

**CHARGE
Professional Day
+
18th CHARGE-
Conference**

20th – 23rd June 2024

Oberwesel / Germany

David Brown

20th June 2024

+

22nd June 2024

***„6 things that most people don't know
about CHARGE syndrome”***

6 things that most people don't know about CHARGE syndrome

David Brown
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18th CHARGE Syndrome Conference
Oberwesel, Germany
June 2024

“There are so many bits, this is why I love CHARGE, I mean so many challenges to me as a practitioner that you don't know where to start! I have been so fascinated with CHARGE syndrome because it's a conundrum. You've got to really work the puzzle, work the system, and be forever thinking outside the box.”

Dr Kim Blake 2019 Dallas CHARGE conference

As our understanding of CHARGE syndrome has developed over time the great complexity of the condition has become more apparent with the identification of an ever-growing number of anomalies that need to be taken into consideration. Parents often report describing the child's issues to various professionals and watching the professionals' faces gradually register disbelief that so many things could be affected in one individual. Because of this large number of possible anomalies parenting a child with CHARGE syndrome invariably requires never-ending priority making - deciding priorities, evaluating priorities, re-visiting priorities, adjusting priorities, and worrying all the time, once the life-threatening aspects of the condition have been addressed, that the current priorities might not be the most beneficial.

David Brown

“I know of no 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 People familiar with CHARGE Association often speak about iron willpower, great spirit, and a marked sense of humour being distinctive features of most people with CHARGE Association, though as far as I know this is the first time these particular aspects of the condition have appeared in the literature.”

David Brown “*CHARGE Association*” Talking Sense, Summer 1997

1. Deafblindness

5

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

David Brown
American Journal of Medical Genetics 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.”

CHARGE syndrome involves many more senses than just vision & hearing, and it is not enough only to consider the tactile sense as a compensatory channel.

CHARGE - the most ‘multi sensory impaired’ of all syndromes
Problems with the perception of:

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

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

2017 US National Deafblind Child Count

HEREDITARY SYNDROMES AND DISORDERS	TOTAL
CHARGE Syndrome	933
Down syndrome (Trisomy 21 syndrome)	315
Usher I syndrome	233
Stickler syndrome	137
Dandy Walker syndrome	115
Goldenhar syndrome	102
Cornelia de Lange	99

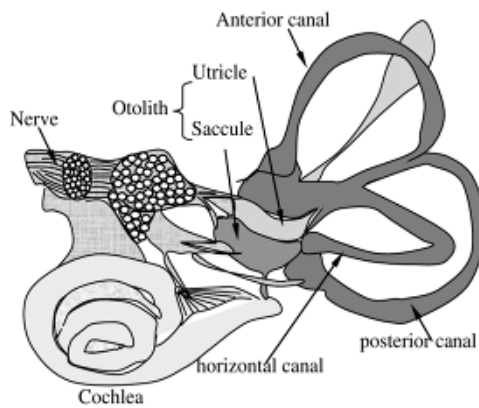
2. Vestibular dysfunction

“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



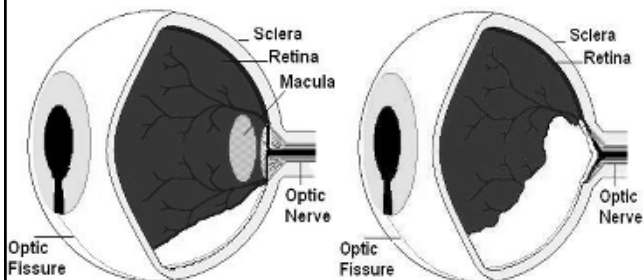
Effects of vestibular problems

- Disorganization of ALL sensory information
- Postural insecurity and low muscle tone
- Poor use of residual vision
- Problems with perception & processing of sound
- Difficulty remembering auditory sequencing
- Poor speech/language development
- Poor memory development
- Behavioral challenges
- Bilateral coordination
- Breathing, feeding, digestion, nutrition
- Sociability
- Fatigue
- Self-regulation

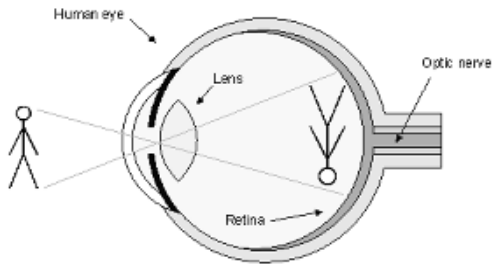
3. Some Vision Issues

- a. Visual field loss
- b. Visual fixation

Retinal Coloboma



The lens inverts the image onto the retina

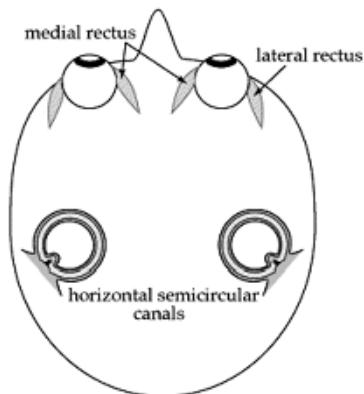


Images are inverted on their way to the retina at the back of the eye

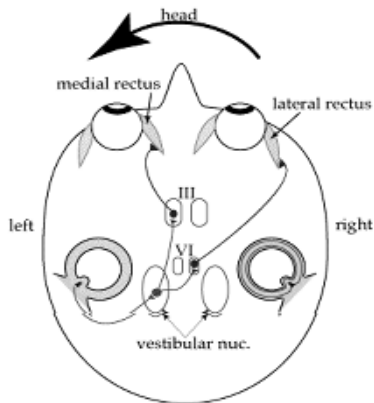
The Vestibulo-ocular Reflex (VOR)

In normal head movement the eyes move in the opposite direction to the head, and at the same speed, to stabilize the retinal image for visual fixation.

The Vestibulo-ocular Reflex (1)

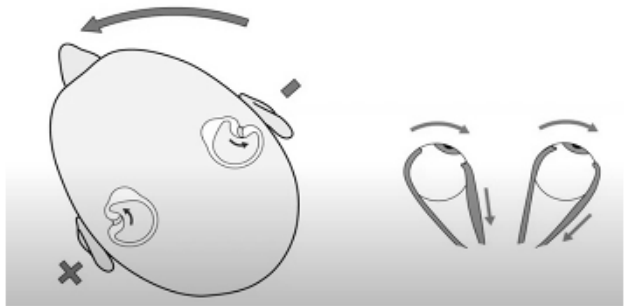


The Vestibulo-ocular Reflex (2)



The Vestibulo-ocular Reflex (VOR)

In normal head rotation the eyes move in the opposite direction to the head to stabilize the visual image on the retina.



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.

4. Balance/Walking

“Human walking is a unique activity during which the body, step by step, teeters on the edge of catastrophe... children begin to walk to trace desires no one will fulfill for them”

Rebecca Solnit, *Wanderlust: A History of Walking*, 2000

“For children with CHARGE syndrome gravity sucks”

Maryann Girardi, Physical Therapist at Perkins School for the Blind

Why is walking a big problem?

- Bone malformations
- Loose joints & low muscle tone
- Visual impairment
- Other sensory issues (Proprioceptive & Vestibular!!!)
- Breathing problems
- Cerebral palsy?
- Stress, depression, FEAR
- Medication
- Distractibility (one thing at a time)
- Low expectations & over-protection
- Too many faster, safer ways of moving!

How do we achieve balance?

Three separate components make up the “Equilibrium Triad”:

- Input from the eyes (vision)
- Input from the muscles and joints (proprioception)
- Input from the vestibular organs (vestibular)

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

Jean Ayres

Where is my head?

Head weaving

Head binding (hat, sweatband, scarf, string, sock, bucket, box)

Head holding/ head tapping

Head pressing (or head standing!)

Jaw clenching

Teeth grinding

Biting/ chewing/ sucking





Early mobility

- Rolling
- Side-winding
- Back scooting
- Bottom shuffling
- 5-point crawling
- Regular crawling

Walking

- Walkers
- Hiking 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)

Active Learning

- Little Room



- Resonance Board



These postural behaviours have always been present but largely unseen. When they are noticed there is often amusement because the postures seem weird, or the child is corrected with no attempt to understand or recognise or honour what the posture means and what function it serves.

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.

One of the most important early assessment questions we can ask the child is: "What do you do?".

"What can you do?" comes later.

Functions of these postures

- For the brain to locate the body (especially the head)
- To confirm postural security
- To stabilize the body, to stabilize the head, to stabilize the eyes for visual tasks
- To ease the discomfort of chronic constipation
- To open up the airway
- To increase desired sensory inputs
- To self regulate

5. Dental Issues

Dental Issues

- Oro-Facial clefting
- Facial palsy
- Misaligned teeth
- Delayed (or early) eruption
- Poor quality dental enamel
- Reflux and vomiting
- Medications
- Pouching of food
- Teeth grinding

6. Fatigue (often identified as laziness)

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

“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

*Communication with one's own body

*Communication with one's immediate environment

*Communication with the wider world

How do children with CHARGE syndrome fool people?
