



Saturday, August 3, 2019

Breakout Session B10 • 1:30-2:30 pm • Chantilly Ballroom East

## “Why are Children with CHARGE Syndrome So Lazy?”

David Brown, Deafblind Educational Specialist

### Presenter Information

David Brown is a deafblind educational specialist who has been working with children with CHARGE syndrome since 1983. In the United Kingdom he was the Head of Family & Children Services for Sense. He moved to California in 2000 to work with the state deafblind project, based in San Francisco. He retired in 2014 but still works extensively as a freelance educational consultant. He has given presentations about CHARGE syndrome in 14 different countries, and in 24 states in the US. His articles about CHARGE syndrome have been translated into at least 12 different languages. In 2005 David was given the Star in CHARGE award by the CHARGE Syndrome Foundation, and in 2013 he received the Lifetime Achievement Award from Deafblind International.

### Presentation Abstract

This session will aim to provide an overview of the many issues which present challenges for the development and behavior of people with CHARGE syndrome. The connection between these issues and the behavioral outcomes they provoke is often missed or misunderstood, and the session will clarify those connections and also suggest strategies which can help to improve developmental and behavioral outcomes.

### Learning Objectives

- To acquire a better understanding of the many issues confronting people with CHARGE syndrome.
- To improve understanding of the implications of these issues for development and behaviour.
- To gain some awareness of effective strategies to ameliorate the impact of these issues.

## “Why are children with CHARGE syndrome so lazy?”

Reflections on fatigue, caution, self-preservation, adaptive abilities, function, efficient use of energy, and the ways that these can be misinterpreted

David Brown  
14<sup>th</sup> International CHARGE Syndrome  
Foundation Conference  
Dallas, Texas  
August 3<sup>rd</sup> 2019

Children with CHARGE syndrome frequently seem to be uniquely driven and goal-oriented, full of energy, curious, narrowly focused and insistent on getting what they want, and unwilling to compromise. Even though children with CHARGE can be lazy, the way all of us can be at times, “lazy” is just about the last word I would use in any description of them, so it surprises me how often I hear the word “lazy” used when people talk about them. The word usually crops up in connection with the fact that a child is not yet walking unsupported even though it looks as if they could, or sometimes because they often like to stretch themselves out in the horizontal position, or they rest the side of their head on the table for periods of time. The word “lazy” is almost the most commonly used adjective for children with CHARGE yet in my experience the children are almost all determined to get up and go when the time is right, to such an extent that I don’t understand why they are not much more lazy than they actually seem to be.

David Brown (2013)

- \* Multiple sensory issues
- \* Multiple health issues
- \* Fatigue
- \* Attention
- \* Priorities
- \* Interests/Motivators
- \* Suggestions

The idea that I am lazy or inactive seems anathema to who I truly am, and yet this was a label that stuck to me for a long time. It’s a label I hear other autistic people share too. Why are autistic people so often accused of laziness? Why is it something that haunts us so? Because of the exhaustion. It is my greatest and most enduring foe. The exhaustion of autism is my biggest difficulty. It is not inherent in the autism itself, it is built out of how the world works. I know this, because when I am at home, in my routine and structure, in my safety and joy, I do not suffer with that soul-sapping tiredness.

Autism & Expectations [autistri.com](http://autistri.com)

### 21 CHARGE issues which can contribute to fatigue

- Visual impairment
- Hearing impairment
- Vestibular dysfunction
- Poor proprioceptive perception
- Poor tactile perception
- Low/high muscle tone
- Skeletal malformations
- Breathing problems
- Poor nutrition
- Anaemia
- Hypoglycaemia
- Hypothyroidism (slow metabolism, low energy)
- Sleep problems
- Heart problems
- Constipation
- Gastro-intestinal issues
- Migraine
- Dental issues
- Joint pain (growth related?)
- Seizure disorder
- Medications
- Poor self-regulation

## Pain

RESEARCH REVIEW

WILEY DISCOVER SOMETHING GREAT  
medical genetics  
Behavioral/Health Genetics

### Behavior in CHARGE syndrome

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Unusual behavior is often associated with genetic syndromes, and may constitute a behavioral phenotype. In contrast to providing a psychiatric diagnosis, a behavioral phenotype describes what is unique to the behavior associated with different syndromes. While behaviors in CHARGE are as complex and variable as other aspects of the syndrome, there are some commonalities that raise the question of common sources for these behaviors. This article addresses how pain, sensory issues, and anxiety may impact the behavior of individuals with CHARGE syndrome, and how the development of self-regulation skills might help to mitigate some of the behaviors.

#### KEYWORDS

anxiety, behavior, CHARGE syndrome, pain, self-regulation, sensory impairment

## CHARGE syndrome

Problems with the perception of:

- Vision
- Hearing
- Touch
- Proprioception
- Temperature
- Pain
- Vestibular
- Smell
- Taste

David Brown - Am.J.Med.Gen. 2005

“Children with CHARGE syndrome are truly “multi-sensory impaired”, having difficulties not only with vision and hearing but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell, as well as problems with breathing and swallowing, eating and drinking, digestion, and temperature control.”

## Deafblindness/multi sensory impairment

“Most people with CHARGE that I have met satisfy the criteria for being considered as having deaf-blindness, even if they have some useful vision and hearing. This is a disability that is defined in functional, not clinical, terms and for each individual with CHARGE it is mostly about difficulties in accessing information not just from the world around them but even from their own bodies.”

David Brown (2011) *Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome* reSources Volume 16 Number 3

“A good and effective educational program, while being very positive and having high expectations, should always take account of the fact that everything that a child with CHARGE syndrome does is likely to take more thought, more attention, and concentration, and energy, and time for them than it does for us.”

David Brown (2011) *Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome* reSources Volume 16 Number 3

The brain is connected to the body through the senses

I believe that most children with CHARGE syndrome are not in touch with/ do not feel their bodies very well

Everything that children with CHARGE syndrome do has meaning, and the first obligation on the teacher is to ascertain that meaning (or at least to come up with a really good guess).

## What do you do?

Most people focus on the child's disabilities, but close attention to their abilities, and to the things that they do, can reveal more about the difficulties they face and the strategies they use to function effectively.

When the proprioceptive sense is not working properly one common outcome may be:

Seeking strong pressure, stretching, or twisting inputs, eg. squeezing into tight spaces, crossing or twisting limbs around each other, twisting a foot or a leg around the leg of a chair, binding parts of the body with cloth or string or rubber bands, pulling downwards on the teeth and lower jaw, grinding the teeth, tapping the teeth, hand clapping or flapping, leg swinging or kicking, hanging from a bar, jumping up and down, banging the head, hammering objects, standing on the head.

### Postures involving pressure on the head



### Postures involving the legs



“Of all the many sensory impairments associated with CHARGE syndrome, absence of, or significant damage to, the vestibular sense is perhaps the most far-reaching in its implications, the least understood, and the most overlooked.”

Brown “Consequences of Vestibular Dysfunction” in Hartshorne, Hefner, Davenport, Thelin (2011) “CHARGE Syndrome” Chapter 5, pp 51

“In the future it is likely that we will discover close links between significant vestibular impairment and many of the currently ‘unexplained’ CHARGE behavioural features such as difficulty with the self-regulation of arousal levels, sudden and apparently unpredictable mood changes, poor memory, and executive dysfunction.”

Brown “Consequences of Vestibular Dysfunction” in Hartshorne, Hefner, Davenport, Thelin (2011) “CHARGE Syndrome” Chapter 5, pp 53

## Effects of vestibular problems

- Organization of ALL sensory information
- Postural security and muscle tone
- Use of residual vision
- Perception & processing of sound
- Remembering auditory sequencing
- Memory development
- Speech/Language development
- Behavioral challenges
- Bilateral coordination
- Breathing, feeding, digestion, nutrition
- Sociability
- Fatigue
- Self-regulation

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“After air to breathe, postural security is our next most urgent priority.”

Jean Ayres

“For children with CHARGE syndrome gravity sucks”

Maryann Girardi, Physical Therapist at Perkins School for the Blind

“Why Aren’ t You Paying Attention? The Interaction of Posture & the Executive Function of Attention”

Three-part webinar by Dinah Reilly

<http://www.idahotc.com/Webinars/tabid/218/categoryid/4/5.aspx>

*P Hodges, et al (1991) Postural activity of the diaphragm is reduced in humans when respiratory demand increases Journal of Physiology 537.3: 999 1008*

Every muscle in the trunk is both respiratory and postural muscle, especially the diaphragm. If breathing is compromised, postural muscle activation is reduced to focus on immediate needs of respiration and thus posture is de-stabilized.

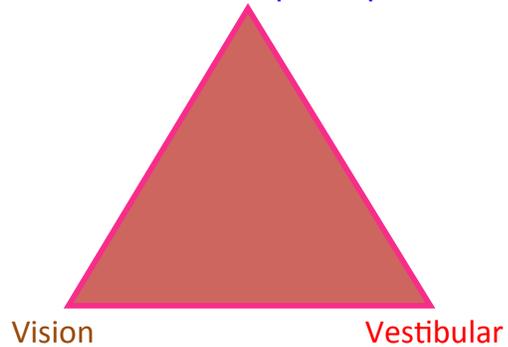
*L Yardley, et al (1999) Effect of articulatory and mental tasks on postural control NeuroReport 10:215-219*

Articulation increases postural sway in stance in adults.

You fix the body.....  
to fix the head.....  
to fix the eyes.....  
so you can use your vision  
in the best, most reliable,  
and most comfortable way  
possible.

## The Equilibrium Triad

Touch/Proprioception



## Walking

- Walkers
- Walking poles
- Rolling gait
- Flat feet/ bent knees
- Foot slapping
- Tip-toe walking
- Feet roll inwards/ knees knock together
- Arms extended to the sides or in front
- Fingers crossed/hands clenched
- Eyes fixed on a visual target ahead (holding on with the eyes)



## Functions of these postures

- To locate the body (especially the head)
- To confirm postural security
- To stabilise the body, to stabilise the head, to stabilise the eyes for visual tasks
- To ease the discomfort of chronic constipation
- To open up the airway
- To self regulate

## What helps? (1)

- Advice from PT and OT
- Activities which improve strength, muscle tone, and controlled movement, and reinforce the body/brain connection (eg. Tai Chi, yoga, wrestling, climbing, dancing, APE, horse riding)
- Deep pressure inputs (eg. jumping, massage, swimming)
- Binding (eg. spandex pressure vest)
- Good physical support & appropriate postures for efficient functioning

“Spandex is the best friend of children with CHARGE syndrome”

Maryann Girardi, Physical Therapist at Perkins School for the Blind



Yes, I believe that posture should be included as an important “self-stimulation” and “self-regulation” behavior for people with CHARGE syndrome (and all the rest of us too)

- What helps? (2)**
- Variety in postures and movement
  - Rest periods for re-organization
  - Controlled environments
  - Self-taught and taught strategies
  - Strategies that are motivational
  - Appropriate vocabulary (for body parts, for physical feelings, for emotional states, for desired activities)

- So, what am I saying? (1)**
- Everything joins up – there may be many unsuspected influences at work. Functional vision & hearing therefore depend upon, and vary in relation to, many things apart from eyes and ears.
  - There is a fundamental sensory hierarchy to which we are all subjected.
  - Vision is a directional sense so body awareness, postural control, orientation, attention, and stamina are all required (all these are probably all compromised by CHARGE syndrome).
  - We can help to prepare children to use their vision and hearing optimally by improving these things.
  - With their spontaneous behaviors, especially their postural behaviors, children might be showing us the best way to work with them, the best way to present materials, and the most energy-efficient ways to proceed.

## So, what am I saying? (2)

- The multi-sensory perspective is crucially important
- Sensory need comes first with self-stimulation behaviors, social meaning is acquired later
- The concept of the sensory diet can provide insightful and powerful strategies
- Sensory needs and sensory inputs control attentional priorities
- Living with CHARGE syndrome is difficult and tiring, so the easier we can make things the better
- Notice posture and movement requirements!
- These things apply to all of us

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**“Why are children with CHARGE syndrome so lazy?”  
Reflections on caution, self-preservation, adaptive abilities,  
function, efficient use of energy, and self-awareness, and the way  
that these can be misinterpreted.**

by David Brown  
CDBS Educational Specialist

Children with CHARGE syndrome frequently seem to be uniquely driven and goal-oriented, full of energy, curious, narrowly focussed and insistent on getting what they want, and unwilling to compromise. These characteristics can be observed even in infants who have not yet acquired any independent mobility, as well as in older children. Even though children with CHARGE can be lazy, the way all of us can be at times, “lazy” is just about the last word I would use in any description of them, so it surprises me how often I hear the word “lazy” used when people talk about them. The word usually crops up in connection with the fact that a child is not yet walking independently even though it looks as if they could, or sometimes because they often like to stretch themselves out in the horizontal position, or they rest the side of their head on the table for periods of time. The word “lazy” is almost the most commonly used adjective for children with CHARGE (“naughty” seems to be the most popular, but then it would be, wouldn’t it?) yet in my experience the children are almost all determined to get up and go when the time is right, to such an extent that I don’t understand why they are not much more lazy than they actually seem to be. How many parents have felt as if their child with CHARGE was never ever going to walk, then one day at the age of three or five or eight the child has just, apparently quite suddenly, launched themselves into space and become a walker? Readiness is all!

Very few professionals get any training about vestibular issues, and they get no training at all about CHARGE syndrome of course, so that often their rather negative comments might simply come from them being unaware of all the complexities involved for the child. It seems like a good idea to try to clarify what some of these characteristic behaviors might mean, and what they result from, so that the “I” word can be removed from discussions of development and behavior.

## **Standing and walking**

The challenge of developing good equilibrium (balance) and postural security for independent standing and walking for children with CHARGE is an outcome of many different aspects of the syndrome including visual problems, deafness, vestibular issues, low muscle tone, loose

connective tissue (the tendons and ligaments that bind the joints), skeletal issues, poor proprioceptive perception, breathing problems, poor body awareness, difficulties with executive attention, and who knows what else. Let's take a brief look at some of these considerations in turn.

## **The Equilibrium Triad**

Although the vestibular sense is often called "the balance sense," balance actually develops from the interaction of vestibular input with proprioceptive input and visual input. Since all three of these senses are usually missing or impaired in children with CHARGE it should be no surprise that independent standing and walking develop only very slowly. And while that development results from growing experience, and strength, and confidence, it also develops from a host of smart adaptive strategies that the children have to learn to use in order to compensate for what is missing from each of these three crucially important sensory systems. The references at the end of this article will all touch on these aspects of development in more detail.

## **Vestibular Issues**

These are probably the biggest and most significant obstacles for children with CHARGE to overcome and alone could explain why walking often develops quite late. One problem with developing effective standing and walking is that most children devise their own adapted ways of moving from one place to another to minimize the impact of their vestibular deficits and the challenge of resisting gravity. Movement strategies like the five-point crawl, the back scoot, side-winding, and rolling, become so safe, so quick, and so effective that risking the catastrophe inherent in standing and moving on only the two feet would be a foolish choice for the child to make until they know that they are absolutely ready for it. Maryann Girardi once told me that for children with CHARGE "gravity sucks," and we would do well to remember that when the child seems stubborn or fearful or resistant to our efforts to get them walking.

## **Vision Problems**

A child with missing vestibular sense and poor proprioception might eventually look as if their balance is quite good when they are standing, and especially when they are walking, but in fact they might be using their vision to fixate on a distant object to guide them in keeping upright and moving on target as they walk. If this is the case the child does need to maintain their gaze on that target object for the entire journey until they arrive. If they get distracted and take their visual attention away from the target during the journey they are very likely to veer off course, or even collapse to the floor like a tree being felled. The biggest problem with this over-reliance on visual input is that it doesn't work if there are no clear visual objects around on which to focus the eyes, it doesn't work in bright light with lots of glare (especially if the child has iris colobomas), it doesn't work in low light or darkness, and it prevents the eyes from moving around during the journey (known as ocular motility) to check for obstacles on the way, especially down at floor level and in the left and right peripheral visual field. Visual field loss in CHARGE is almost always in the upper visual field, so when a child regularly trips over obstacles on the floor people get confused and assume that there must be lower visual field loss as well. In fact the lower visual field might be fine, but the child cannot make their vision available for checking the floor because their vision is serving the much more urgent need to keep them

upright and moving in the right direction. As a result of all these visual factors a child might walk independently at certain times in certain places, but revert to back scooting or some other safer and easier method of moving at other times and in other places.

## **Breathing Difficulties**

In her three-part webinar PT Dinah Reilly makes many important and relevant points even though she is not speaking specifically about CHARGE syndrome. One of the things I liked most about her talks is when she mentions the way that difficulties with breathing get in the way of good postural control and balance. She quotes from an article by Hodges:

“Every muscle in the trunk is both respiratory and postural muscle, especially the diaphragm. If breathing is compromised, postural muscle activation is reduced to focus on immediate needs of respiration and thus posture is de-stabilized.”

This quote also reminds me of Jean Ayres’ comment that after air to breathe, postural security is our next most urgent priority. Given the high frequency of breathing issues in the population of children with CHARGE this connection needs to be remembered and recognized when the focus is on the development of walking.

## **Challenges for the Physical Therapist**

Maryann Girardi, at Perkins School for the Blind, has made a very interesting short webcast called “CHARGE Syndrome: Providing Physical Therapy.” She speaks from a long experience of working with children with CHARGE, and perhaps the most interesting thing about her presentation is that she spends very little time on conventional PT skills and techniques. Instead she talks about things like the importance of building a positive relationship with the child, being consistent and predictable, communicating effectively with the child, using the child’s motivators and setting realistic goals for the session, observing to ensure that the child is in a good level of arousal (not over-excited or drowsy), and allowing the time necessary for the child to be actively involved in what is going on. Her concern is not with getting the PT right but with creating a situation where her PT skills can be delivered effectively and involve the child.

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## What Does “Follow the Child” Mean?

by David Brown  
CDBS Educational Specialist

### Assessment woes

Why is it so difficult to assess children with deaf-blindness successfully? Many different people in the education/psychology field assess these children for many different reasons in many different ways, but very often the children end up being (usually) wildly under-estimated or (sometimes) just as wildly over-estimated. Clinical assessment approaches, medical, psychological, and educational have a part to play but may yield a very misleading view of the child's current abilities and developmental potential. Many people are familiar with the declaration that a child is untestable, or is too disabled or too non-compliant for any useful assessment data to be obtained, but this is the fault of the assessment approach being used rather than any failing on the child's part of course. Why is it thought preferable to make a child fit an assessment procedure rather than make the assessment procedure fit the child? Jan van Dijk has always made it very clear that we must alter our attitudes and approaches on every level when working with this population:

“The multi-sensory impaired person is a unique human being with a unique line of development, who is more dependent on the professional's willingness to accept this and act accordingly than any other group of disabled persons,” (van Dijk, 2001).

### What goes wrong?

The problems encountered by children and families in their dealings with assessors are legion, and it is not hard to identify the mistakes that are commonly made by people who are supposed to and assumed to be trained and skilled in conducting an assessment:

- people use inappropriate assessment tools
- they ignore the child's motivators and use materials and activities which have no meaning for the child
- they fail to adapt to the child's needs for individualized pacing and physical positioning and support
- they repeatedly attempt to assess the wrong things
- they attend only to the part of the child that they are aiming to assess (e.g. eyes, ears, hands, and so on) as if the rest of the child has no relevance
- they focus on only a single sensory channel and ignore all the other senses, as if each sense works in isolation from all the others

- they approach the child already knowing what it is that they want to see, so that the child is expected to perform to order
- they bring a 'pass or fail' mentality to the task
- they communicate with the child in meaningless and inappropriate ways
- they look for inappropriate responses and ignore the responses that the child does make
- they misinterpret the child's behaviors
- they come to the assessment with their opinions already formed in advance, and then engineer the assessment process to prove their preconceptions correct

While there is general agreement that the assessment process is especially challenging with this population, many people have no problem with committing all or most of the errors listed above and then basing crucially important decisions about school placement, support services, and educational approaches on very faulty information. We also notice that people seem to be searching constantly for the ideal assessment tool, the binder or DVD or checklist that will guide them through a straightforward and fairly quick and easy procedure that will result in a successful in-depth picture of the child. This tool does not exist of course. There are helpful assessment materials which have been designed or adapted to be used with children with sensory impairments including deaf-blindness, but the growing diversity and complexity of the population means that even these materials need to be used with care and caution, and can only ever be one part of the comprehensive assessment process. A very helpful booklet from Design to Learn Projects (Chen, Mar, Rowland, & Stillman, 2010) gives helpful advice on how to approach the issues of assessment, and also reviews twelve instruments that are commonly used to assess children with deaf-blindness in the US. Unfortunately there are many more assessment tools out there that have not been designed or adapted for this population at all, and these are often used because the assessors have no knowledge of anything more appropriate. Staff training in appropriate assessment approaches is also extremely scarce.

### **Follow the child**

The idea that, rather than starting our assessment with a published instrument or a fixed set of questions in our mind, we should 'follow the child' has become a well-known concept in the field of deaf-blindness. For almost 5 decades Jan van Dijk has been suggesting that we should follow the child (van Dijk, Oster, & McDonnell, 2009), and in 2001 I wrote an article called 'Follow the Child – Approaches to Assessing the Functional Vision and Hearing of Young Children with Congenital Deaf-Blindness' (Brown, 2001), which attempted to explain just how helpful this approach could be. A seminal article by Robbie Blaha also offers invaluable advice for working successfully with this population of 'untestable' children through careful observation (Blaha, 1996). Unfortunately, this idea still seems to generate concern and misunderstanding and suspicion, so it seems a good idea to re-visit it and try to clarify what it means and exactly why it can be such a successful approach.

In view of critical responses that I have often received after talking about this approach, it might be helpful first if we think about what 'follow the child' does not mean, just to clear up mistaken assumptions and misunderstandings. It is not a recommendation that a child be left to do whatever they like all day without any adult intervention and interaction.

Neither does it mean that potentially damaging self-injurious behaviors should be ignored and tolerated. It does not mean that adults should have no preconceived ideas of what they could be doing with the child and should just allow the child to determine the entire course of all their interactions. It also does not mean that the child should be left to self-stimulate all day, although self-stimulation behaviors should be respected as a valuable source of useful assessment data for us. The ‘follow the child’ approach is primarily concerned with finding out, as quickly as possible, who the child is and how they operate, and in the process starting to build a relationship with them with minimal aversive episodes. The relationship you build is the essential component that should enable the child, one day, to follow you.

### **Assessment questions**

I like to think of assessment as a process of getting to know the child, and if done correctly that should also result in the child beginning to get to know me too. We assess because there are things that we don’t know but we want to find out. As a result we have certain questions in our minds to which we want to find the answer, and having the right questions in mind will usually guide us to some helpful answers, even if the answers to our much bigger questions may still remain to be explored. I also like to think of assessment, and the process of getting to know a child, as asking a series of questions about the child to obtain information that I want to know



but don’t have at the moment. I may ask the parents and others who know the child better than me, and their information will form a major part of my assessment findings. Pediatric Ophthalmologist Dr. Gordon Dutton recently told me that his initial interview with a parent can provide up to 90% of the information that he needs to carry out an assessment. I may not ask the child these questions directly, but they will be in my mind as crucially important things that I wish to discover, and they will guide and color my behavior in a way that I hope will show the child that I am interested in these questions that demonstrate my respect for them. These would not be any of the obvious big questions that are often the sole focus of people’s assessment procedures, questions like “What can you see?”, “What are your hearing thresholds?”, “How many manual signs do you recognize and understand?”, “How many steps can you walk unsupported?”, “Can you stack three one-inch cubes?”, and so on.

### **The First Questions**

Before we begin to address any of these specific questions about the child’s precise, measurable skills we could usefully occupy our minds with a much more personal set of questions. In an article published in 2001, Nelson & va Dijk suggest four key steps to beginning an assessment:

- Make the child at ease
- Determine the child's bio-behavioral state
- Determine the child's interest
- Follow the child's interest

In my article, also published in 2001, I suggested a similar approach but in a rather different way:

“Basing the assessment approach on the child's curiosity and personal satisfaction, on current abilities and interests rather than on current deficits, on function rather than on structure, on motivated behavior rather than on sterile performance, is now seen as a legitimate and effective way of beginning the process. The approach needs to be individualized and holistic, so that every aspect of the child is taken into consideration even if only one sensory or skill area is being assessed. The emotional needs of the children will exert a direct and powerful influence on their ability to function, so that serious consideration of questions like 'How do you feel?' 'What do you like?' and 'What do you want?' will provide the best basis for successful assessment. People often think that 'What can you do?' is the key question to pose to any child during an assessment, but with this group a better question to begin with would be 'What do you do?'" (Brown, 2001, p. 2)

My first four questions are important in providing a good basis for successful relationship building, and they match the 4 key steps of van Dijk & Nelson well. Although I refer to them as 'first' questions I think they should be in your mind all the time in all interactions with the child, not just as your starting point. They are not age-specific but apply to all chronological ages and developmental levels. I have seen teachers use these four questions to structure their observations and their preliminary guessed interpretations, and from this get a fairly quick and useful idea about who the child is and how they function.



### **How do you feel?**

'Make the child at ease' and 'Determine the child's bio-behavioral state' are both obviously included in my very first question 'How do you feel?'. This is a population particularly likely to show good days and bad days, even good moments and bad moments, and we have known for many years that extreme variability of functioning and attention is a characteristic of most younger children with deaf-blindness. Issues of poor self-regulation, rapid changes in arousal levels, complex health issues, pain, physical discomfort, neurological involvement that results in

variability of sensory perception and processing, are all likely to be at work here. How can we deal with this in an effective and reasonably well-informed way? Knowing the child is the only thing that helps as far as I know, or relying upon guidance from those who know the child well.

### **What do you like?**

'Determine the child's interest' is clearly a main feature of the question 'What do you like?'. I have written at length about the crucial importance of motivators for learning, and the fact that it is motivation that makes brains grow and develop (Brown, 2009). Of course, once motivators have been identified they have to be used in creative ways appropriate for the individual child.

### **What do you want?**

The next question 'What do you want?' takes us directly to the child's already existing expressive behaviors and immediately credits them with the ability to indicate their feelings and desires to those who bother to observe and get to know them, even though this may not be being done intentionally by the child in the early stages of development. Eye pointing, toe wiggling, hyperventilating, rhythmic grunting, and freezing like a statue can all indicate very specific feelings and wishes in certain contexts once you really know the child.

### **What do you do?**

The fourth and last question might be the most important of all since it leads us to the essence of the individual. Sometimes a person will claim that a child does nothing when they are left entirely alone, even though this is obviously unlikely to be true. We could categorize this as very careless observation, which tells us a lot about the person's attitudes and preconceptions (and their poor observational skills) rather than anything at all useful about the child. More often I am told that if left alone the child will only stim, 'stimming' being the disapproving abbreviation for self-stimulation, which is widely considered as a thoroughly bad thing, to be opposed and stopped as a first priority. For me a child's self-stimulation behaviors provide vital insights into who they are, what their sensory needs and preferences might be, and how they self-regulate to arouse or calm themselves. These are vitally important things to know about a child if successful interactions are to take place and if learning is to result from those interactions. I have outlined thoughts about this aspect of a child's behavior in two articles which are now available on-line (Brown, 2008; Brown, 2009), and a further article with a specific focus on self-stimulation will be available soon.



### **What can you do?**

This is typically a teacher's question, to get an idea of the thresholds of the child's skills so that you have a good idea where to start teaching. Much of the information needed to answer this question will come from direct interaction with the child to try to elicit responses from them. It is an important and highly relevant question, but it needs to be posed after the first 4 described above.

### **So how do you ‘follow the child’?**

Leaving aside questions of attitude and expectations, which are the essential starting points, success in this form of assessment is based upon meticulously careful observation of the whole child, collaboration with others (including especially those who know the child better than you do), interpretation of your observations, testing of those interpretations, and amending things until you are fairly certain that you have got things right, at least for the time being.

This intensity of observation is a quiet and undemonstrative business—I sometimes observe a child intently for an hour or so and then get asked when I intend to start my assessment because people don’t realize that I am already doing exactly that! It is exhausting work if done thoroughly for several hours—it is not unusual for me to leave a school visit tired and with my brain spinning with impressions and possible interpretations—but it can be done in short periods too, with your observations compared and consolidated over time. On occasion I can observe a child carefully for just 30 minutes and then feel confident enough to intervene, introduce myself, and make a success of the interaction. This approach works, and nobody has ever shown me a better way to start to prepare for effective intervention and teaching. It is also an honorable and valid way of assessing and working with children, and Jan van Dijk, more than anyone else, has provided this validation for us in his lifetime’s work. Watch his webcast on ‘Child Guided Assessment’ (van Dijk, 2011) and see wisdom, respect, practicality, and genuine caring at work.



### **What are you observing for?**

Blahe gives useful ideas on what we should be observing for. Again, van Dijk (Nelson & van Dijk, 2009) has given us useful information about where our focus should be rather than the narrow skills-based emphasis of most conventional assessment procedures. His short list follows this sequence:

- Ability to maintain & modulate state. How well does the child achieve and remain in a receptive state of arousal which promotes perception and attention and learning, how well can they increase or decrease their state of arousal to maintain attention, and what activities and sensory inputs facilitate these processes?
- Preferred learning channels. Is the child a visual, an auditory, a tactile, or a movement learner? Does this vary, and under what circumstances? How are their sensory hierarchies impacted by both internal and external environmental factors?

- Ability to learn, remember & anticipate routines. This is a fundamental feature of caregivers behavior with infants, even on the simplest level of playing peep-bo or tickling games, and routines are used extensively in the field of deaf-blind education going up to a very complex and sophisticated level.
- Accommodation of new experiences with existing schemes. Once a routine is memorized, and recognized, how does the child cope with unexpected changes in the routine? Do they even notice the change, and what problem solving abilities do they show?
- Problem solving approaches. What abilities does the child demonstrate to perceive a problem and then find a solution to it? Do they give up easily, do they experiment with alternatives to find a way through the problem, do they ask others for help, or do they react with frustration and episodes of greatly raised arousal?
- Ability to form social attachments and interact. This includes recognition of others, memory of previously established social interaction patterns, differentiated behaviors with different people, and expansion of the social circle from a single special partner to others.
- Communication modes. Use of a multi modal language and communication approach to identify 'the child's preferred modes' over time (remembering that the preferred mode for expressive communication might be different to the preferred mode for receptive communication).

To this list I would add the importance of observing the child's postural preferences and the way they transition from one posture to another, which can offer insights into their distractibility issues, sensory needs, and self-regulation strategies.

I hope that anyone with responsibility for assessing a child they do not know will consider following the child in the ways that I have described as a first step, before they intervene to elicit responses from the child.

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*This article is the first installment in a two-part series. Part Two will be published in an upcoming edition of reSources.*

## **Deaf-Blindness, Self-Regulation, and Availability for Learning: Some Thoughts on Educating Children with CHARGE Syndrome**

by David Brown, Educational Specialist

Sigmund Freud claimed that there was no such thing as bad weather, only the wrong clothing (obviously they never get tornadoes in Vienna). In much the same way we can say that there is no such thing as a child with CHARGE syndrome who cannot be educated successfully in a local school, only programs which are not the right 'fit' for the child. Is it easy to get this right? Usually not, and it tends to become more of a challenge as the child grows older and moves into middle and high school, but there is evidence from a growing number of successful situations that demonstrates both the challenges and the attitudes and solutions that can lead to success. Reports from a large number of parents and education professionals over the past few decades confirm again and again what a challenge this can be. Often the biggest difficulties relate to behavioral features of the children, but these are usually compounded by inappropriate responses from the education world itself. The chief culprits that I see are lack of creative thinking, stereotypical and inflexible attitudes, inappropriate assessment procedures, a refusal or inability to genuinely observe children, and a misinterpretation of things that are observed. Attempts are made to force the children into a pre-existing, standard mold, often with behavior plans as a significant extra component, and the end result is almost always to intensify the problems and move even further from any desired educational outcomes.

People with CHARGE generally have to cope with much more than just functional vision and hearing problems because this condition often involves multiple systems and all, or most of, the senses. In my experience most people with CHARGE develop amazing adaptive behaviors and almost all of them end up "fooling" other people into thinking that everything is easier and more straightforward for them than it actually is. People who imply that a child's vestibular or vision or hearing issues have resolved and are no longer a problem have clearly misunderstood the compensatory skills that the child is having to use in order to function at their current level. A good and effective educational program, while being very positive and having high expectations, should always take account of the fact that everything that a child with CHARGE syndrome does is likely to take more thought and attention and concentration and energy and time for them than it does for those of us who work with them. The education establishment seems to be unduly fixated on, almost obsessed with, considerations of cognitive skills and potential to the exclusion of all else, yet for most children with CHARGE cognitive issues appear to be of lesser

significance compared to the overwhelming impact of multiple sensory and neurological issues that we are still struggling to identify and understand.

The key challenges are listed below:

### **Deaf-Blindness**

Most people with CHARGE that I have met satisfy the criteria for being considered as having deaf-blindness, even if they have some useful vision and hearing. This is a disability that is defined in functional, not clinical, terms and for each individual with CHARGE it is mostly about difficulties in accessing information not just from the world around them but even from their own bodies. In 2005 I wrote that,

“People with CHARGE syndrome are truly ‘multi-sensory impaired’, having difficulties not only with vision and hearing but also with the senses that perceive balance, touch, temperature, pain, pressure, and smell, as well as problems with breathing and swallowing, eating and drinking, digestion, and temperature control.” (Brown 2005)

People who suggest that a child’s functional vision and/or hearing is fine inspite of the presence of features like ocular defects, persistent ear infections, very low muscle tone, poor tactile and proprioceptive perception, and significant vestibular dysfunction should be regarded with scepticism. If anyone wishes a broader view of vision issues in CHARGE syndrome, for example, and the complexities involved, go to [www.cadbs.org/newsletter/](http://www.cadbs.org/newsletter/) (Brown 2010).

The philosophy of deaf-blind education offers the best match for these multiple, complex, and overlapping sensory problems, as outlined in the article “Educational Needs of Children with CHARGE Syndrome” (Majors and Stelzer 2008). The list of teaching strategies at the end of this article gives a particularly good idea of the range of approaches that might need to be considered in order for the educational program to be successful. Once a child is regarded as having deaf-blindness a very special and appropriate world of resources and information is opened up. With the Internet, and websites such as those of the CHARGE Syndrome Foundation, DB-LINK, the Texas School for the Blind and Visually Impaired, California Deaf-Blind Services, and many others, advocates for this population now have a significant and growing body of information to assist them.

### **Curriculum Delivery**

For children with CHARGE it is particularly important to think carefully about ways that the curriculum is being delivered. Although a child might ‘perform’ well with an adult in quite structured activities, these activities could sometimes impose significant levels of stress if they continue for too long a period of time, or if the child’s positioning becomes uncomfortable or insecure. Because a child seems to have a good level of cognitive ability and potential there might be an assumption that they should therefore be able to sit for 20 or 30 or more minutes at a time and attend and learn in quite structured ways, rather like most other students of their

age with a hearing loss or a visual impairment. But cognitive ability/potential is only one of the things needed to learn and function in this kind of way, and in fact the child might have a range of disabilities, and especially sensory difficulties, that are likely to present big obstacles to managing these (for them) lengthy periods of immobile desk-learning.

An activity-based approach to curriculum (i.e., working on the appropriate curriculum goals in areas like math and language and social and self-help skills, but all embedded within certain functional activities that require the child to move around periodically) can often help to keep stress levels and arousal levels down. Many children with CHARGE enjoy and respond well to moving around and physically ‘doing’ things, and can learn a lot of the curriculum while they are doing this. They also, and paradoxically, often need to get into a fully-supported horizontal position, or at least place their head in this position for periods of time. Of course, a certain level of ‘desk-learning’ should be introduced as a part of each day also. Keeping these sessions fairly brief, requiring the child to move when they seem to be losing concentration, and using materials and concepts that are motivating and meaningful to them should help to guarantee a fair degree of success. Sometimes the child might need lots of time to gather information, work at things and get them right, but at other times they might need activities that do not last too long and that move at a good brisk pace; otherwise they might become stressed or bored and may need some kind of specific sensory input. In particular, people need to learn how to ‘read’ the child to see when they are becoming over-aroused, or stressed, or tired, and so are needing to be helped to transition smoothly to a more appropriate activity.

### **Communication & Language**

There are many important and complex considerations in the area of communication and language. For a variety of reasons, many children with CHARGE have difficulties with accessing purely spoken language, especially with using it themselves for expression. But they do show great interest and skill in receiving information visually (i.e., signs, gestures, familiar objects) and in expressing themselves gesturally. While it might be important to continue using appropriate spoken language with the child, I think it is important for most children in this population to offer them language in sign and to think about using gestures, facial expressions, and objects communicatively as well, and going for a Total Communication approach (see Majors & Seltzer). If the child does later opt for speech as their expressive language mode, there may be several specific reasons why they have some difficulties with the articulation of speech (see the list below), and giving the child access to sign as a backup for their expressive speech should also help to reduce any potential frustration levels at not being immediately understood by other people. The child may well then make their own choices and develop their own preferences about how and where and when and with whom they use speech or sign. Many children, if they have a range of communication options available to them, both receptively and expressively, clearly show their skill at choosing what suits them best from moment to moment. It is not at all unusual for a person with CHARGE syndrome to develop good levels of language but to receive in one mode (often speech) and to express in a different one (often sign). There is also evidence of gradual or sudden deterioration of hearing and/or visual abilities in some people with CHARGE, which adds further weight to the argument that Total Communication is a wise option to maintain.

One additional point that needs to be mentioned is the growing likelihood that many people with CHARGE might have executive function disorder (see below). This carries many implications, but as far as communication is concerned it may make it difficult for children to initiate it, so that even if they have the language (spoken or signed), and even if they know exactly what they want or need to say, they might be unable to do it unless another person gets them started (e.g., by asking the child a question like “Are you okay?”, or “Do you want something?”, or “Do you need to go to the bathroom?”). Sometimes, a tool like a written or pictorial daily calendar system might be appropriate and helpful, along with some kind of individualized communication book that includes a vocabulary of relevant written words or drawings of signs, which the child can refer to to help get themselves started. These concrete systems might offer children an important amount of support and reassurance even though they may not obviously refer back to the calendar often throughout the day. My experience suggests that many students with CHARGE benefit from simply knowing the calendar and/or the communication book is there and can be consulted if needed.

### **Potential Barriers to Speech & Sign Articulation**

When we consider the many anomalies that are associated with CHARGE syndrome we find many that are likely to have a negative impact on the development of well-articulated speech or signing. I will list these significant anomalies first from the perspective of barriers to effective speech articulation:

- Hearing impairment
- Vision impairment
- Facial palsy
- Low muscle tone
- Poor tactile perception
- Oro-facial clefting
- Enlarged tongue
- Poor tongue movement
- Small lower jaw
- Larynx/pharynx anomalies
- Breathing difficulties and poor lung capacity
- Swallowing difficulties
- Dental anomalies
- Delayed/immature eating skills
- Chronic congestion/infections

In spite of this daunting list many children with CHARGE do opt for speech as their preferred expressive mode, although articulation difficulties may make their speech very difficult to understand. The list of anomalies that may have a negative impact on good sign articulation is just as imposing, although generally speaking it is easier for children to form intelligible signs than intelligible words. Here is the second list, from the perspective of barriers to effective sign articulation:

- Low or poorly modulated muscle tone
- Poor tactile perception
- Poor proprioceptive perception
- Poor vestibular perception
- Poor spatial awareness
- Dyspraxia
- Poor body awareness
- Poor bilateral coordination
- Poor sense of equilibrium
- Various postural difficulties
- Low vision, blindness or visual field losses
- Skeletal anomalies

### **Avoid Working at Thresholds**

I think of most children with CHARGE syndrome as a people who, for much of the school day, are probably operating at the very thresholds of their sensory abilities, which is a challenging and potentially stressful level at which to be functioning for more than short periods of time. This is possibly the situation even at times when the children seem quite relaxed and confident and the activities seem well within their various competencies. Consequently it is a good idea to try to ensure that activities are simplified and made as easy as possible for each child so that the essential teaching point is clear and accessible and the child is not having to think about and prioritize other considerations (e.g., dealing with background noise, or challenging physical positions, or visual clutter or glare, or other kinds of sensory overloading, or anxiety about unknown future plans, and so on). The children's level of alertness or fatigue appears to show itself in their posture and movement patterns and attention levels, and people involved with them need to familiarize themselves with these indicators and know how to adapt each situation so that the child is supported and does not become too tired or too stressed.

### **Complex Health Issues**

These are typically complex and severe in nature, and often and repeatedly are life-threatening in nature, so they take priority over educational considerations. Thus there are significant considerations around the availability of medical/para-medical services, and the strategies required to balance health and educational needs. In the broad special education field there is extensive literature about these kinds of issues in school settings. It is not so very rare to see a child with CHARGE in school with a full-time nurse, maybe also with a full-time educational intervener or a sign interpreter, and in my experience this has always seemed necessary and appropriate in each case I have observed. If nursing and healthcare procedures take up much of the school day that is where the education program can focus. So much of the curriculum can still be accessed during these procedures which are not an excuse for abandoning any idea of dialogue, communication and language development, numeracy and literacy work, orientation and mobility activity, turn-taking, for example. In this population early vocabulary will often include the words/signs for such things as ventilator, G-tube button, suction, pills, and so on. It

may also be possible to work towards independence with some of these nursing/para-medical procedures; it seems to me quite iconic and typical that the first child I ever saw give himself lunch through a G-tube entirely independently (while he continued to converse in sign language one-handed) should have been a teenager with CHARGE!

### **Executive Function Disorder & Self-Regulation Issues**

With the passage of time I have come to consider this as probably the most challenging and least understood long-term aspect of this condition for the children themselves, their families and educators. Whenever I am approached and asked to help with a student with CHARGE it is almost always these issues that are the main focus of the challenge and so of the solutions. Several people in the field are now investigating, among them Tim Hartshorne who wrote this in the CHARGE Syndrome Foundation Professional Packet:

“Children with CHARGE develop behavioral difficulties, some of which may be described as autistic-like, and obsessive-compulsive, with attention difficulties and tic disorders also present. There are potentially multiple sources for these difficulties, and research is attempting to sort them out. Multi-sensory impairments, communication frustrations, and physical pain and discomfort all have been implicated. However, some children with fairly good sensory abilities, adequate communication, and little apparent discomfort may still have challenging behavior. Cognitive impairment has been implicated in some but not all cases. It seems likely that some neuropsychological problems exist. Recent research supports the presence of executive dysfunction, or problems with shifting, initiating, inhibiting, or sustaining actions based on prefrontal cortex activity. Another area being explored is the presence of a regulatory disorder making it difficult for the child to regulate complex processes such as their sleep-wake cycle, hunger-satiety cycle, their ability to console themselves, to manage their emotions, and to plan their motor activities.” (Hartshorne 2005)

So many issues encountered in a significant portion of the population of people with CHARGE – poor self-regulation, rigid dependence on routines, difficulties managing transitions, maintaining an appropriate emotional perspective, urgent but unusual postural needs, sensory needs and resultant self-stimulation behaviors - all add layers to the puzzle. The first step seems to be to try to ascertain when these issues, and which of them, are present before anything effective can be done about them. My observations suggest that most children with CHARGE are not always and not fully available for learning due to this multi-layered set of issues, and that this needs to be considered urgently before other aspects of the academic curriculum can be addressed effectively. We are learning from examples of creative thinking and good practice, much of it crisis-driven. The use of interveners (see <http://intervener.org/> for example), an idea that comes from the field of deaf-blind education, has proved very effective in many cases – in aiding more efficient ongoing assessment, providing more effective access to the curriculum in fulfillment of IEP requirements, in facilitating interactions with peers and an ever-growing social circle, and in offering reassurance and effective strategies when self-regulation issues begin to arise. We still have a long way to go, but at least the focus now seems to be more clear and more appropriate.

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# Behavior in CHARGE syndrome

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Unusual behavior is often associated with genetic syndromes, and may constitute a behavioral phenotype. In contrast to providing a psychiatric diagnosis, a behavioral phenotype describes what is unique to the behavior associated with different syndromes. While behaviors in CHARGE are as complex and variable as other aspects of the syndrome, there are some commonalities that raise the question of common sources for these behaviors. This article addresses how pain, sensory issues, and anxiety may impact the behavior of individuals with CHARGE syndrome, and how the development of self-regulation skills might help to mitigate some of the behaviors.

## KEYWORDS

anxiety, behavior, CHARGE syndrome, pain, self-regulation, sensory impairment

## 1 | INTRODUCTION

In 1996, there was a panel on behavior in CHARGE at the Australasian CHARGE conference. The next year at the International CHARGE Conference in the United States, Denno and Bernstein (1997) presented on behavioral characteristics in CHARGE. Concern about and interest in the behavioral aspects of CHARGE have increased since. In 2007, Wachtel, Hartshorne, and Dailor (2007) reported that 37 (43%) of individuals with CHARGE from their sample were on at least one psychotropic medication: 15 were on one, 13 were on two, 8 were on three, and one person was on five psychotropic medications. We suspect the number overall has increased. Behavioral issues are common topics on the various CHARGE syndrome social media pages

and behavioral presentations continue to draw large crowds at international conferences.

Problem behaviors in CHARGE are as complex and variable as other aspects of the syndrome. Physical behaviors may include scratching, hair pulling, biting, pinching, kicking, shoving, throwing objects, smearing feces, undressing, self-injury, and resistance. Verbal behaviors may include repetitive statements or questions, yelling, and complaining. Non-verbal behaviors may include agitation, pacing, invading personal space, and withdrawal. Psychiatric diagnoses are often assigned to individuals with CHARGE, the most common being autism, obsessive-compulsive disorder, and attention-deficit/hyperactivity disorder. But we have also seen individuals diagnosed with Tourette disorder, bipolar disorder, schizophrenia, borderline personality disorder, oppositional defiant disorder, anxiety disorder, and major depressive disorder.

Hartshorne and Cypher (2004) argued that “it may not make sense to add diagnoses on to that of CHARGE, such as CHARGE and autistic disorder, or CHARGE and ADHD, but rather to recognize that a diagnosis of CHARGE may indicate the potential development of behaviors that are like those in other disorders” (p. 50–51). As Waite et al. (2014) point out, “High rates of ASD have been reported in syndromes such as Cornelia de Lange and fragileX. However, there is debate about whether the ASD profile of behaviors that triggers a diagnosis in these syndromes is the same as in individuals with idiopathic ASD” (p. 470). Unusual behavior is often associated with

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genetic syndromes, and may constitute a behavioral phenotype. A true behavioral phenotype would potentially allow for the diagnosis of a syndrome on the basis of behavior that is unique to the syndrome (Harris, 2006). Hartshorne (2011) has described a behavioral phenotype for CHARGE syndrome (Table 1). While these seven characteristics appear to be descriptive of behavior in CHARGE, additional research is needed to compare behavior in CHARGE with behavior in other syndromes (Waite et al., 2014). Nevertheless, they provide an alternative to a psychiatric diagnosis and a foundation for future research.

Challenging behavior in genetic syndromes may be seen as deriving from four sources (Einfeld, 2004). These are vulnerabilities stemming from intellectual disability when present, from the child's life experience, from factors in the immediate environment, and from the specific genetic cause. While we cannot change the genetics, another approach to behavior is to try and understand why it might develop, assuming it has an experiential/environmental etiology. Through our work with individuals with CHARGE and their families we have come to see three sources of behavioral issues in CHARGE beyond what might be genetic. These are the experience of pain, sensory issues, and anxiety. Figure 1 shows these in the form of a triangle. In the middle is self-regulation, as we believe learning to self-regulate pain, sensory systems, and anxiety can help to mitigate the behavioral issues.

This article addresses how pain, sensory issues, and anxiety impact the behavior of individuals with CHARGE syndrome, and also briefly addresses the development of self-regulation.

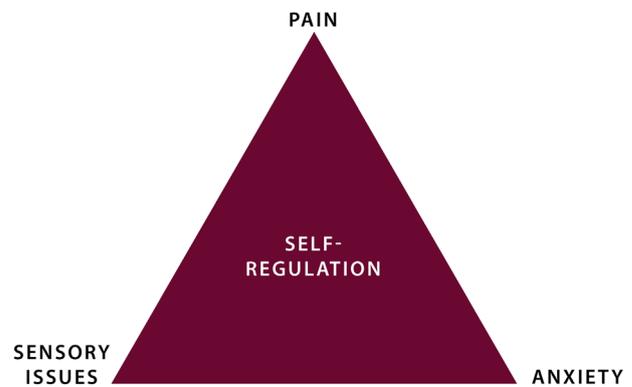
## 2 | PAIN

Historically, individuals with developmental disabilities who present with limited communication have been portrayed as having a higher threshold for pain than the general population and some have gone as far as to say these individuals do not feel pain (Sobsey, 2006). Similar statements, without any evidenced based research, have also been proposed for individuals with CHARGE, such as suggesting a high pain threshold. More recent research, however, suggests that individuals with developmental disabilities, including those with CHARGE, experience more frequent and intense pain experiences (Breau, Camfield, McGrath, & Finley, 2003; Stratton & Hartshorne,

**TABLE 1** CHARGE syndrome behavioral phenotype

Low normal cognitive functioning
Very goal directed and persistent with sense of humor
Socially interested but immature
Repetitive behaviors, increase under stress
High degree of sensation seeking
Under conditions of stress and sensory overload find it difficult to self-regulate and easily lose behavioral control
Difficulty with shifting attention and moving on to new things; easily lost in own thoughts

From ref. Hartshorne (2011). Used by permission.



**FIGURE 1** Behavior Triangle: Major sources of problem behavior in CHARGE syndrome

2010; Stallard, Williams, Lenton, & Velleman, 2001) and that they may communicate pain in a different way due to communication and sensory deficits (Oberlander & Symons, 2006; Stratton & Hartshorne, 2010).

Individuals with CHARGE endure a significant number of painful experiences including multiple, intensive medical procedures, recovery, acute complications, and various therapies (e.g., physical therapy) to name a few. Stratton and Hartshorne (2010) found that individuals with CHARGE experience considerable amounts of reported intense pain, largely from complications related to CHARGE including migraines, constipation, surgery-related pain, chronic otitis media, sinusitis, gastroesophageal reflux, dental issues, and breathing-related pain. It has also been hypothesized that all 12 cranial nerves can be impacted in CHARGE and the extent of these anomalies' relationship to pain is unknown (Blake, Hartshorne, Laward, Dailor, & Thelin, 2008). Migraine headaches, however, have been implicated in cranial nerve V functioning (Hargreaves, 2007). Blake et al. (2008) report cranial nerve V anomalies in over half of children with CHARGE. Migraines were reported by parents to be the most intense painful experience for children with CHARGE in the Stratton and Hartshorne (2010) investigation.

In addition to substantial and intense acute pain experiences, individuals with CHARGE have chronic pain (Stratton & Hartshorne, 2010). Parents were asked to rate their child's frequency of pain for a variety of identified painful experiences. Of those experiences, gastroesophageal reflux, difficulty swallowing, breathing difficulty, hip/back pain, abdominal migraines, and muscle pain were reported to occur 95 or more days a year, often in combination with one or more other painful experiences. In addition to prolonged conditions eliciting chronic pain, surgery pain is frequent. The reported average number of surgeries was 12 (range of 1–47) between the ages of 1 and 18.

Due to its chronic nature, individuals with CHARGE may develop a high degree of pain tolerance, and so underreport it. Those with communication challenges may not have the tools to indicate the experience of pain. When not reported clearly, underreported, or when communication attempts are missed by others, treatment to reduce pain is not sufficient or even missing. As the gold standard for

treatment of pain remains patient self-report, individuals with CHARGE are placed at greater risk for experiencing long-term, unnecessary pain that can impact their quality of life (Oberlander & Symons, 2006), overall adaptive skills (Breau et al., 2007), mental health (Kassam-Adams, 2006), and behavior.

With pain, problem behaviors increase. For those individuals with CHARGE who have a combination of limited communication and heightened pain experiences, it is expected that problem behaviors will be elevated and at times will be intense. Even with more formal and functional communication in place, pain can impact adaptive functioning including understanding and using language, and socializing (Breau et al., 2007), thereby reducing the self-report of pain. In one investigation, individuals with an intellectual disability were found to have between 21% and 29% reduction in functioning when in pain (Breau et al., 2007). It is unknown to what degree reductions in adaptive behavior and communication occur in individuals with CHARGE when their pain experience intensifies or occurs for a prolonged period. However, it is clear, based on our experience, that these are replaced by problem behavior, an alternative form of communicating one's needs.

As noted, many children with CHARGE develop extremely challenging behavior (Hartshorne, Hefner, & Davenport, 2005; Lauger, Cornelius, & Keedy, 2005; van Dijk & de Kort, 2005). It has been shown that challenging behaviors increase with pain, including aggression and self-injury, and may serve as indicators of pain or alternative ways to communicate pain to others (Cook, Niven, & Downs, 1999; Symons & Danov, 2005). Further, longer periods of time associated with elevated pain have also been found to be associated with elevated ratings of self-injurious behaviors (Symons & Danov, 2005; Symons, Harper, McGarth, Breau, & Bodfish, 2009). Given this, it is imperative for parents, medical professionals, educators and the like to first rule-out pain when problem behaviors have presented, spiked, and/or appear unexplainable. All behavior is communication and challenging behaviors like aggression, property destruction, tantrums, and self-injury, are strongly communicating that there is a problem, and it is often due to pain.

The CHARGE Non-Vocal Pain Assessment (CNVPA) (Stratton & Hartshorne, 2012) is a non-vocal pain measure that can be used to screen for pain based on an individual's presenting behavior. This 30-item measure assesses five subscales including vocal, social, facial, activity/challenging behaviors, and body limbs/physiological and uses a Likert rating scale. The CNVPA is reported to have strong psychometric properties, good social validity, and has been found to discriminate pain well from non-pain behavior for individuals with CHARGE.

In summary, problem behaviors serve a purpose and often in CHARGE are communicating discomfort or pain. Due to multisensory impairments, many individuals with CHARGE present with limited communication that can be further limited by a painful experience. Rather than using the historical model of "They don't feel pain or they need to experience more pain to feel it," more recent research suggests individuals with CHARGE tolerate a significant amount of pain and likely communicate this through problem behavior. Non-vocal pain

assessments can be useful to identify pain early when problem behaviors begin/increase and can lead to earlier treatment of pain/discomfort and a reduction, or elimination in problem behavior. Teaching strategies to indicate and communicate pain at the child's developmental level is strongly suggested, particularly when pain is present. As more formal communication increases, problem behaviors are less likely to develop.

### 3 | SENSORY ISSUES

As they develop, children learn how to create responses to their sensory experiences (Dunn, 1997). According to Dunn, sensory processing difficulties can affect social, cognitive, and sensorimotor development. They can also impact the child's reactivity to their environment, and this can lead to behavioral issues. Fox, Snow, & Holland (2014) found sensory processing difficulties in well over half of 38 five- to nine-year-old children at risk for conduct disorders.

Sensory impairments and differences in response to sensory stimulation are frequently associated with genetic syndromes, and understanding these and how they influence behavior is important (Waite et al., 2014). Many of the puzzling behaviors demonstrated by children with CHARGE syndrome originate from the complex multisensory impairments that are prevalent in the condition (Davenport & Hefner, 2011). The behaviors begin because they serve important functions for the children, and it is necessary to understand the functions of these behaviors before attempting to modify them (Murdoch, 1997). The brain is only connected to the body and to the outside world through the sensory systems (Coren, Porac, & Ward, 1984) and in CHARGE syndrome all of the sensory systems may be malfunctioning for various reasons. There may be specific problems with the sensory receptors (the eyes, the ears, the vestibular apparatus, etc.), there may be problems with the nerve pathways that connect the sensory receptors to the brain, specifically, frequent impaired function of the cranial nerves (Blake et al., 2008), and there may be malformations of the brain itself (Feng et al., 2017), and all of these will have a significant negative impact on overall sensory functioning. It is not unusual for a child to have all three kinds of these problems, (with sensory receptors, with nerve pathways, and with the brain), which makes functioning (for them), and understanding and interpretation (for us) very difficult.

We rely on intact sensory systems to gather information about our environment. It is important to remember that damage to any one sensory system can present functional challenges to integration with other, apparently unrelated, sensory systems, because information is missing. In this context even minor issues with a sensory system, things which should be easily managed and compensated for by most people, may have a magnified impact on overall sensory functioning and behavior for a child with CHARGE syndrome. For example, vestibular problems may contribute to functional vision deficiencies (Möller, 2011). Clinical assessment might suggest that a child's vision or hearing or balance are fine, particularly if examination of eyes and ears

suggests that everything is anatomically intact. However, close observation of the child is likely to reveal significant problems with function, not least because of the interdependence of the senses (Smith, Smith, & Blake, 2010). As a result it is most helpful to consider children with CHARGE syndrome as multi-sensory impaired, and to utilize well-established educational practices from the broad field of deafblindness.

The sensory systems of children with CHARGE syndrome are unusually vulnerable to deterioration or further damage. A sensory problem may be temporary because of an infection (e.g., of the eye or the ear, joint pain, or a skin infection). But it may also be the case that the sensory system itself has sustained permanent damage as a result of such things as persistent and severe infections over time, unfortunate side effects of surgical intervention, falls resulting in impact to the head, or the child's own self-stimulation behaviors which can develop in intensity and frequency until they become self-abusive (e.g., head banging, teeth grinding, eye poking, skin picking). Any worrying change in behavior (e.g., not looking at things, not responding to sounds, moving less, resisting being touched, or lifted and carried, sleeping too much or not enough, self-stimulating with more intensity or more frequency) should result in an investigation of sensory status.

Significant variability of sensory functioning may also result from internal factors (such as fatigue, hunger, chronic constipation, pain, poor body temperature control, anxiety) and external factors (such as light levels, noise levels, sensory distractions, postural insecurity). Sometimes aggressive behavior towards self or others may be reduced, or eliminated altogether, by measures taken to change these internal and external factors. Pain control, deep pressure massage, removal of excessive environmental stimuli, regular movement, and better physical support in sitting, can all help a child to become less distressed, and more available for social interaction and learning.

An additional consideration is that many of the children have sensory systems that are not integrated together effectively so that they do not respond to stimulation in a well-modulated way (Brown, 2005). Such sensory processing issues are considered to underlie behavioral and functional problems in Autism Spectrum Disorder (Case-Smith, Weaver, & Fristad, 2015). This is likely the same in CHARGE given the challenges of multi-sensory impairment. As a result, apparently minor incidental sensory inputs (air movement or quiet noise from a heater or air conditioner, shadows created by people walking between the child and a window, a wet or sticky patch on the tabletop where the child places their hands) may overload the child and create strong aversive responses, high levels of over arousal, or a significant increase in possibly damaging self-stimulation behavior. Good deafblind educational practice is founded upon a multi-sensory view of the child and their environment, so that all of these internal and external factors should be automatically considered, particularly when a child's behavior is causing concern.

Just like all infants, young children with CHARGE syndrome self-stimulate in order to learn about their bodies, to extend and practice skills, to self-regulate, to amuse themselves, and to make themselves feel more confident and comfortable (Murdoch, 2000). With

multi-sensory impairment the need to self-stimulate becomes more intense and more persistent because the brain is not in good contact with the body. Characteristic self-stimulation behaviors in early childhood include rocking side to side (just the head or the whole body), kicking the legs, sucking the fingers, grinding the teeth, rubbing or scratching the body with the hands, staring at light sources (often while waving a hand in front of the eyes to create shadows and visible movement), and adopting postures which create strong proprioceptive stimulation (crossing the fingers and the legs, arching the back, curling into a fetal position, pressing the head, squeezing into tight spaces). These behaviors can serve many important functions (Moss, 1993), including confirming that the body has postural security, reducing the impact of joint pain, helping to maintain an open airway, reducing discomfort from constipation, stabilizing the visual field, calming down, and gaining and maintaining alertness. In the early days all these behaviors give the child's brain a better idea of where the body is, what it is doing, and what it is capable of doing. As a result these kinds of spontaneous behaviors are inherently self-regulating, and they offer us important insights into the child's challenges and needs, and valuable pointers for our interventions (Haney, Hartshorne, & Nicholas, 2015). But if we do not notice and value self-stimulation behaviors, or even worse, if we consider them nonfunctional or negative and try to eliminate them as our only goal, then this is likely to have a negative impact on the child's self-confidence and sociability, and in turn their behavior. If we believe that children with CHARGE engage in unusual behavior because it is in some way functional for their self-regulation (Ramirez, Hartshorne, & Nicholas, 2014), then attempts to block these behaviors may frustrate and challenge the child to engage in even more serious behaviors. Our goal must be to understand why the child might have chosen these behaviors, and then help them to find alternatives that still provide, among other things, the necessary sensory stimulation.

## 4 | ANXIETY

Anxiety, anxious behavior, and diagnosed anxiety disorders are common concerns among individuals with CHARGE and their caregivers. A survey of 87 families found that anxiety disorders were the most common mental health diagnoses reported in individuals with CHARGE (Wachtel, Hartshorne, & Dailor, 2007). Blake, Salem-Hartshorne, Daoud, and Gradstein (2005) surveyed caregivers of individuals with CHARGE and 43% indicated that their child was diagnosed with obsessive-compulsive disorder while 37% percent indicated an anxiety disorder diagnosis. Similarly, Hartshorne et al. (2016) found that about 50% of their participants with CHARGE indicated difficulties with anxiety and/or obsessive-compulsive behaviors. Overall, it is clear that anxiety is a prevalent concern in this population.

Anxiety has many different causes. Individuals with CHARGE syndrome are exposed to circumstances that contribute to a higher level of anxiety (Wachtel, 2011) including concern over explained or

unexplained pain, sensory overload or underload, and lack of predictability in the environment. A meta-analysis of children with chronic illnesses found a small but meaningful difference in anxiety compared to levels of anxiety in the general population (Pinquart & Shen, 2011). Specifically, they found that there were higher levels of anxiety in individuals with sensory impairments. The authors noted that lack of control over an illness can lead to elevated levels of anxiety. Anticipation of pain can also lead to higher levels of anxiety in individuals with CHARGE and these higher levels of anxiety can lead to deficits in tolerance of chronic pain (Nicholas, 2011).

Sensory impairment and difficulties gathering and processing information can lead to a lack of understanding of one's environment and the progression of events. This lack of predictability can result in higher levels of anxiety and increases in behavior aimed at communicating distress or obtaining security. Use of interventions that help mark changes in routine and anticipated events can make the environment more predictable (Blaha, 2001), which may help to reduce anxiety.

Anxiety can have a direct positive or negative impact on behavior; moderate amounts can prove useful while too much can be debilitating and too little can be dangerous. Children with CHARGE syndrome exhibit behaviors that are common with anxiety: negative thinking patterns, anger, aggression, tantrums, crying, physical complaints, avoidance behaviors, sleeping difficulties, eating disturbances, and withdrawal from activities or family interactions (Eugster, 2007). Research indicates that while anxiety is a concern for individuals with varying genetic syndromes, there are differences in how that anxiety is exhibited (Crawford, Waite, & Oliver, 2017; Leyfer, Woodruff-Borden, & Mervis, 2009). CHARGE syndrome has a behavioral phenotype (Table 1) (Hartshorne, 2011) that may represent a unique profile of anxious behaviors.

Individuals with CHARGE syndrome often exhibit obsessive-compulsive behaviors and many are diagnosed with Obsessive Compulsive Disorder (OCD) (Blake, Salem-Hartshorne, Daoud, & Gradstein, 2005; Hartshorne, 2011; Hartshorne et al., 2016). These behaviors include repetitive behaviors, sorting objects, linking up objects, checking, and an all-consuming focus on one idea, activity, or item (Wachtel, 2011). Hartshorne and Cypher (2004) asked one hundred parents of children with CHARGE about the behaviors that their child exhibits and about a quarter to a third of participants indicated that their child engaged in behaviors linked to a typical OCD diagnosis, including the need for items to be arranged in a certain order or symmetrically, seeking reassurance on doing or not having done an activity, the need to touch certain parts of the body or to blink the eye, and doing the same things over and over again. There can be an adaptive function to such behaviors. For example, ritual organizing and ordering of items may reduce the anxiety over where things are for someone who is multi-sensory impaired. Repetitive behaviors can provide a sense of regularity to the environment and the individual's experience.

Individuals with CHARGE syndrome sometimes exhibit over-responsivity to sensory input (Hartshorne, 2011). Sensory over-responsivity (SOR) is defined by behavioral responses to sensory stimuli that are atypical or exaggerated. While the cause of SOR in

individuals with CHARGE and how it might be related to sensory impairment are not known, evidence shows that SOR is associated with anxious behavior, and anxiety diagnoses. In a study of college students, individuals with high levels of sensory responsiveness were more likely to be anxious (Levit-Binnun, Szepeswol, Stern-Ellran, & Engel-Yeger, 2014). Conelea, Carter, and Freeman (2014) found 88 children with a clinical diagnosis of anxiety were likely to exhibit behaviors related to SOR. They also found that SOR was significantly correlated to an OCD diagnoses. Similarly, Lewin, Wu, Murphy, and Storch (2014) found that sensory concerns and SOR were linked to OCD-like behaviors. Therefore the heightened levels of OCD behaviors present in the CHARGE population could partially result from their sensory over-responsivity.

Individuals with CHARGE syndrome exhibit behaviors that are common in children with anxiety. Anxiety may be the result of pain, sensory impairment, and the unpredictability of the environment. Interventions should assist with pain management, sensory issues, and making the environment more predictable.

## 5 | SELF-REGULATION

Self-regulation is the ability to monitor and change one's responses to achieve a desired goal. This process involves overriding the body's automatic responses to achieve an outcome or state. There are two components to self-regulation: 1) having a desired goal; and 2) self-awareness. Together these two components allow individuals to monitor their progression toward a goal and to alter their cognition, emotions, behavior, or physiology to maintain movement toward the goal (Ramirez et al., 2014).

There are four major systems involved in self-regulation: cognition, behavior, emotion, and physiology. The body must coordinate these systems to achieve a goal after receiving input from internal and external sources. Cognitive self-regulation is the ability to regulate one's thoughts and mental processes through attentional regulation, shifting, inhibition, and working memory to achieve a desired goal. Self-regulation of behavior requires an individual to monitor one's behavior to achieve a goal, which may be done by inhibiting activity, regulating movement, or delaying gratification. Emotional self-regulation is the ability to react to situations with an emotional response that is appropriate in timeliness and intensity. Physical self-regulation involves the body's ability to maintain homeostasis after receiving internal and external stimuli. This includes maintaining equilibrium within the somatic, endocrine, and autonomic nervous systems (Ramirez et al., 2014).

Self-regulation develops through the interplay of physiological development, caregiver responsiveness, and the child's interaction with the environment (Ramirez et al., 2014). As all three may be diminished for children with CHARGE, self-regulation strategies must be deliberately taught. Interventions that teach self-regulation skills may help individuals with CHARGE better regulate pain, sensory systems, and anxiety in order to alleviate negative behaviors that may arise from these issues.



**FIGURE 2** Upside down: A child with CHARGE hangs upside down in her chair

When individuals experience pain, they often experience maladaptive thought patterns, negative emotions (i.e., depression, irritability), and compulsive and repetitive behaviors (Nicholas, 2011). Improving self-regulation skills may help individuals with CHARGE to become better aware of negative thoughts and to shift attention away from ruminating on feelings of pain. Self-regulation skills may support an individual's ability to identify and express emotions about pain in ways that do not include behavioral outbursts. For example, self-regulation skills can help an individual identify where the pain is occurring and communicate the intensity of the pain with a preferred mode of communication. Additionally, improved self-regulation skills may allow individuals to inhibit repetitive, or compulsive behaviors that may arise when in pain. Interventions like biofeedback, cognitive behavioral strategies, and relaxation skills can teach self-regulation to address pain (Sauer, Burris, & Carlson, 2010).

Individuals with CHARGE syndrome experience sensory issues including difficulties perceiving balance, experiencing touch, and sensing pressure (Brown, 2005). These can result in poor memory, difficulty coordinating hearing and vision, maladaptive thinking, and stereotypic behaviors. A behavior such as hanging upside down, commonly seen in children with CHARGE (Figure 2), may be a form of self-regulation, as it can support upper visual field loss, vestibular challenges, and provide proprioceptive feedback. Self-regulation can help the child to manage shifts from a calm sensory state to hyperarousal (Dunn, 1997). Acquiring additional self-regulation skills can help an individual to better process stimuli, inhibit triggering sensations, and manage body movements. Interventions like physical therapy and

occupational therapy can help improve self-regulation when experiencing sensory issues (see Brown (2005), for other interventions).

Anxiety can result in maladaptive thought patterns and repetitive and compulsive behaviors (Campbell-Sills & Barlow, 2007). Improved self-regulation skills may help an individual inhibit negative thoughts about past or present anxiety provoking situations, and reframe these into more adaptive thought patterns. Additionally, self-regulation may allow an individual to inhibit compulsive or repetitive behaviors that occur because of anxiety, or help individuals to communicate to others the purpose of their adaptive repetitive or compulsive behaviors. Regulating by developing a predictable routine and being prepared for changes may also lower anxiety. Calendar systems are one example of how assistive technology could be used to make the environment more predictable and encourage social-emotional regulation (Blaha, 2001). Interventions like deep breathing, mindfulness, development and availability of a "safe place" and a predictable environment may help individuals with CHARGE regulate their physiology, cognitions, emotions, and behaviors when experiencing anxiety (Kennert, Ramirez, Hartshorne, Deuce, & Nichols, 2015).

## 6 | CONCLUSION

Challenging behavior is not inevitable in individuals with CHARGE syndrome, but it is highly predictable given problems with pain, sensory issues, and anxiety. Pain should always be checked first, particularly if there has been a change in behavior. Helping children to compensate for multi-sensory impairment is essential. Anxiety can be reduced by increasing routine and predictability. Children need to be supported in the development of self-regulation skills.

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