



German Professional Day Oberwesel - 2014

Program

Thursday, July 3rd, 2014

up to 1 p.m.

From 2.00 p.m.

Arrival of participants

Claudia Junghans, President

Dr. Julia Benstz, Vice president

Official welcome and introduction of the lecturers

Organizational information

2.30 p.m.

Lecture by Dr. David Brown

“CHARGE Syndrome after 31 years”

4.30 p.m.

Coffee break

5 p.m.

Lecture by Dr. Kim Blake

“All you need to know about the cranial nerves and more”

From 6 p.m.

Dinner and get together

From 7 p.m.

Dr. Andrea Wanka : Interview with Claudia Junghans and Dr. Julia Benstz

„My child with CHARGE-Syndrome“

An insight into everyday life from the view of a mother

8 p.m.

Open Space conversation circles with the CHARGE Experts/ lecturers – topics:

- *„Communication and behavior“*
- *„Puberty and becoming an adult“*
- *„Medical issues“*
- *„Aggression and how to cope with it“*

Friday, July 4th, 2014:

From 7.30 a.m.

9.00 a.m.

Breakfast

Lecture by Prof. Dr. Tim Hartshorne

„Self regulation in CHARGE“

10.45 a.m.

Lecture by Gail Deuce

„What educational provision is likely to be needed“

12.00 o'clock

End of the Professional Day and departure

From 2.30 p.m.

Beginning of the German CHARGE Conference for affected families
(If you are interested please also join the following lectures)

14.00 Uhr: Dr. Kim Blake

“CHARGE 101: Anaesthesia, Botox, Sleep Apnea und FAQ´s”

16.00 Uhr: Prof. Dr. Conny van Ravenswaj

“All you want to know about CHARGE and genetics”

Sponsored by

BARMER GEK

CHARGE Syndrom e.V. - Elternkreis betroffener Kinder

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CHARGE
CHARGE Syndrom e.V.

CHARGE syndrome after 31 years

David Brown

CHARGE Konferenz in Rahmen des 8. Bundestreffens
Oberwesel
Thursday, July 3rd, 2014

What is special about CHARGE?

- Strong common characteristics BUT a very wide range of ability and function
- Large number of anomalies (up to 38?)
- True multi-sensory impairment
- Frequently high developmental potential plus features that normally suggest low developmental potential
- Brilliant adaptive abilities but also bewilderingly patchy development
- Sensory integration dysfunction that affects all areas of development, learning, and behavior
- Significant difficulties with self-regulation and with executive function
- Inherent high levels of stress

Resources & literature

- CHARGE Syndrome Foundation (Professional Packet, AJMG, ASHA video, Parent Manual, CHARGE Accounts, webinars, links)
- Perkins School for the Blind CHARGE webcasts
- CHARGE Lab at Central Michigan University
- DB-LINK
- Texas School for the Blind & Visually Impaired
- California Deaf-Blind Services
- Sense UK CHARGE Information Packet
- Books from the USA, from the CHARGE Association of Australasia, and from Germany
- Face Book

1981 - 6 anomalies

- C - Coloboma
- H - Heart Defects
- A - Atresia of the Choanae
- R - Retarded Growth/Development
- G - Genito-urinary Defects
- E - Ear anomalies

2005 - 38 anomalies???

- Mutation of gene CHD 7
- Facial Palsy
- Cranial Nerve anomalies
- Semi-circular canal anomalies
- Dental anomalies
- Sense of smell anomalies
- Larynx & Pharynx anomalies

Cranial Nerve Anomalies

- Nerve 1 - Smell (42%)
- Nerve 2 - Vision (80%)
- Nerve 7 - The Face (43%)
- Nerve 8 - Hearing & Balance (80%)
- Nerves 9 and 10 - Swallowing (50%)

(From CHARGE Foundation Parent Manual - 1999)

2005 – 38 anomalies?? (Continued)

- Atresia of the esophagus
- Skeletal anomalies
- Sleep apnea
- Tracheo-esophageal fistula
- Cleft lip / Cleft palate
- Hypocalcaemia
- Seizure disorder

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.”

CHARGE - the most ‘multi sensory impaired’ of all syndromes

Problems with the perception of:

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

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

“...the immense difficulties that children with CHARGE face in almost everything that they do, and, as a consequence, the very high levels of stress with which they must live for much or even all of their lives. Time spent trying to reduce stress levels, and trying to give the children acceptable strategies for doing this for themselves, must be one of the most precious gifts we can offer them, and one of the biggest favors we can do ourselves as educators, therapists, and family members.”

10

David Brown on CHARGE - 1997

“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”

The Early Months/Years

- Major medical issues take precedence
- Life and death issues dominate
- A large team of specialists becomes involved
- Outcomes are unknown & prognoses are grim
- Major surgeries take place
- Bonding between child and parents is interrupted
- Procedures take place without warning or preparation
- Communication with the child is minimal
- Hostile environments
- Multi-sensory impairment

12

Most children with CHARGE function as if they have deaf-blindness in their early life, and benefit from the earliest possible application of good deaf-blind educational principles and practice



13

With infants with CHARGE mental retardation should never be assumed until it is proven



14

Most children with CHARGE significantly under-function at birth, and have more potential for development than they reveal to most early assessment procedures



15

The educational needs of infants with CHARGE should be addressed as soon as possible after birth even when medical issues seem to be the only, or the top, priority



16

What do you mean - "Too early"!!??!

- Light sources and levels
- Sound sources and levels
- The cot environment
- Touch cues (firm touch)
- Personal markers
- Slow and fully supported lifting and moving
- Reciprocal interaction games
- Communication/Language
- Educating others

17

"The Forgotten Senses"

PROPRIOCEPTION

The receptors are in the muscles and joints throughout the body

Tells us about the position of our body and all of our limbs, and if anything is moving

VESTIBULAR

The receptors are in the Inner Ears

Tells us about head position & the pull of gravity, detects motion, and it has very close links with the eyes and vision

When the proprioceptive sense is not working properly some common outcomes may be:

- Inability or reluctance to push up on the hands and arms when laying face down, because of an inability to “feel” and control the joints in the fingers, wrists, elbows & shoulders.
- Inability or reluctance to stand and bear weight because of an inability to “feel” and control the joints in the toes, ankles, knees & hips, while also maintaining vertical stability in the spine.
- Frequent use of the arms and hands to prop the head/upper body, or leaning against furniture, walls, posts, trees, or other people.

When the proprioceptive sense is not working properly some common outcomes may be:

- Feet stamped or slapped repeatedly on the floor when cruising or walking to maximize the tactile and proprioceptive sensation (“feeling the feet”).
- Later on, walking tip-toe to maximize the pressure input through the feet, ankles, calves, knees, thighs and buttocks (another way of “feeling the feet”).
- Clumsy, poorly coordinated movements, often with self-taught correction strategies.

When the proprioceptive sense is not working properly some common outcomes may be:

- Use of too little force, or excessive force when touching, patting, grasping, pushing or pulling things, or lifting and placing things - may often drop objects.
- Seeking strong pressure or stretching 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 body parts 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.

Dr. George Williams “Balance in CHARGE”
CHARGE Syndrome Foundation Manual

Vestibular function has a role in...

- Detecting motion
- Detecting & responding to gravity
- Providing stability during body movement
- Locating body parts & developing body schema
- Influencing muscle tone and posture

22

Dr. George Williams “Balance in CHARGE”
CHARGE Syndrome Foundation Manual (cont.)

- Facilitating the crossing of the midline
- Motor control, coordination & sequencing
- Assisting with auditory & visual perception
- Modulating arousal & alertness for attention and calming

23

Early effects of poor (or missing) vestibular function
in children with CHARGE Syndrome

(Brown American Journal of Medical Genetics, March 2005)

- Low muscle tone (“floppy muscles”)
- Poor head control & poor ability to resist gravity
- Strong postural insecurity when held upright
- Marked preference for being flat on the back
- Delayed mobility & unusual movement patterns (eg. back scooting, side-winding, five-point crawl)

24

Early effects of poor (or missing) vestibular function in children with CHARGE Syndrome
(Brown, cont.)

- “W” sitting for broader, more secure base
- Better visual, auditory and fine motor skills in supine than in the upright position
- Poor bilateral coordination, hand and eye dominance is either total or missing
- Fatigue after trying to resist gravity for periods of time
- Difficulties with self-regulation

25

Health issues

Complex health issues

Availability of medical/para-medical services

Balancing health and educational needs

Making health care educational

CHARGE management issues

- Constipation - Autonomic nervous system?
- Toilet training delays– nerve abnormalities?
- Sleep disturbances - abnormal circadian cycle
- Cyclic vomiting/abdominal migraines
- Sensory integration issues
- Behaviors

27

*Communication with one's own body

*Communication with one's immediate environment

*Communication with the wider world

Communication Steps

- 1) Alert the child to your presence
- 2) Introduce yourself (who are you?)
- 3) Alert the child to the coming activity
- 4) Introduce the activity
- 5) Do, *and discuss*, the activity
- 6) On completion, review what you have done (eg. What is different now?)
- 7) Let the child know that you are leaving

29

Communication Options

- Objects & calendars
- Sign language
- Speech with hearing aids
- Speech with a cochlear implant
- Visual programs
- Signed English
- Reading and writing

30

The way to success?.....

.....the child's preferred mode(s) of communication

31

Obstacles to the clear articulation of speech for people with CHARGE (1)



- Hearing Impairment
- Vision Impairment
- Facial Palsy
- Low muscle tone
- Poor tactile sense
- Oro-facial clefting
- Enlarged Tongue
- Poor tongue movement

32

Obstacles to the clear articulation of speech for people with CHARGE (2)

- Small lower jaw
- Larynx/Pharynx anomalies
- Breathing difficulties
- Swallowing difficulties
- Dental anomalies
- Delayed/immature eating skills

33

Obstacles to the clear articulation of manual signs for people with CHARGE (1)

- Low or poorly modulated muscle tone
- Poor tactile sense
- Poor proprioceptive sense
- Poor spatial awareness
- Dyspraxia
- Poor body awareness



34

Obstacles to the clear articulation of manual signs for people with CHARGE (2)



- Poor bilateral coordination
- Poor sense of balance
- Various postural difficulties
- Low vision or blindness / visual field losses
- Skeletal anomalies

35

Groping for understanding over the years

- “social immaturity”
- “lack of empathy”
- “sudden unpredictable extreme mood changes”
- “lack of a shared frame of reference”
- “disproportionate emotional responses”

[Self-regulation]... “is defined as the capacity to manage one’s thoughts, feelings and actions in adaptive and flexible ways across a range of contexts”

Jude Nicholas, CHARGE Accounts, Summer 2007

37

What is ‘self-stimulation’ ?

- The constancy of sensory feedback.
- Any sensory input that we seek which is not directly the result of a specific activity (such as making coffee, drinking from a glass, getting dressed, walking).
- Any sensory input that we seek which facilitates an activity but is not an inherent part of it.

Statement 1

Sensory inputs have a significant & direct impact on arousal levels

The 9 levels of arousal (Carolina Record of Individual Behavior)

- Uncontrollable agitation
- Mild agitation
- Fussy awake
- Active awake
- Quiet awake
- Drowsy
- Active sleep
- Quiet sleep
- Deep sleep

40

1. Where are you on the ladder of arousal?
2. Where do you need to be?
3. How can you get there?

Statement 2

We all self-stimulate (a lot) to maintain alertness, to wake up, to calm down, to maintain postural control, to keep/get comfortable, to occupy our minds, to self-regulate, to maintain attention, to keep sane, and generally to improve our functioning to achieve our goals

Statement 3

CHARGE is the most multi-sensory impaired syndrome that we know, and poor self-regulation is a very common feature of this population

What is CHARGE Behavior??

- Impulsivity
- High anxiety
- Obsessions
- Self stimulation
- Poor self-regulation
- Executive function disorder
- Attention Deficit Hyperactivity Disorder
- Pervasive Developmental Disorder
- Obsessive Compulsive Disorder
- “Autistic-like” behaviors
- Deafblindness
- Multi-sensory problems.....(and so on!)
- Laziness, stubbornness, aggression

...behavioral difficulties, some of which may be described as autistic-like, and obsessive-compulsive, with attention difficulties... There are potentially multiple sources for these difficulties... 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... 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.

Tim Hartshorne, CHARGE Foundation Professional Packet

Statement 4

Most children with CHARGE are not in touch with/do not feel their bodies very well

Yes, I believe that posture should be included as a “self-stimulation” and/or a “self-regulation” behavior (especially for people with CHARGE syndrome)

Statement 5

Sensory deficits and poor sensory perception make children with CHARGE self-stimulate in mostly normal ways – but with more intensity, more persistence, and for a longer period of their lives than “normal”

Statement 6

For various reasons children with CHARGE may have poor social awareness, so self-stimulation behaviors may be more obvious

Statement 7

Attempts to stifle and stop self-stimulation behaviors are likely to result in worse self-regulation and generally less good functioning

If it isn't dangerous or illegal, ask "What does it mean?", and then intervene to try to answer that question, NOT to stop the behavior as the primary aim

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

"Many people with CHARGE demonstrate difficulties with vocabulary recall, initiating communicative exchanges, and with clearly articulated expression, in the abstract forms of spoken and/or sign language. Provision of a communication mode with a concrete component (eg. objects, symbols, pictures, written words) can be of immense help in aiding recall, in encouraging initiations, in clarifying meaning, and in generally fostering a more confident, animated, and fluent communicative style."

52

"Exploring executive functioning"-Amanda Kirby

- **Activation** – organizing & prioritizing, initiating, getting started
- **Focus** – sustaining & shifting, completing
- **Effort** – regulating alertness
- **Emotion** – managing frustrations, modulating emotions, keeping perspective
- **Memory** – remembering, accessing recall, recognizing & remembering a sequence
- **Action** – monitoring & regulating self-action without impulsivity, or poor context or poor pacing

Statement 8

Observing how and when a child self-stimulates will offer invaluable insights into who they are and how they work, for assessment, teaching, behavior management, and relationship building

Teaching Strategies (Brown 2005)




- Individualization
- Relationships
- Stress Control
- Positioning & support
- Motivators
- Pacing
- Consistency
- Routine-based
- Adaptations
- Appropriate communication/ language
- The Just Right Challenge

The Future

- Medical research & advances - genetics, immunology, digestive system, osteoporosis, vestibular issues, sleep disturbance, low muscle tone
- Improved identification and access to services
- Age-related developments, both medical and behavioral
- Investigation of behavioral issues, especially self-regulation & executive dysfunction
- Improved educational strategies
- Improved awareness of the correct assessment procedures and teaching strategies

"All you need to know about the cranial nerves and more":



Dr. Kim Blake
Professor Pediatrics
IWK Health Centre and Dalhousie University
kblake@dal.ca

Oberwesel, Germany 2014

Halifax, Nova Scotia, Canada



Objectives

After this presentation you will understand more about:

1. The cranial nerve anomalies in CHARGE Syndrome, particularly in relation to feeding.
2. The Cutting edge research involving the Zebrafish in relation to the cranial nerves.

Let's Rate Your CHARGE's Eating Difficulties Over the Years

0	1	2	3	4
None	A little (reflux, choking, no G or J tubes)	G or J Tube, less than 12 months	G or J tube feeding more than 12 months	Extreme difficulties, one of the biggest problems

CASE HISTORY

4 Major & 3 Minor

MAJOR

- C - Coloboma [Left Eye].
- C - Choanal Atresia [Right].
- C - Cranial Nerves [VII (Right), VIII, IX, XI].
- C - Characteristic Ears [Severe SNHL].



M.C.


MINOR

- C - Cardiac - aberrant subclavian artery, bicuspid aortic valve.
- C - Characteristic CHARGE face.
- D - Developmental delay - balance, expressive speech.

Hidden Structural Problems

CASE HISTORY

- Feeding Issues
- Severe renal hydronephrosis
- Abnormal temporal bones



Cochlear transplant 2000

Nissens fundoplication and tonsillectomy 2001

Blake et al 1998 CHARGE Association - An update and review for the primary Pediatrician.

Feeding Issues

- Poor sucking and swallowing
- Velopharyngeal in-coordination
- Gastroesophageal Reflux (GER)



Dobbelsteyn C, Blake KD. 2005. Early Oral Sensory Experiences and Feeding Development in Children with CHARGE Syndrome: A Report of Five Cases. Dysphagia. Vol : 89-100.

Feeding Question #1

“My 2 year old has been getting more picky and will not eat lumps. We never needed a tube but she’s losing weight and now has regular hiccups. She was on ranitidine as an infant but we weaned her off this.”

The family doctor feels that this is just the terrible two’s and not to worry.

Cindy Dobbelsteyn, et al. Feeding Difficulties in Children with CHARGE Syndrome: Prevalence, Risk Factors, and Prognosis. Dysphagia. 2008 Vol. 23, No. 2, p. 127

Treatments for Gastroesophageal Reflux (GER)

1. Behavioral treatment – raising the bed, small frequent meals, limiting foods that promote reflux such as tomatoes, meat, chocolate.
2. Medical management
 - o ranitidine 8mg/kg per day in 1-2 divided doses (for babies 3 divided doses)
 - o Prevacid (lansoprazole)- 1-2 mg/kg per day at the beginning of the day (occasionally twice a day)
 - o Domperidone (Motilium) – 4 times a day before meals

Also consider cow’s milk protein intolerance

Feeding Question #2

After gastrostomy removal some children cram their mouths with food, why?

- oral hyposensitivity
- Need for substantial amount of food in mouth before bolus preparation occurs



Two friends having lunch.

“Hot Dog in 3 Seconds Flat”



Ate quickly and swallowed without chewing


Ideas for Treatment

- external pacing - Therapist
- small manageable bites
- wait until mouth is clear before offering more



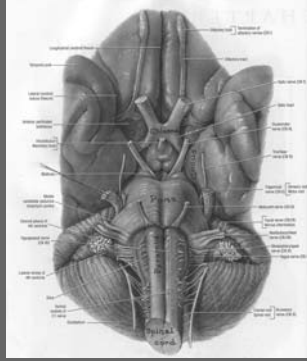
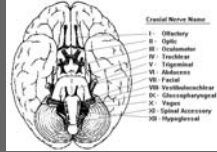
Cranial Nerves

- I Olfactory
- II Optic
- III Oculomotor
- IV Trochlear
- V Trigeminal
- VI Abducens
- VII Facial
- VIII Vestibulocochlear
- IX Glossopharyngeal
- X Vagus
- XI Accessory
- XII Hypoglossal



Yale Center for Advanced Instrumental Media's Web Site: <http://info.med.yale.edu/caim/cnerves>

Cranial Nerves Arising from Base of Brain

Tenth Edition Grant's Atlas of Anatomy

Cranial Nerves – 12 Pairs Motor & Sensory


I	Smell - anosmia
II III IV VI	Eye movement
V	Weak chewing & sucking, migraines
VII	Facial nerve weakness
VIII	Hearing & balance problems
IX X	Internal organs (heart, gut)
XI	Shoulder movements
XII	Tongue

Blake KD, et al. Cranial Nerve manifestations in CHARGE syndrome. Am J Med Genet A. 2008 Mar 1;146A(5):585-92.

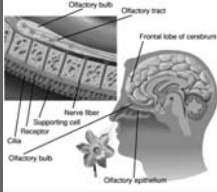
How many of you have CHARGE's with suspected cranial nerve problems?

No 1 2 3 More

CHARGE hands up



Olfactory Nerve (CN I)




There is a test kit available

Chalouhi C, Faulcon P, Le Bihan C, Hertz-Pannier L, Bonfils P, Abadie V. Olfactory evaluation in children: application to the CHARGE syndrome. Pediatrics 2005

The Cranial Nerves of the Eye

II	Optic
III, IV, VI	Eye muscle movement



Retinal Nerve Coloboma

In CHARGE syndrome visual perception (II) affected, less often eye movement.

McMain K, Blake K, Smith I, Johnson J, Wood E, Tremblay R, Robitaille J. Ocular features of CHARGE syndrome. 2008 Oct;12(5):460-5.

Eyes are at Risk With Facial Palsy

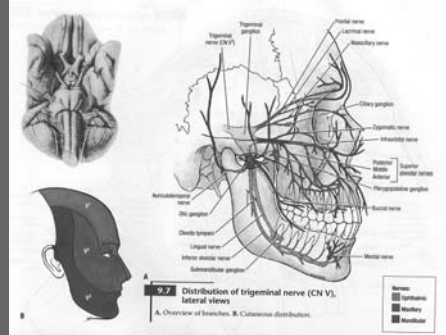


- Dry eye
- Damaged cornea
- Light sensitivity

Using weights in the eyelids



Trigeminal Nerve (CN V)



Tenth Edition Grant's Atlas of Anatomy

Muscles of Mastication – Cranial Nerve V



Feeding issues are often severe.

Two friends, MC and KW, having lunch.

Cranial Nerve VII - Facial



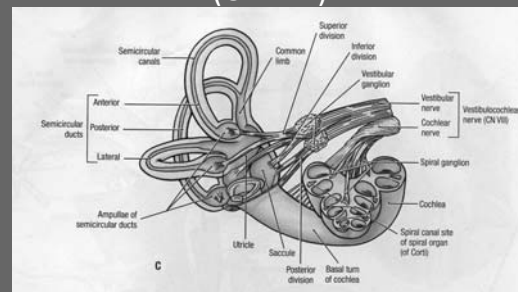
Web Site: <http://info.med.yale.edu/caim/cnerves>

Mobility & balance in CHARGE has improved with physiotherapy



International CHARGE Conference 2011

Temporal Bones – Balance & Hearing (CN VIII)



Tenth Edition Grant's Atlas of Anatomy

Lower Cranial Nerves IX-XI

Cranial Nerve	Function	Symptom of Dysfunction
IX	Taste Salivation Swallowing	Gag reflex Swallowing
X	Phonation Swallowing	Gag reflex Swallowing
XI	Head and shoulder movement Laryngeal muscles	Shoulder drop Winging scapula

IX X XI Cranial Nerves – Abnormality in the supranuclear region.
 Poor suck – swallow coordination, neonatal brain stem dysfunction (NBSD)

Cranial Nerve IX

Tenth Edition Grant's Atlas of Anatomy

Cranial Nerve X Vagus

Tenth Edition Grant's Atlas of Anatomy

Cranial Accessory Nerve XI

Tenth Edition Grant's Atlas of Anatomy

Summary of Cranial Nerve (CN) Findings in CHARGE syndrome

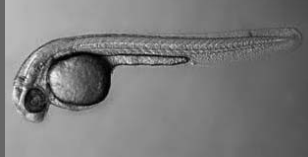
- Dysfunction of cranial nerves is more frequent and multiple.
- The extent and involvement of cranial nerves may reflect the clinical spectrum.
- CN VII - is more frequently associated with other CN's
 - is seen in those individuals more severely affected.
- CN V – “muscles of mastication” affected in CHARGE.
- Structural brain malformations highly associated with CN.

Role of Chd7 in Zebrafish: A Model for CHARGE Syndrome. PLoS One. 2012;7(2):

Patten SA, Jacobs-McDaniels NL, Zaouter C, Drapeau P, Albertson RC, Moldovan F.
 Sainte-Justine Hospital Research Center, Montreal, Quebec, Canada.

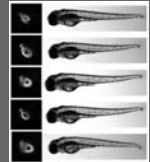
The Zebrafish

- Zebrafish make an excellent model organism to study rare pediatric single gene diseases because:
 - Conserved genetics
 - Ease of genetic manipulation
 - Embryonic transparency
 - Rapid development



Zebrafish and CHARGE

- CHD7 gene is conserved in the zebrafish
- CHD7 knock down has demonstrated the following physiological effects in the zebrafish:
 - Dysmorphic heart
 - Smaller eyes
 - Curvature of the body axis
 - Disruption in the number, organization, and patterning of the cranial nerves (mainly V, VII, and X)



Continuing Research

- Teaming up with Dr. Berman, who has expertise in modeling rare diseases in zebrafish, we hope to explore three main areas of CHARGE syndrome:
 1. Gut motility and function:
 2. Heart anomalies and genetics
 3. Cranial nerve anomalies



Thank you!

Young
Researchers



Questions:

Behavior as self-regulatory adaptation, or “I can’t believe my child just did that!”

Tim Hartshorne
Central Michigan University




Behavior often seen in persons who are Deafblind

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

How to make sense of it

- The kid has a syndrome!
- It’s pathological and should be eliminated
- It’s due to frustration and pain
- It’s communication
- It works for the kid

Not because the behaviors guarantee success, but because they serve a purpose



•The purpose is Self-Regulation

The primarily voluntary regulation of cognition, behavior, emotion, and physiological states for the purpose of goal-directed actions

But it does not always work well


•Self-Regulation

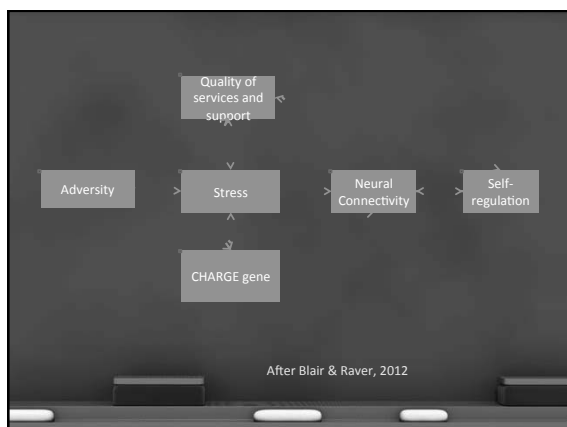
- Managing the threshold of arousal
- Processes of self-control
- Both suppresses and encourages; inhibits and promotes
- Supports homeostasis of the system
- Critical to development



•Self-regulation problems in CHARGE

- Rapid changes in arousal levels
- Melt downs
- Unfocused behavior
- Diagnoses
 - OCD – a way to reduce stimulation and exercise control
 - ADHD – a problem with regulating sensory and behavioral stimulation and focusing on a goal
 - Tic disorder – a stress response to lack of control over environment
 - Autistic-like behavior – the failure of regulation strategies, and the adoption of dysregulated behavior






Adversity

- Fragile health
 - Breathing problems
 - Multiple hospitalizations
 - Multiple surgeries/anesthesia
 - Multi-sensory impairment
 - Defects in major organs
- Nervous parents
- Sources of stress
 - Social relationships
 - School
 - Family
 - Abuse



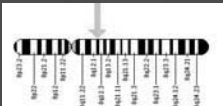
•Quality of Services and Support

- Medical or specialist knowledge
- Needs multi-disciplinary medical and educational teams
- Parent-Professional relationships
- Extent of social support
- Parent and family resilience




•CHD7 Gene

- Regulatory gene
 - Neural crest
 - Placode cells
- Multisensory impairment
- Major organs may be affected
- Vestibular functioning impaired
- Epigenetics
 - NICU experience




•Stress

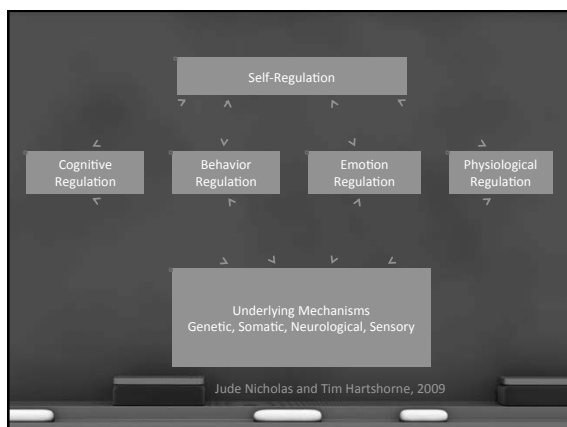
- Endocrine regulatory system
- Perception of adversity
- Availability of resources
- Response of professionals
- Response of family



•Neural Connectivity

- Prefrontal cortex and executive function
 - Reactive forms of learning and behavior
 - Reflective forms of learning and behavior
- Neuropsychological control over behavioral schemas
 - Routine control
 - Supervisory attentional system

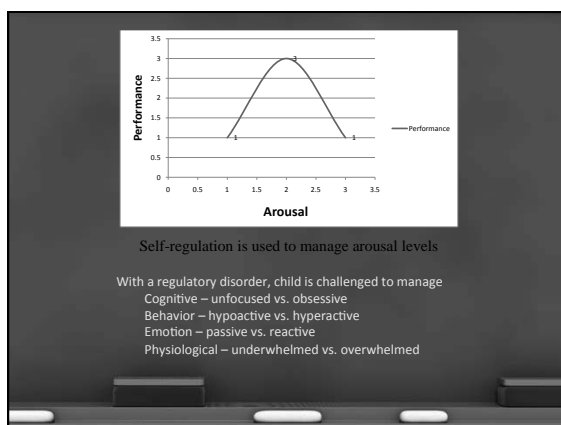




- ### Self-regulation Scale
- I have a hard time paying attention and my mind tends to wander.
 - When I really need to pay attention I can focus my mind.
 - I can readily prioritize the things I need to get done in a day.
 - I become overwhelmed when faced with too many things to take care of.
 - I get upset a lot and cannot find any way to get rid of those feelings.
 - When I really need to control my feelings I can do it.
 - When there is nothing going on I have to create it.
 - When I am in a noisy crowd I have to find a way to leave.

Dunn Conceptual Model

Arousal of thoughts, behavior, feelings, sensations	PASSIVE Self-regulation Strategies	ACTIVE Self-regulation Strategies
Habituation	Non-reactive Tune it out	Sensation Seeking
Sensitization	Reactive to Stimuli	Sensation Avoiding



- ### Self-regulation begins with a goal
- What do you want to have happen?
 - What must you do to make it happen?
-

- ### Study for an exam
- Cognitive
 - Behavioral
 - Emotion
 - Physiological
- Strategies?
-

•Cognitive Regulation

Today I mean to polish an article. But so far it has been hard to keep my mind focused. When a new email pops up on my computer I am distracted, and if I answer it, I may even forget what I had been doing. I think about an appointment I have later in the day and my eyes leave the manuscript. I read a passage, and then wonder if that is really what we meant to say, and then my mind is no longer on the writing. To counter these forays away from the task, I re-focus my attention, re-energize my motivation, and keep telling myself to stay working and avoid the distractions.



•Cognitive Regulation

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



•Behavior Regulation

Flossing teeth is not an easy habit to acquire. At least it was not for me. After having to endure a bit of dental surgery due to the state of my gums, I had a new goal in mind: never repeat that. At first I hated flossing. I had to force myself every evening before bed. But my goal was strong and so I stuck with it. Gradually it became something I did pretty much automatically. Even on those late evenings when I just wanted to fall into bed, I found I could not succumb until those teeth were flossed. This is now a very well regulated behavior. My dentist is very pleased with me.



•Behavior Regulation

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



•Emotion Regulation

I am starting to be stressed by an article. I wanted it completed some time ago. As my emotions get on edge, I become a bit grumpy, and find that I am less pleasant with the people around me, particularly my co-authors. As I notice my feelings become more aroused, I remind myself that this is a group process, we are all busy, we are making progress, and it will be good. Telling myself these things helps me to relax a bit and calm myself down. I have lowered my level of emotional arousal and am better able to focus on the task at hand. I have self-regulated my emotions.



•Emotion Regulation

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



•Physiological Regulation

Once in graduate school when I was studying at my favorite study carrel in the library, someone sitting right behind me started tapping the desk with his pencil. It slowly drove me crazy. The noise completely distracted me, I began to squirm in my chair, I started to sweat, and my stress levels increased dramatically. I lost the ability to keep myself in a nice, calm state. Several interventions posed themselves, one was rather violent; but, the most reasonable was to move to a different carrel.



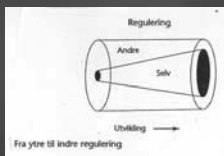
•Physiological Regulation

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



•Supporting self-regulation

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



•Scaffolding

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



•Components of Scaffolding

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

•The Shape Sorter



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

•Interventions

- Cognitive
 - Help the child learn to break down larger goals, tasks, or problems into shorter, more discrete tasks
- Behavioral
 - Provide feedback to the child on how others react to their behavior and help child to understand the consequences
- Emotion
 - Help the child by creating environments that assist the child to self-sooth
- Physiological
 - Squeeze technique
 - Hand on arm or leg
 - Brushing and joint compression

•Supporting self-regulation

- Make goals more explicit
- Use social stories
- Utilize prompts
- Provide social support
- Utilize scaffolding
 1. Identification of the problem to be solved
 2. Focus activities on outcomes and goals
 3. Frustration control
 4. Reducing the complexity of the task
 5. Marking critical relevant features
 6. Modeling



•Summary

- Children with disabilities often have poorly regulated systems
- This is centrally related to stress, deriving from adversity, quality of supports, and genetics
- The child's attempts to self-regulate manifest as peculiar behavior, often labeled as challenging
- They will do better socially and academically if they can learn to self-regulate
- They can only develop self-regulation skills slowly while they experience a lot of scaffolding from the adults in their lives

•Thanks to my Lab


- Maria Ramirez
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- Gretchen Imel




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Educational provision for learners with CHARGE



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




Types of educational provision

Types of school	% of children from study
Hearing impaired/ visually impaired	27.5%
Physical impairment	7%
Severe learning difficulties (including 4 children in deafblind provision)	27.5%
Mainstream	31%
Independent/ non-maintained	7%

Deuce, Howard, Rose and Fuggle (in press)

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Choosing an educational placement

“The main driving force must be clear identification of the child’s educational needs and how these are to be met.”

The CHARGE Information Pack for Practitioners: Factsheet 16


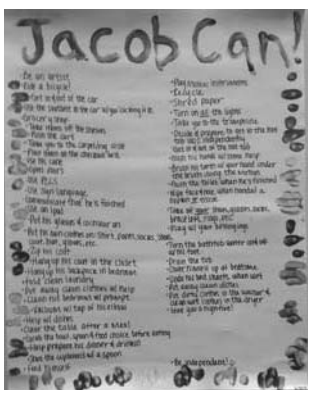
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First Steps

Spend time identifying the child or young person’s strengths and needs

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Choosing the educational approach

“The philosophy of deaf-blind education offers the best match.”
(Brown, 2011)

“The learner with CHARGE syndrome additionally must cope with multiple challenges along with limited access to both vision and hearing.”
(Majors, 2011)

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Features of CHARGE that may affect learning



- Visual impairment and hearing impairment
- Other sensory impairments (vestibular, proprioceptive, touch, smell, taste)
- Sensory integration difficulties
- Low tone
- Neurology (executive function difficulties)
- Cranial nerve anomalies
- Sleep disturbances
- Medical intervention, hospitalisation and pain
- Potential later onset features

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What might be required? 1



Input from professionals including:

- Specialist teacher of the deaf
- Specialist teacher for the visually impaired
- Specialist teacher for multi-sensory impairment/deafblind (with knowledge and experience of CHARGE)
- Specialist teacher for physical disabilities
- Speech and language therapist (speech and communication/ dysphagia)
- Physiotherapist
- Occupational therapist (plus sensory integration)
- Paediatric mobility or habilitation officer
- ICT specialist
- Support from an intervenor
- **PLUS** involvement from the child or young person and their family

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What might be required? 2



An appropriate and planned response to:

- Sensory needs (meeting visual, auditory and other sensory needs)
- Communication needs- access to appropriate modes of communication and communication systems
- Physical needs- to develop gross and fine motor skills; seating and positioning
- Orientation and mobility needs
- Any fatigue issues- a reduced timetable, a safe rest area
- A need for clear routine and structure
- ICT requirements (including alternative recording methods)
- Behavioural issues

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9

What might be required? 3



A response to the child/ young person's sensory integration difficulties:

- A sensory integration assessment by a qualified and experienced occupational therapist, leading to the creation of a 'sensory diet'
- Strategies to manage sensory overload
- Strategies to manage fatigue and facilities for safe rest periods (may include adopting a horizontal position)
- Alternating between active and passive activities throughout the day
- Providing sensory breaks that allow the child time to refocus and organise the sensory information received
- Use of specific techniques identified and monitored by the occupational therapist

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What might be required? 4



A response to the child/young person's executive function difficulties. Increasing recognition is being given to the occurrence of executive dysfunction in individuals with CHARGE. These can create difficulties in:

- social and emotional development
- monitoring and regulating behaviour
- transitions between activities, different environments and people
- working independently
- initiating activities and interactions
- self-help and self-organisational skills
- completing a task independently
- maintaining a well-regulated sensory state

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11

What might be required? 5



Strategies for responding to executive function difficulties:

- Use of a concrete component to aid recall, encourage initiations, and clarify meaning
- Use of a communication timetable to support independence and self-organisation
- Teaching improved self-organisational skills (preparing a swimming bag, planning and shopping for ingredients to cook a simple meal, etc.)
- Support to complete a wider range of tasks more independently; providing support at the beginning of a task and then reducing the level of support, but being available to help re-focus on the task when distracted
- Develop templates for often repeated procedures
- Teaching functional life skills
- Carefully planned transitions
- Clearly lay out expectations
- Plan in advance
- Establish a response to help support the 'stop' function

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12

What might be required? 5



Support for the child or young person's social and emotional development:

- Developing and consolidating emotional vocabulary and what it means when an emotion is being felt
- Support to identify and label own emotional state. If the child is upset it may be difficult to have this conversation at the time, and may need to be done retrospectively
- Using role play to model emotions and how to respond in specific situations. Young people with CHARGE sometimes have in adopting another persona and the possibility of undertaking role play through a set of figures
- This idea can also be used to re-enact a previous situation –Different potential responses can be acted out in turn and then a discussion undertaken of what would be the best response

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What might be required? 6



- Rewarding positive, appropriate emotional responses (e.g. using Positive Behavioural Support)
- Using other concrete aids such as a colour or face chart to describe feelings or a 'traffic light'/number scale to describe the strength of a feeling
- Preparing beforehand for situations that are likely to be emotionally difficult and equipping with strategies to help regulate their emotional state without the need for an outburst.

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14