, which is what motivates babies | integration of multiple types of sensory input, with a          |
| to lift their heads and begin          | particular negative influence on Balance.                       |
| strengthening extensor muscles.        |   |
| Normal movement patterns               | May see some abnormal movement patterns due to seeking          |
|  | or avoiding particular types of sensory input                   |

### Balance

- 1. Balance is the ability to:
  - Stay upright against gravity when the body is static (holding still) and when it is in motion (dynamic)
  - Transition between positions without falling over
  - Reach for something outside of our base of support without falling over
  - Protect, support, and recover when balance is challenged

2. Balance is the result of the **vestibular**, **proprioceptive**, and **visual** systems working together. The ACCURATE registration, processing, and integration of vestibular, proprioceptive, and visual information generates GOOD BALANCE.

| TYPICAL  | IN CHARGE SYNDROME                        |
|--|---|
| Normal PROPRIOCEPTION, so child can feel what his        | Poor proprioception and poor body         |
| body is doing.   | awareness                                 |
| Normal VISION, so child can get a visual fix on vertical | Visual field cuts and/or reduced visual   |
| and horizontal markers that help him understand that     | acuity                                    |
| his head is upright, or if he is falling off center.     |   |
| Normal VESTIBUALR sense, so child gets verification      | Absent or malformed semicircular canals   |
| from inner ear to confirm visual sense that his head is  | that receive vestibular information based |
| upright, or if he is 'falling off center.                | on movement of the head in space.         |
| Normal STRENGTH  | Reduced strength                          |

3. Since all three sensory systems responsible for Balance are impaired in CHARGE Syndrome, plus strength is usually reduced, it is much more difficult for these children to achieve motor milestones in the same timeframes as children without the same obstacles. They will generally follow the same sequence of development, but it will take more time. Sitting, crawling, and walking all require Balance.

### How Can We Help Support Motor Development?

| DO THIS   | BECAUSE  |
|---|--|
|   |  |
| Relax. Take a deep breath and don't fret.   | Children with CHARGE Syndrome usually do learn<br>to walk. It just takes longer than for children who<br>don't have the same challenges to contend with.   |
|   |  |
| Position the child so he or she can obtain<br>maximum physical benefit from an activity. Your<br>child's physical and occupational therapists will<br>help you with this. <i>Sometimes</i> the best position is<br>upside down. | Muscles have to be used – and used against<br>resistance – in order to get stronger. Different<br>positions will encourage strengthening of<br>different muscle groups, all of which are<br>important.   |
|   |  |
| Play, practice, and allow mistakes to happen in a safe way. Use hand-UNDER-hand facilitation.   | ACTIVE PARTICIPATION OF THE CHILD is<br>ESSENTIAL to motor learning. Nobody ever<br>learned anything by being passively moved<br>through a motor sequence. Trial and error is how<br>the brain makes adjustments to a motor plan to<br>make it more efficient and effective. The more<br>practice, the better it gets at sending more<br>accurate instructions out to the muscles. |
|   |  |
| Remember that the MOTIVATION TO MOVE has to be STRONGER than the FEAR OF FALLING (so you better make it FUN!).  | MOTIVATION<br>Fear of falling<br>We move in order to work, play, explore, and to<br>take care of ourselves and others. We have to<br>WANT to MOVE - Otherwise, none of us would<br>have learned to walk.   |
|   |  |
| Celebrate each small bit of progress along the way.   | Children who have CHARGE Syndrome usually<br>have a strong will and lots of determination.<br>These are GIFTS that will help your child to<br>overcome many of the obstacles that CHARGE<br>Syndrome presents.   |

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8-2-2019 14<sup>th</sup> International CHARGE Syndrome Conference Dallas, TX, USA

### CHARGE 102 CHARGE SYNDROME AND BEHAVIOR CHALLENGES

Laurie S. Denno, PhD, BCBA-D, LABA Behavior Analyst Behavior and Learning Consultation laurie.denno@verizon.net

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### Common Behavioral Challenges in CHARGE Syndrome

- Behavioral phenotype
- Very common in genetic conditions
- Predisposition not a certainty
  - Not everyone with the condition will show the behaviors

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### Examples

- Down syndrome higher probability of depression
- Prader-Willi syndrome higher probability of over-eating
- Williams syndrome musical ability and high social engagement
- CHARGE syndrome
  - 1. Generalized anxiety disorder (GAD)
  - 2. Obsessive compulsive disorder (OCD)
  - 3. ADHD
  - 4. Sleep difficulties

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### Definitions

From the Diagnostic and Statistical Manual, 5<sup>th</sup> edition, (DSM-5) Generalized anxiety disorder (GAD)

- The presence of excessive anxiety and worry about a variety of topics, events, or activities. Worry occurs more often than not for at least 6 months and is clearly excessive.
- The worry is experienced as very challenging to control. The worry in both adults and children may easily shift from one topic to another.
- The anxiety and worry are accompanied with at least three of the following <u>physical</u> or cognitive symptoms (In children, only one symptom is necessary for a diagnosis of GAD)

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#### Definitions

**OCD** - Recurrent and persistent thoughts, urges, or impulses that are experienced, at some time during the disturbance, as intrusive and unwanted, and that in most individuals cause marked anxiety or distress

ADHD - individuals younger than 17 must display at least 6 of 9 inattentive and/or hyperactive impulsive symptoms listed in DSM-5.

Insomnia (sleep disorder) - dissatisfaction with sleep quantity or quality, associated with one (or more) of the following symptoms:

Difficulty initiating sleep.

Difficulty maintaining **sleep**, characterized by frequent awakenings or problems returning to sleep after awakenings.

- Many students with CHARGE display challenging behavior of all types: tantrums, crying/whining, hitting, silly behavior that makes parents and teachers unhappy, self-injurious behavior (SIB), self-stimulatory behaviors and frequently, repetitive behaviors
- These are challenging behaviors that many youngsters display. They are not unusual. They are usually not of huge concern unless the frequency and intensity is high and interferes with learning or other activities
- These types of behaviors, from a behavioral perspective, are usually displayed to get something (attention, toys, computer time, snack, drink, stimulation, etc.) or avoid something (the doctor's office, hard work at school, peers who tease or adults who yell or are likely remove preferred items)
- However the number of individuals with CHARGE who display one or more of these behaviors is high: BE AWARE

### **Behavior is Complex**

- · Many behavior challenges are directly related to the
- environment
- Parent/child interactions
- Being "spoiled": getting what they want by screaming
- Getting all the best attention for "being bad" or "being silly"
- · Some behavior challenges may be directly related to CHARGE
  - Lining up all the toys over and over
  - Emptying out all half full containers
  - Awake during the night
  - Repetitive questions about the same thing

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- Some behavior challenges are a combination of environment and CHARGE
  - Individual is missing communication cues because hearing is poor or he can't see the sign language
  - Individual is constipated and is irritable and he doesn't follow directions or avoids activities
  - Individual has an undiagnosed ear infection and poor communication skills. He starts to hit his head (maybe it competes with the pain)

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• It is important to recognize the <u>sensory</u>, <u>communication and</u> <u>physiological factors</u> that contribute to these difficulties.

 In order to make changes for the better, look at behavior through the lens of the <u>science of behavior</u>

Applied behavior analysis (ABA) is the science of behavior, with a 70 year history of of studying how individuals learn to *do* or *not* to *do* a behavior

- The science of ABA builds on what others have learned in the past to extend knowledge and understand new relationships between behavior and the environment (both internal and external)
- People who practice this science are usually called **Behavior Analysts**, but other professionals also can practice the science such as psychologists, educators, guidance counselors and medical providers and business leaders

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### What is ABA?

Behavior analysts look at behavior in the context of the environment, sometimes to decrease troublesome behavior but primarily as a teaching strategy. The GOAL is to teach socially significant behavior

#### First steps:

- Define behavior in clear, observable terms. Be specific. What does the individual do? Example: Don't' say "aggressive," say "hit, kick, hair pull"
- 2. Contextual variables are important: who is there, what are the requests, materials, noise, lighting, past learning history, etc.
- Is this behavior related to inability to communicate, inability to understand, pain, fatigue, sensory deficits, sensory overload?
- 4. What happens before the behavior of concern and what happens after the behavior of concern?

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- The first steps outlined above are combined into an initial assessment
- If the assessor is experienced and skilled, a hypothesis about why the individual does the behavior is developed: this is called a "functional behavior assessment" (FBA). It is why the behavior occurs.
- Once the assessment is completed, the behavior analyst has a basis for designing a plan
- No plan should be designed without a complete assessment

- In educational settings the plan is called a behavioral intervention plan (BIP) and in adults services, often a behavior support plan (BSP)
- Every BIP/BSP is based on the unique characteristics of the individual and the unique environment with the individual spends his time
- There is no one correct response to aggression, repetitive behavior or work refusals. It depends on why the individual does the behavior, the "function" of the behavior

### **Functions of Behavior**

Two main things happen when challenging behavior is observed: individuals either get something or avoid something

- 1. Attention: social feedback, eye contact, reprimands, touching, bargaining, yelling, reasoning, "little talks"
- 2. Tangible items: toys, food, electronics, games, trinkets
- 3. Sensory input: lights, noise, deep pressure, movement
- Escape/avoidance: less preferred things such as work, specific people and places, sometimes related to pain, discomfort, fatigue

Behavior can have more than one function

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### What To Do? Tips For Change

Making a behavior change involves rules, reminders and reinforcement

- Making changes in the environment to <u>prevent</u> the behavior
   Using prompts to <u>increase</u> the desirable behavior so reinforcement can be forthcoming
- Changing consequences (what happens after the behavior) to increase more desirable behavior (using reinforcers) or to decrease behavior (using planned ignoring if behavior is maintained by attention from adults or students)
- A rule is a stated behavioral expectation (clarify your expectations)
- A <u>reminder</u> is a prompt about what to do and when to do it
- <u>Reinforcement</u> will increase the behavior that it follows (reinforcers are also unique to individuals and should be delivered immediately after the behavior you want to increase, even if you use a reminder)

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#### **Tips for Generalized Anxiety Disorder**

- Tell the individual what to expect
- Use pictures if necessary
- Make a schedule and use it all the time
- · Follow a routine as much as possible
- Give advance warning of changes, including what will happen instead of the regular thing. Say it more than one time.
- Give advance warning of unpleasant activities and schedule a preferred activity after a less preferred activity (first, the thing you don't like and then, then the thing you love)
- In some cases, anti-anxiety medication can be very helpful in "taking the edge off" and making the above strategies more effective

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### Tips for Obsessive Compulsive Disorder (OCD)

#### Diagnosing OCD

- In a study of 29 students with CHARGE, using the Compulsive Behavior Checklist, CBC, 72% displayed repetitive behavior that resembled OCD and for most of those, the behaviors significantly interfered with their daily routine.
- CBC has five categories with specific compulsions listed: ordering, completeness, cleaning, checking/touching and unusual grooming behaviors
- OCD is very, very common in CHARGE: it can be difficult to diagnose because many individuals with CHARGE cannot report having intrusive thoughts, only display the compulsive behavior. Both things are part of the diagnostic criteria (DSM-5)

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#### Tips for dealing with OCD

- Keep the individual busy with useful and fun activities
- Prompt away from behaviors either verbally or physically
   BEWARE: in some cases this may result in behavioral escalation
- Use strategies for generalized anxiety disorder: OCD is an anxiety disorder
- Allow OCD behavior during specific scheduled times when it will not interfere with learning
- In some cases, a prescription for medication (often an SSRI) can be very helpful in reducing these behaviors and/or making strategies more effective
- Finding the best medication for OCD can be a trial and error process. <u>Schedule a visit with a psychiatrist and follow dosing</u> recommendations for the best results

### Tips for ADHD

- Order, structure and organize the day
- Provide extra cues and prompts in the environment (text, pictures, rules, calendars, visual or tactile aids)
- Allow for lessons/activities with frequent breaks. Use reinforcers for completing tasks
- Teach individuals to monitor their own behavior and request breaks when they notice their attention is wandering or they start to fidget
- Add vigorous exercise throughout the day, especially at the beginning of the day. Schedule periodic stretching and movement.
- In some cases, ADHD medication can be very helpful in improving attention and activities completed

### **Tips for Sleep Disorder**

Sleep disorders are very common in people who are blind or have low vision, both falling asleep and staying asleep. It is important to practice good "sleep hygiene"

- Go to bed and get up at the same time every night
- Follow a consistent, quiet, relaxing routine before bed
- Refrain from all screens (TV, computer, video games, tablet, smartphone, etc.)
- Sleep in a cool, dark, quiet room
- Avoid caffeine and large meals before bedtime
- · Exercise during the day but not right before bedtime
- In some cases, sleep medication can be very helpful in improving asleep. Work with a physician to select the best type for your individual's sleep issues.

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#### Beware

- This is a very complex process
- Not all "behavior consultants" know what they are doing
   Many people skip the assessment piece which is a big mistake
  - They throw interventions at behaviors and sometimes they get lucky and they work. Sometimes they make things worse.
- Get qualified help, use a professional BCBA if you can find one. They are licensed in 26 states
- Go to <u>www.BACB.com</u> and search by zip code. There are 35,000 BCBAs in the US. They will not know about CHARGE but they will know about behavior and how to shape it. Behavior is learned the same way by all individuals.

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