



**Friday, July 31, 2015
Breakout Session #17: 2:15 - 3:15pm
Schaumburg E-F**

CHARGE 102

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

Presenter Information:

Nancy is an Assistant Professor of Psychology at Delta College in Bay City Michigan and a School Psychologist. She has authored articles and book chapters about developmental outcomes for individuals with CHARGE syndrome. Her young adult son Jacob has CHARGE syndrome and lives in his own home. Nancy has been involved with language and education of children with CHARGE for 20 years. She advocates for individuals with disabilities, teamwork, thorough planning, and forward thinking for quality life outcomes for all individuals.

Presentation Abstract:

Nancy will present an overview of what we know about behaviors and developmental outcomes in CHARGE and introduce communication options

COMMUNICATION

Communication, communication, communication!!!!

After Medical/Health Issues...

- Enabling expressive and receptive communication for the individual with CHARGE syndrome is the most crucial goal. It underlies all other learning.

Language: What's so hard? Isn't it a Natural Process?

**Sure it was, for you and most of the people you know. But
Imagine...**

- you were just born into the world.
- you are hooked up to tubes, monitors, and pumps, some of which are painful.
- you have just been given a tracheostomy or gastrostomy.
- you have been in the hospital for weeks on end.
- you are recovering from surgeries, tests, and procedures, with pain and fatigue.
- your body isn't working right and you just plain don't feel good.

Now: How "ready" are you to learn?

Let's add a few things: Imagine...

- your vision is limited: You can only see through one eye, or part of one.
- your hearing is limited: You can't hear much from one ear, and nothing from the other.
- you cannot move to what you want to explore: Your muscles are weak; you can't tolerate the feel of the carpet/floor on your hands or feet.
- your sense of balance is limited or nonexistent. What you see moves around and you can't get oriented. Being held and carried may even cause you distress.
- all of this doesn't allow for growthful parent/baby interaction.

Now: How "available" are you to learn?

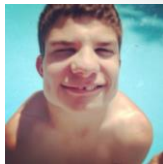
Factors affecting language development in CHARGE syndrome

(Adapted from Swanson, L., in CHARGE Syndrome, 2011)

- Vision and mobility/balance
- Hearing loss
- Facial palsy may cause difficulty with speech
- Tracheostomy: problems vocalizing
- Difficult to stimulate language early because of medical issues
- Language delays may affect cognitive skills
- Parent-child interactions: difficult when the child is less available because of sensory/medical issues.
- Serious illness and hospitalizations

Distance Senses

- Smell
 - Can identify a person
 - Can evoke strong memories
 - Is smell available? Not always, in CHARGE syndrome



Adapted from <http://www.dproject.mn.org/commbubble.html>

Distance Senses

- Hearing
 - Important for understanding oral language
 - Also gives environmental cues
 - How much hearing is available? From one ear or both? How much does an ear infection affect this? Do they wear hearing aids or cochlear implants?

Adapted from <http://www.dbproject.mn.org/commbubble.html>

Distance Senses

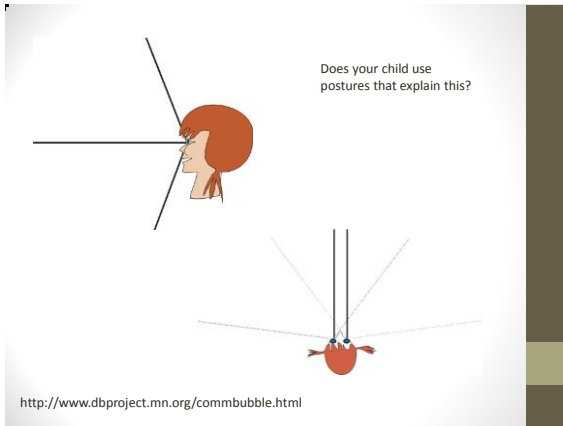
- Vision
 - Tells you what is in your environment
 - Tells you if things or people are moving toward you or away from you, or standing still
 - Tells you if people are trying to communicate with you through sign or gestures
 - How far up, down, and side to side can they see? Do they have blind spots? How far away can they see clearly? How does lighting affect their vision? What about contrast? These things may be able to be determined through a Functional Vision Assessment.

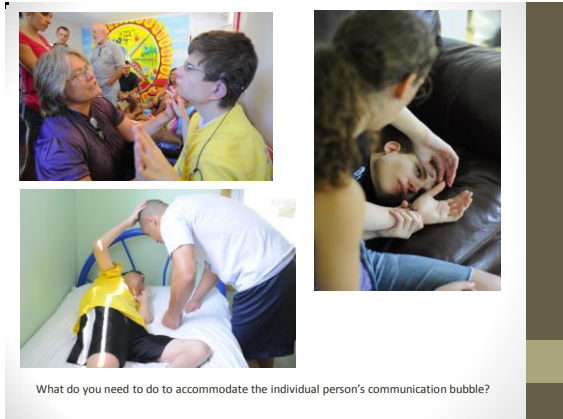
Adapted from <http://www.dbproject.mn.org/commbubble.html>

The Communication "Bubble"

"A child with CHARGE may not see you unless you are at a specific distance and in the individual child's visual field, or the child may see only parts of you and not as a person. These children also may not hear your natural voice, or they may not hear you at all. These same children may not smell properly in order to identify food, perfumes, or other common odors and may not have enough balance to attain normal motor milestones. These children are *input impaired*."

<http://www.dbproject.mn.org/commbubble.html>





Communication/Language in Children with CHARGE Syndrome: Some data

Makes reactions or noises or behaviors which can be difficult to interpret	n =20	16.1%
Uses behaviors such as gestures, sounds, body movements	n = 12	9.7%
Uses single words, signs, picture symbols, or object symbols to represent basic needs	n =15	12.1%
Uses some 2- to 5-word phrases and sentences using speech, signs, picture symbols, etc.	n = 17	13.7%
Uses verbal or sign language in complete sentences	n = 59	47.6%

All children were 4 or older. Hartshorne, T.S., Unpublished Data

Communication Modes Used by Individuals with CHARGE Syndrome (Adapted from Swanson, L., CHARGE Syndrome, 2011.)

- **Manual Sign**
 - *Sign Language*: The "official" language used by Deaf culture in your country
 - *Signing Exact English*: Using English word order, instead of the "language" of sign
 - *Cued Speech*: Using gestures while speaking to enhance understanding of lip-reading
 - *Fingerspelling*: Spelling out words letter by letter using a signing alphabet
 - *Tadoma*: a method of touching a person's face and throat to feel what they are saying
 - *Home sign*: Signs used/invented by an individual that are specific to them

- **Speech**
 - Using verbal forms of language, both formal and informal
- **Visual Symbols**
 - Using objects, pictures, or textures to communicate
- **Voice Output Communication Aids (VOCA)**
 - Using electronic equipment to communicate a message
- **Gestures and Vocalizations**
 - Pointing, showing, push-pull, vocal noises to communicate, without the use of signs
- **Idiosyncratic Behaviors**
 - The individual's own form of communication when other forms are not adequate, available, or accessible. Examples: crying, smiling, pain behavior, or any other way of trying to get a message across.

- **Total Communication: recommended for ALL**
 - The use of any form of communication available to the individual. Often, individuals choose to use more than one form.
 - For example, a person may use some sign language to answer questions, some picture symbols to ask for food, some push-pull to show you what he/she wants, and some idiosyncratic behaviors when in pain, because that is what works for them.
 - There is a growing consensus to allow for this to happen, as any communication helps the individual to be heard, no matter how it's done.
 - Limiting the individual to only one mode of communication may cause frustration as it blocks their ability to make themselves heard.

Some General Guidelines

- Let the child take the lead in communication: follow them. Get into their world; don't force them into yours.
- Turn-taking is an important first skill in communication, even if there are no words involved. How? Respond to signals to get a conversation started, just like with a developmentally typical baby; mimic sounds and gestures, extend them, expand them. Don't ignore them. All behavior is communication!

- Validate the individual's attempts at communication. Don't assume you know what they are going to say. Let them say it before you respond.
- Make sure the individual has access to all of their communication tools at all times, (all pictures, all devices) even if the answer to their request is "no." Never take away their ability to use their words, in whatever way they use them.

- Use total communication: Allow/encourage the individual to tell you what they need to tell you in any way they can.
- Ask for clarification if you are not sure what the individual is saying. Use a consistent signal/sign/word for "What?"
- Use calendar systems: help the individual to anticipate what will be happening during the time period. This helps to alleviate anxiety, to give them choices about their activities, and to understand time: before/during/after.

Behavior in CHARGE Syndrome

Common Deafblind Behavior

- Eye pressing
- Finger flicking
- Rocking
- Tapping body/objects
- Self-injurious behavior
- Mouthing objects
- Tactile defensiveness
- Clinging
- Spinning
- Vocal tics
- Feces smearing
- Lining things up
- Extreme preferences
- Darting/running off
- Learned helplessness
- Submissive
- Stare at lights
- Inappropriate vocalize

We see all of these in children with CHARGE

-Beth Kennedy, DeafBlind Central

Tim Hartshorne (CHARGE Syndrome, 2011) has studied behavior and found that "certain similarities and patterns are emerging." These could possibly related to the genetic differences in CHARGE syndrome. Not all individuals with CHARGE will have all features. However, these seem to come up often.

1. Low-normal cognitive functioning: There is a wide range of abilities, but Salem-Hartshorne and Jacob (2003) found that about half have low-average to average abilities, which was surprising, given that medical reports had often said all had mental retardation.

2. Very goal-directed and persistent with a sense of humor: "Children with CHARGE syndrome seem to know what they want and persist in their intentions. This stubborn persistence helps them to learn to walk, to eat, and to achieve beyond all expectations." However, sometimes they have trouble letting go of the idea of what they want, and this can be a problem. At other times, they may be unable to communicate what it is they want, causing frustration and escalation of behavior. Happily, they often have a great sense of humor as well.

3. Socially Interested but Immature: Children with CHARGE syndrome often have trouble making and keeping friendships. At the same time, they seem to be more interested in relating to others than others who are deafblind or have autism. However, they often "have difficulty understanding other children and often miss or misunderstand social cues." Part of it may be because of their problems with vision and hearing, but they seem to have difficulty with taking turns, backing off when necessary, and playing nicely with others.

4. Repetitive Behaviors that Increase under Stress: Many children with CHARGE are diagnosed with Obsessive Compulsive Disorder because of these behaviors. OCD is an anxiety disorder, and behaviors will increase with stress. The behaviors tend to soothe the anxiety. Whether or not individuals with CHARGE syndrome have OCD is still in question. Still, they often engage in repetitive behaviors, lining things up, and keeping strict routines. It seems to make sense that with limited sensory input, the world can be a bit chaotic. Using these behaviors may simply help to keep anxiety levels down.

5. High Degree of Sensation Seeking: "Light flicking, hand flapping, body shaking, and rocking are among the self-stimulatory behaviors that are common in children with CHARGE." Although these behaviors can get in the way of learning at times, they seem to serve a function for individuals with CHARGE syndrome. Because they often lack sensory stimulation from vision and hearing, these behaviors may serve to make up for that lack, to keep the brain alert (just as you or I might tap a pencil, chew gum, or sway our calf back and forth to keep alert.) Some individuals with CHARGE may simply need a sensory break to pull themselves back together before working again. "A hard-working student might suddenly drop everything and shake his or her whole body for a moment, and then be able to go back to work."

6. Under Conditions of Stress and Sensory Overload, Find it Difficult to Self-Regulate and Easily Lose Behavioral Control: Most of us are able to self-regulate well. We can concentrate even though there's a little bit of noise in the background, or we know that when we are overwhelmed in a noisy environment we need to get someplace quiet. Everyone is different in what they can tolerate to their senses, and in how they deal with it. Individuals with CHARGE have difficulty regulating this. Sometimes they may want extra stimulation, such as rubbing their hands on a textured surface repeatedly. Then, suddenly, it becomes too much. The sensation "might send them over the sensory edge and cause loss of behavioral control." They may crave the sensation of staring at a colorful disco light toy, but then knock the light away when it becomes too much.

7. Difficulty with Shifting Attention and Transitioning to New Activities: Easily Lost in Own Thoughts.

Many individuals with CHARGE syndrome become very focused internally. This may in part be because of sensory impairments, but there seems to be more. They seem to have difficulty shifting their attention, and "moving freely from activity to activity."

Additionally some may have difficulty initiating new activity. They may want to, and may be ready, but might have difficulty getting started.

They may also have trouble with monitoring their own behavior and how it affects others.

These individuals will need assistance to learn to do these things.

Behavioral Threshold:

This is the point at which someone loses control. Consider anger in you or me. Most people may be able to control their behavior when they are angry, but then someone does or says something, and it's the "last straw," and they may lose control.

Likewise, an individual with CHARGE syndrome may hold it together as long as they are under the behavioral or sensory threshold. Once things become too much, they can have an abrupt change in behavior, seemingly coming out of the blue.

Three things can put an individual with CHARGE syndrome over the threshold for behavior:

1. Sensory stimulation: As stated before, what begins as a welcome need for stimulation can often escalate to be too much. It can change from "pleasant or bearable to painful and upsetting very quickly."
2. Buildup of stress in the environment to the point that they can no longer cope. Consider all of the medical interventions they must go through, for example. In addition, consider the mere exhaustion factor when trying to see, hear, stand, walk, and all of the other things these individuals must work hard at, all day.
3. Pain: This may be the most important thing to look at. Often, pain is subjective. We cannot see it. A child with CHARGE may not be able to tell us that their throat or ear hurts, or that they feel achy. There are many other types of pain that are specific to CHARGE syndrome (see next slide). A sudden change in behavior without explanation should always lead to an immediate investigation of what may be causing the person pain.

Frequent Sources of Pain in CHARGE

- Otitis Media (middle ear infections)
- Sinus infections
- Migraines
- Abdominal Migraines (migraines felt in the belly)
- Gastroesophageal reflux disease (GERD)
- Gas
- Constipation
- Muscle pain
- Tactile defensiveness (too much sensation at the skin level)
- Stoma pain (inflammation around the skin at the entrance of the feeding tube in the stomach.)

Why pain is so important

- Can affect normal brain and nerve development
- Can affect sleep
- Can interfere with exploration of the environment and learning
- Can interfere with the development of attachment and trust
- If they begin to get used to it, it may affect their health if not taken care of.

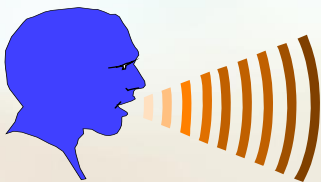
(Hartshorne, T.S. 2012)

When you are in pain, do you ...

- Whine?
- Complain?
- Seek attention?
- Make demands?
- Act out?



ALL BEHAVIOR IS COMMUNICATION



Sometimes behaviors make perfect sense!

This is normal viewing posture...

...when you have
no vestibular
sense, upper
visual field loss,
poor tactile &
proprioceptive
perception, & low
muscle tone.

(-David Brown)



How can you help?

- Communication, communication, communication
- Always interpret behavior as communication.
- Develop a pain management plan.
- Figure out if the behavior serves a purpose for the individual. It doesn't always have to be changed or eliminated!
- If it is determined that the behavior should be changed or eliminated (maybe they are injuring themselves or others), we must teach them alternative ways to communicate their needs.
- Sensory diet: deep pressure/squeeze hugs, brushing and joint compression (ask your occupational therapist for help). These things can help regulate the sensory and behavioral thresholds by getting the nervous system reorganized.
- Break down larger tasks into smaller, more manageable chunks.
- Help the individual learn ways to self-soothe.
- Build sense of safety and trust
 - Consistency
 - Routine
 - Calendar System

EXAMPLES

David Brown (2005).
CHARGE Syndrome
**"Behaviors": Challenges or
Adaptations?** *American
Journal of Medical
Genetics*, 133A, 268-272.



- A young child who was said to be self stimulating "all the time" was actually practicing and developing his mobility and orientation skills, and using vision and touch to explore objects, very creatively. While doing this he needed to get onto his back on the floor to reorganize his sensory system with brief episodes of limb shaking and hyper-ventilating every 10 to 20 minutes. (sensory break)

- A kindergartener was often self-abusive when he got distracted and over-aroused by incidental touch and air movement caused by people repeatedly walking behind his chair. Once his chair was placed with its back securely against a wall he was less self-abusive and more amenable to social interaction. (Too much stimulation)

- People were concerned when a young boy began to insist on the unusual idea of wearing band-aids wound tightly around the tips of all his fingers and thumbs every day. He was expressing his need for more and stronger pressure and touch inputs as a part of his sensory diet. (Needed more pressure/stimulation)

- Every morning in a pre-school program a student refused to sit on the floor with her class to watch the teacher sign a story. When an appropriate chair was provided the student sat and attended with great interest and a growing level of participation. (balance problems)

- A girl was described as very disruptive during sessions that required the class to sit still and participate in a signed conversation with the teacher for up to 30 minutes. When the teacher used a strategy of asking the student to move periodically to carry out small chores during these sessions (to fetch a pen, open a door, bring a book, take a paper to the school office) the disruptive behaviors largely ceased. (visual fatigue)

- A teenager enjoyed, and was quite good at, soccer in the school gym, but was unwilling or unable to play it outdoors due to the absence of strong vertical visual markers to aid equilibrium. The ability to participate in such complex physical activities outdoors did not develop until significant adaptations were introduced. (need for visual “handles” in the environment to know where his body is in space)

- During Orientation and Mobility sessions a teenager was refusing to stand still to receive spoken/signed instructions, but the problem was solved when the student was allowed to stabilize himself by leaning against a pole or a tree or a wall, or by placing one hand on the instructor's shoulder during these conversations.

(balance problem/stabilizing body/head)

Developmental Considerations

Three Developmental Domains

- In studying human development, we look at three domains:
- Physical Development
- Cognitive Development (Intelligence)
- Social/Emotional Development

Physical Development

- We've already discussed the physical differences in individuals with CHARGE syndrome. We have discussed senses, sensory systems, surgeries, medical problems, and many other aspects. What's left?

- **Motor Milestones**

Normal Gross Motor (Large Muscles) Milestones

- What's typical for a non-CHARGE baby?
 - Hold head up
 - Roll over
 - Sit up
 - Crawl on belly, then on all fours
 - Pull to stand
 - Cruise holding onto furniture
 - Walk independently (Average Age=12 months)
 - Stairs, hopping, running, skipping, etc.

CHARGE Gross Motor Milestones

- Arching back
- Back scooting (rub hair off back of head)
- 5-point crawl (four limbs plus head)
- Sitting
- Bottom Shuffling
- Crawl
- Pull to Stand
- Cruise around furniture
- Walk independently (average age = 42 months)
 - (Age ranges from 30 to 84 months)
- May continue to have unsteady gait

With what you know...

- About sensory, vestibular, and other CHARGE systems, can you hypothesize why these milestones might occur differently?
 - Visual field reasons?
 - Vestibular reasons?
 - Keeping head stable reasons?
 - Tactile defensiveness reasons?

Vestibular

- We use eyes, joint pressure, and semi-circular canals to balance. Many children with CHARGE have little vision, and malformed or missing semi-circular canals. In order to walk, they must rely on joint pressure, primarily. Does this explain the wide, slow, toddling gait many have?

Normal Fine Motor (small muscles) milestones

- Suck, swallow
- Eat puree
- Chew, bite
- Grasp with whole fist
- Grasp with finger and thumb
- Manipulate/trade objects between hands
- Hold/draw with crayon, etc.

With what you know....

- About eating, tactile defensiveness, and low muscle tone, what might this mean about fine motor milestone achievement for children with CHARGE syndrome?

- Early tactile defensiveness (inability to tolerate some sensory input to skin) limits how much touch and object manipulation the child can handle without going over the "sensory threshold." This may delay fine motor milestones.
- Of course, eating/swallowing/breathing issues will delay eating milestones.

Cognitive Ability

- We gave the Adaptive Behavior Evaluation Scale to 100 Children with CHARGE
- They showed a higher range of ability than once thought: 54 had scores higher than 70 (Average score is 100. Below 70 is typically considered mental retardation)
- Those who walked earlier, had fewer medical problems, and had better hearing and vision scored higher. Age at walking had the strongest result.

Salem-Hartshorne N, Jacob S. 2004. Characteristics and development of children with CHARGE association/syndrome. *Journal of Early Intervention* Vol 26: 292-301.

What does this mean?

- It means that **we can/should have high expectations** of these children from the beginning, because we absolutely cannot tell what the outcome might be, based alone on how sick they are as babies.
- It **MAY** mean that we can possibly predict how well a child will do based on when they start walking. (This is definitely not set in stone, but has been found in other studies as well.)

Age at Walking? Why?

- The ability to walk depends a great deal on the vestibular system. Some children may have more severe problems there, and therefore walk later.
- It's not walking age that causes problems, though. The vestibular system can affect a LOT of things that have to do with learning. Walking is just one thing it delays.
- So, does having more problems with the vestibular system mean the child may have more developmental problems, overall?
- It's what we are beginning to think, and it makes sense, but there is no clear proof of this.

Social Skills Deficits

- Acquisition deficit - never learned the skills
- Performance deficit - prefers not to use the skills
- Fluency deficit - knows the skills but not very good at applying them

Most children with CHARGE appear to have a fluency deficit rather than the other two.

This means:

- They know what to do.
- They want to do well.
- They have trouble doing it well.

Social Skills they Need

- Getting along with peers
 - Managing their behavior
 - Classwork skills
 - Cooperation
 - Being assertive
- They will need practice, guidance, and support to do well at these things.

- Management of emotions can be difficult, because of that emotional threshold that is difficult to come back from. Melt-downs may be common.
- Need to teach them strategies for self-soothing, talking to themselves about what's going on, and recognizing when they are approaching the threshold.
- How much is emotional management associated with sensory overload? We do not know. But sensory breaks seem to help.

ATTACHMENT/BONDING



Infant Attachment Problems

- It's difficult to attach when there is little to no eye contact, and no hearing of one another's voices, between parent and baby. Add in tactile defensiveness (not wanting to be touched), and it can become very hard.
- Medical procedures, and just trying to stay alive can have a huge effect as well.
- The shock of having a baby with a disability can also play a part.
- Many parents report delayed bonding with their child with CHARGE syndrome.

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

Toileting

Salem-Hartshorne, Blake, and Hartshorne (CHARGE Syndrome, 2011) surveyed information for 44 children with CHARGE, ages 9-21.

Independence with urine 73%
The rest needed assistance, reminders, or did not use the toilet

Independence with bowel 65%
The rest needed assistance, reminders, or did not use the toilet

Independence with overnight 54%
The rest needed protective undergarments, reminders, or physical help.

WHY?

Prerequisites for Toilet Training

Denno, L. CHARGE Syndrome, 2011

- Feel the need to eliminate
- Walk to bathroom
- Pull down pants and underpants
- Sit on toilet
- Eliminate in toilet
- Use toilet paper
- Put toilet paper in toilet
- Stand up
- Pull up underpants and pants
- Flush toilet
- Wash hands
- Dry hands
- Exit bathroom

For what reasons might these skills be difficult for an individual with CHARGE syndrome?

Balance/mobility (if you can't balance, it's difficult to undress standing up.

Can the individual button/snap/zip?

Inability to hear urine stream

Inability to smell feces

May not be socially motivated to try

There is some discussion of the possibility that they may have limited sensation because of problems with the nerves or muscles

Finally, they will likely get a late start because of all of the medical/physical problems (walking late, for example)

So....

Toilet training is likely to happen later with individuals with CHARGE.

Some may never be totally independent.

It is still a really important teaching opportunity, in terms of communication, turn-taking, and development of independence.

SLEEP.....ZZZZZZZZZZZZZZZZZZZZ

Over half of children with CHARGE syndrome have sleep problems.

Why?

- Deaf-blindness: It is well-known that darkness cues the brain to release melatonin, a hormone that causes sleepiness. If you are blind or visually impaired, the darkness may not work as a cue. Therefore, is the melatonin released? Some parents have found over-the-counter melatonin to be helpful in inducing sleep, pointing to a possible deficit.
- Pain from ear infections/other problems keep child awake
- Behavioral difficulties and anxiety (think of that threshold!) leading to troubles settling at night
- Obstructive sleep apnea: Large tonsils/adenoids, choanal atresia, clefts, and other physical anomalies may get in the way of breathing while asleep. This can "fragment" sleep, so that good quality sleep is not obtained, night after night.

Effects of too-little sleep

Think about how you feel when you haven't had sleep.

Some of these children may be having that effect compounded by having fragmented sleep night after night.

Fragmented sleep means they may never enter the Rapid Eye Movement stage....this is when protein is synthesized, dreaming takes place, and the brain regenerates.

Do you think there are behavioral effects of exhaustion?

What to do?

(Heussler, H. S., [CHARGE Syndrome](#), 2011)

- First, take care of obstructions. Surgery to remove tonsils and adenoids, or other medical management may help.
- Teach good sleep hygiene. This includes using a regular bedtime and routines (bath, read a book, teddy). The child will begin to associate these things with getting sleepy.
- Pay attention to sensory preferences. Sometimes a heavy blanket, temperature, or other things will help a child with sensory integration difficulties settle in.
- Try to make dark/light, day/night contrast as clear as possible.

ADOLESCENT DEVELOPMENT

The CHARGE Experience



Kim Blake, M.D.



Nancy Hartshorne, Ph.D.

Adolescent and Adult Issues

Blake KD, Salem-Hartshorne N, Abi Daoud M, Grandstein J. 2005. Adult and adolescent issues in CHARGE syndrome. Clinical Pediatrics. 44:151-159.



- ❑ 30 participants
- ❑ 16 female; 14 male
- ❑ Age range 13 to 30 years, mean 17.6

Medical Issues Found	Number	%
Scoliosis	19	63
Sleep Apnea	13	43
Abdominal Colic (Gas Pain)	12	40
Retinal Detachment/Cataract	10	33
Migraines	8	27
Seizures/Epilepsy	5	17
Urinary Tract Infections	5	17
Hypoglycemia	1	3

Osteoporosis also common

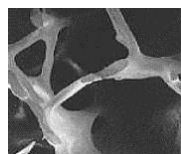
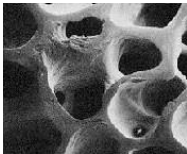
- 15 males and 15 females, ages 13 to 34 (average 19.6)
- Traumatic bony fractures were identified in 30% of the sample
- Delayed puberty was experienced by 87% with only 4 individuals (2 female, 2 males) having experienced normal puberty
- Feeding difficulties, inactivity and delayed puberty are all problems that lead to the development of poor bone health in CHARGE syndrome.

Forward KE, Cummings EA, & Blake KD. (2007). Risk factors for poor bone health in adolescents and adults with CHARGE syndrome. *American Journal of Medical Genetics, Part A*. 143A(8), 839-45.

Osteoporosis

Osteoporosis, or porous bone, is a disease characterized by low bone mass and structural deterioration of bone tissue, leading to bone fragility and an increased susceptibility to fractures, especially of the hip, spine and wrist, although any bone can be affected.

Healthy and Unhealthy Bones



Puberty Development

- Often delayed
- Girls tend to go into spontaneous puberty more often than boys, although it is usually delayed.
- Boys will often need hormone treatments to go through puberty.
- It is very important, medically, to go through puberty, to avoid osteoporosis. During puberty, we lay down bone mass.

Behavior can still be a difficult area in Adolescence

Behavioral Concern	Number	%
Aggressiveness/outbursts	16	53
Self-abuse	15	50
Sleep problems	15	50
Tactile Defensiveness	12	40

How independent were the kids in the study?

Degree of Independence (N=30)

Activity	Degree of Independence					
	None	Little	Some	Most	All	N/A
Dressing self	10		2	5	13	
Toileting self	10		4	2	14	
Washing self	10	4	3	5	8	
Getting to/from work/school	15	1	2	3	6	3
Cleaning	12	4	7	3	3	1
Cooking	16	5	7	2	2	2
Shopping	19	3	3	2	2	1
Finances	25	2	1		1	1

Can you see that there is a bit of a split? About a third have lots of trouble. The rest are learning or are somewhat independent. This gets again at the 50% who have low normal to normal intelligence.

Final Words from David Brown

"There is no other identified sub-group within the population of people with multi-sensory impairment who have so many medical problems, of such complexity and severity, and with so many hidden or delayed difficulties, and yet no sub-group has shown such a consistent ability to rise triumphantly above these problems."