

## Program

(subject to change)

### Friday, 4<sup>th</sup>, July, 2014:

- 12.00 o'clock Start of individual meetings with the CHARGE experts Dr. David Brown, Gail Deuce, Prof. Dr. Tim Hartshorne, Dr. Kim Blake (time lists at the reception desk)
- 1.30 p.m. **Claudia Junghans, President**  
Official welcome  
**Welcoming speech**  
**Matthias Rösch**  
commissioned for the interests of handicapped people in Rheinland Pfalz
- 2.00 p.m. **Lecture by Dr. Kim Blake**  
"CHARGE 101: Anaesthesia, Botox, Sleep Apnea and FAQ's"
- 4.00 p.m. **Lecture by Prof. Conny van Ravenswaaij**  
"All you want to know about CHARGE and genetics"
- 6.00 p.m. Dinner
- 8.00 p.m. Family introductory session, campfire bread (bannocks) and get-together, abseil adventure for everybody who is courageous (children from 8 years)

From 10 p.m. Football World Cup on TV

### Saturday, 5<sup>th</sup>, July, 2014:

From 7.30 a.m. Breakfast

- 10.00 a.m. **Lecture by Dr. David Brown**  
"How do you 'Follow the Child'?"
- 12.00 o'clock Lunch
- 2.00 p.m. **Lecture by Gail Deuce**  
"Person centered planning approach"
- 3.30 p.m. Coffee Break
- 4.00 p.m. **Lecture, Dr. David Brown and Prof. Dr. Tim Hartshorne**  
" Back to the basics and beyond"
- 6.00 p.m. Dinner
- From 8.00 p.m. Presentation of some CHARGE affected young people  
"This was our first CHARGE teenager weekend! "  
Presentation of the teenager-film.

From 10 p.m. Football World Cup on TV

### Sunday, 6<sup>th</sup>, July, 2014:

From 07.30 a.m. Breakfast

- 09.00 a.m. **Lecture by Dr. Kim Blake**  
" Bone health & Puberty - Not a Humerous Issue "
- 10.30 a.m. **Lecture by Prof. Dr. Tim Hartshorne**  
" Resilient Parenting "
- From 12.00 o'clock Final group photo/ Lunch
- 1.00 p.m. **Claudia Junghans / Dr. Julia Benstz**  
Reflection on talks, opportunity to put questions to the CHARGE experts
- 2.30 p.m. End of the conference and departure

#### Please note:

All presentations in English will be **simultaneously** translated.  
Interpreters will also be available (if required) for the individual interviews with CHARGE experts. For a communication without **barriers sign language interpreters** will be on hand.

#### Child care:

- Friday : 2.00 p.m. to 6.00 p.m.  
Saturday : 8.30 a.m. to 6.30 p.m.  
Sunday : 8.30 a.m. to 2.30 p.m.

#### The following extended items will be offered as a part of the child care:

- craft stations run by a team of Gerrit Event Concept
  - High wire course, climbing wall, abseil adventure run by a team of Martin Becker
  - Mobile Vehicles & Games Circus held outdoors
- Sufficient qualified staff will be available, supported by sign language competent persons.

#### **SUPPORTING PROGRAM:**

- Program for our CHARGE teenagers
- Grandparents seminar (Saturday, 2 p.m.)
- CHARGE sales booth
- Tombola
- Books table
- Poster exhibition

Gefördert durch die

**Aktion  
MENSCH**

## CHARGE 101 Anaesthesia, Botox, Sleep Apnea and FAQ's



Dr. Kim Blake, MB, MRCP, FRCPC

Oberwesel, Germany July 2014

## Objectives

- To help you understand the risks of anaesthesia in CHARGE syndrome.
- To explain the use of Botox injections into the salivary glands.
- Research findings about sleep apnea and benefits of tonsillectomy and adenoidectomy.

## There are Always Risks of Complications with Anaesthesia

- "...you sign a consent"
- Are you informed?
  
- Are Individuals with CHARGE Syndrome More at Risk?

## If yes, what are the risks and who should know?



APSEA 2006

## Growing up With CHARGE Syndrome



Age 0-2 years: 7 surgeries



Age 2-4 years: 3 surgeries



Age 4-6 years: 6 surgeries

Kennedy

## Kennedy's Four ICU Admissions

- 2 weeks – open heart surgery
- 6 months – G-tube/fundoplication extubation attempted (x 3)
- 18 months – aspiration pneumonia
- 6 yrs – heart surgery – pneumonia after heart surgery

## Postoperative Airway Events of Individuals with CHARGE Syndrome

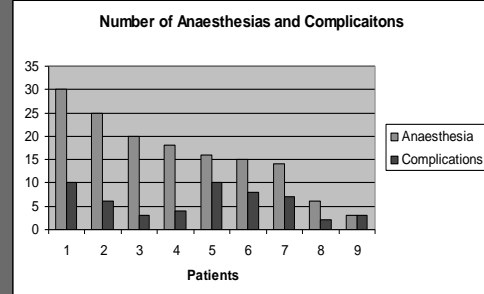
Population n=9  
 Mean age 11.8 years ( $\pm$  8.0)  
 215 surgeries (mean 22 per child)  
 147 anaesthesias (mean 16 per child)



Postoperative events (reintubation for apneas and desaturations, airway obstruction due to excessive secretions)

Blake K, *International Journal of Pediatric Otorhinolaryngology*, 2009

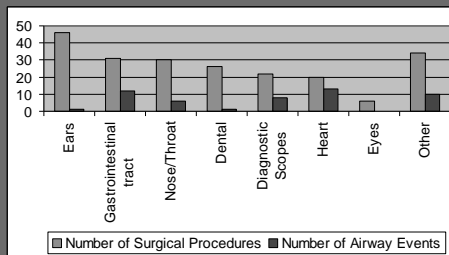
## Results



35% (51/147) of anaesthesias resulted in post-operative events (>60% were major)

## Results

Anaesthesia related events occurred most often with heart, diagnostic scopes (L/B/E) gastrointestinal tract procedures



## Results

Number of surgical procedures per anaesthetics with resulting postoperative airway events.

Number of surgical procedures	Number	Post-operative Events	Percent resulting in airway events
1	94	37	39% (n= 37/94)
2	36	8	22% (n= 8/36)
3+	15	5	33% (n= 5/15)

P=0.1 Combining multiple procedures under one anaesthesia does not lead to an increase in post-operative events.



MacKenzie & Kennedy sharing lunch

## Results

Feeding procedures and rates of postoperative airway events.

	Number of Anaesthesias	Airway Event	No Airway Event	Significance
G/J tube	82	36	46	Yes p=0.0092
No G/J tube	63	15	48	
Nissens fundoplication	79	33	46	Yes p=0.049
No Nissens fundoplication	66	18	48	

Having a G/J tube or Nissens fundoplication increases your child's risk of post-operative airway events

## Summary

- 35% of anaesthetics resulted in post-operative events
- Heart, diagnostic, and gastrointestinal tract procedures result in the most events
- At least one event occurred with every type of surgery except for eyes

Blake K. *International Journal of Pediatric Otorhinolaryngology*, 2009

## Summary cont'd

- High risk of complications in individuals with Nissen fundoplication and/or gastrotomy/jejunostomy tube
- What about individuals with CHD7 mutations who have mild clinical criteria?
  - Will they be at risk in the future?
  - Have they actually been challenged with surgeries?

## Frederick's Story

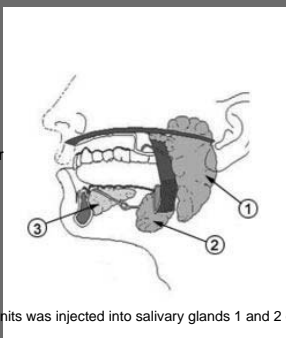


## Freddy at 2 Months

- Difficulty with intubation
- ToF repair, vascular ring repair, PDA ligation
- Increased oral secretions
- Multiple attempts at extubation

## Site of Botox Injections

1. Parotid glands
2. Submandibular glands
3. Sublingual glands



Botox 7.5 units was injected into salivary glands 1 and 2 on each side

## Freddy at 7 Months

- Aspiration pneumonia from oral secretions
- Gastroesophageal reflux
- Required ventilation

## Botox Injection

Submandibular Gland Via Ultrasound and Parotid Gland by Palpation



10 Botox Units/gland

## Botox Injection

Prophylactic Use to Prevent Increase in Oral Secretions and Aspirations (4-5 monthly)



Freddy's growing up!



## Summary - Botox

- Botox injections into the salivary glands may help to reduce the oral secretions (needs repeating every 4-5 months).
- Reduction in oral secretions may help prevent aspiration and pneumonia
- May help prevent and/or removal tracheostomy

*MacCuspie. AJMG 2011*

## Obstructive Sleep Apnea and Post Operative Airway Events



How many of you have sleep issues with your CHARGER's?

## MacKenzie's Story



- 27 surgical procedures
- 18 anaesthetics
- 4 complications
- Multiple ICU admissions

## Understanding Sleep Apnea in Children with CHARGE Syndrome

In Children 0-14 years old

### Authors:

Carrie-Lee Trider  
Dr. Gerard Corsten  
Dr. Debra Morrison  
Meg Hefner  
Dr. Sandra Davenport  
Dr. Kim Blake



Carrie-Lee & Freddy

*Trider CL. International Journal of Pediatric Otorhinolaryngology 76 (2012) 947-953*

## Objectives

- To determine the prevalence of Obstructive Sleep Apnea (OSA)
- Apply two validated questionnaires to the CHARGE Syndrome population
  - Brouillette Questionnaire
  - Pediatric Sleep Questionnaire
- Assess the quality of life after treatment for OSA

## Methods

- Subjects
  - Children ages 0-14, diagnosis CHARGE Syndrome
- Questionnaires
  - CHARGE Syndrome Characteristics
  - Brouillette Score
  - Pediatric Sleep Questionnaire
  - OSAS Quality of Life Survey2

Brouillette Score

Try it out!

**Questionnaire / Observation**  
**D. Difficulty in breathing during sleep?**  
 0=never; 1=occasionally; 2=frequently; and 3=always  
**A. Stops breathing during sleep?**  
 0=no; 1=yes  
**S. Snoring?**  
 0=never; 1=occasionally; 2=frequently; and 3=always

**Brouillette score** = 1.42 D + 1.41 A + 0.71 S - 3.83  
 >3.5: diagnostic for OSA  
 Between -1 and 3.5: suggestive for OSA  
 <-1: absence of OSA bb

## Results (N=51)

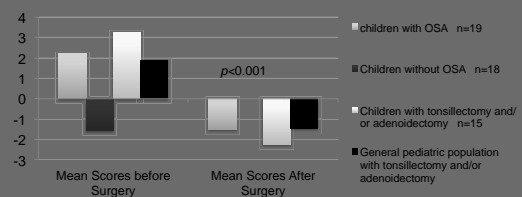
33 / 51 = 65% of children had obstructive sleep apnea (OSA)

- 10 treated with CPAP
- 27 adenoidectomy +/- tonsillectomy
- 9 tracheostomy

Brouillette Scores > 3.5 = OSA < -1 unlikely OSA

## Results (N=51) cont'd

Brouillette Scores for children before and after treatment for OSA



## Results (n = 16)

Pediatric Sleep Questionnaire Scores

Symptom Category Subscale	Mean scores before surgery	Mean scores after surgery	P Value
Snoring*	2.9	0.7	<0.001#
Breathing problems	1.8	0.6	<0.001#
Mouth breathing	1.3	1.0	0.104
Daytime sleepiness*	2.6	1.7	0.011#
Inattention/hyperactivity*	4.2	4.1	1.00
Other symptoms	1.6	1.6	0.333

\*Significantly associated with sleep related breathing disorders on their own  
 # Significant

Chervin RD, et al. Sleep Med 2000;1:21-32.

## Discussion / Conclusion

- Over 80% of the study population had a diagnosis of sleep apnea
- Brouillette scores identified obstructive sleep apnea in CHARGE Syndrome
- Pediatric Sleep Questionnaire – Sub Scale was useful in diagnosing sleep apnea
- The OSA -18 Questionnaire (Quality of Life) demonstrated all treatments for obstructive sleep apnea improved quality of life

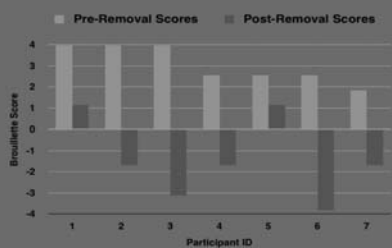
## Friends Chatting



## Tonsils and Adenoids

- How many of your children have had a tonsillectomy and/or adenoidectomy?
- How many of your children had anaesthetics after that?
- How many of your children had improved recovery after subsequent surgeries

## Removal of Tonsils & Adenoids (Preliminary Results)



Scores >3.5 highly predictive of OSA, between -1 to 3.5 suggestive for OSA, <-1 absence of OSA

## Summary – Sleep Apnea

- Obstructive sleep apnea is highly prevalent in the CHARGE Syndrome population
- Remove all tonsils and adenoids may be beneficial

## Take Home Messages

- Your children are at high risk of post-operative anaesthesia complications. Combining procedures during one anaesthesia does not increase the risk of post-operative airway events.
- The anaesthesiologist needs to be aware that, even with simple procedures, the individual with CHARGE syndrome is at high risk of post-operative events.

## Thank You From Four Friends



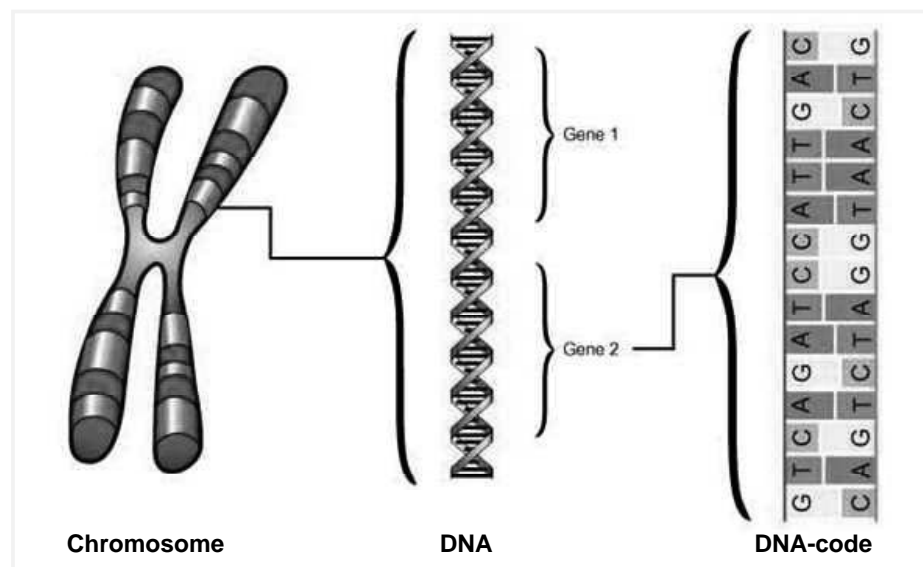
## All you want to know about CHARGE and genetics

By Nicole Corsten-Janssen and Conny van Ravenswaaij-Arts  
Department of Genetics, University Medical Center Groningen, The Netherlands  
July 2013

### What are chromosomes, genes, DNA and mutations?

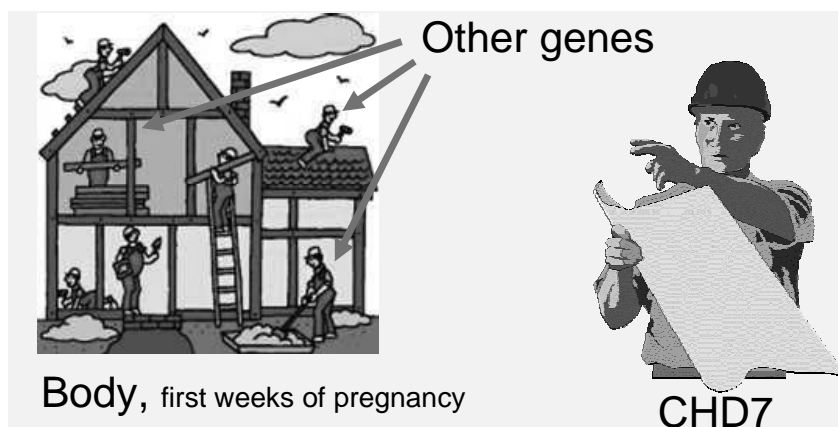
Our genetic information is tightly packed up on structures called chromosomes. Humans have 46 chromosomes grouped into 23 pairs. Everyone inherits 23 chromosomes from his father and 23 chromosomes from his mother. A chromosome consists of tightly packed up DNA.

DNA is our hereditary material and it is made up of four chemical bases (A,C,T,G). Genes are the pieces of DNA that hold the code for proteins, which are important for all kinds of functions in the body. A mutation is another word for a change in the DNA code, such a change in the DNA code can led to a change in the function of the proteins.



### What is the *CHD7* gene and what does it do?

Since 2004 we know that CHARGE syndrome is caused by a change (mutation) in the *CHD7* gene. Every person has two *CHD7* genes, one inherited from their father and the other from their mother. *CHD7* is a regulatory gene. It regulates the work of developmental genes very early on, during the development of the fetus. If there is insufficient *CHD7* protein being made, there is a higher risk of developmental defects occurring in specific organs like the heart, eye, ear, kidney, etc. A change in one of the two *CHD7* genes is enough to result in CHARGE syndrome. However, the syndrome is highly variable and it is not possible to predict the clinical consequences for the child from a specific change in *CHD7*.



*CHD7 regulates the function of genes influencing fetal development during early pregnancy.*

### How is CHD7 analysis done?

CHD7 analysis is performed on DNA. DNA is usually extracted from blood cells, but other tissues, e.g. skin or saliva, can also be used. Different types of mutations can be present in the *CHD7* gene. Most of these mutations will be detected by routine DNA analysis (called “sequencing”). Sometimes a part of the *CHD7* gene may be missing or even the whole gene (called a “deletion”). Deletions of *CHD7* are rare and occur in approximately 1% of CHARGE patients. They cannot be found by routine DNA analysis, but can be identified by other techniques (e.g. array, MLPA).

## Type of mutations

Normal gene

**Tim and his toy**

Point mutation

**Pim and his toy**

Deletion

**Tim and toy**

Insertion

**Tim and his big toy**

Frameshift

**Tim ndh ist oy**

### Why perform DNA analysis?

There are several reasons to perform DNA analysis:

- To provide certainty
- Confirm a tentative diagnosis in a child with atypical features (these two reasons have consequences for the clinical follow-up)
- Confirm the diagnosis, so that parents or siblings know their recurrence risk
- Prenatal options

### What if no *CHD7* mutation is found in my child?

- A diagnosis of CHARGE syndrome can be made by identifying a *CHD7* mutation, or by looking at the clinical criteria for the syndrome. If your child fulfills the clinical criteria, he/she has CHARGE syndrome, irrespective of the results of *CHD7* analysis.
- Some other syndromes have clinical features that overlap with CHARGE syndrome and it is important to exclude these syndromes from the diagnosis.

- Current techniques are not good enough to identify all *CHD7* mutations.
- Other genes might also contribute to CHARGE syndrome (see below).

### What is the risk that CHARGE syndrome will re-occur if we have another child?

Since familial CHARGE syndrome is extremely rare, the recurrence risk for parents who have a child with CHARGE syndrome is, in general, low. If parents want to learn more about their risk for future pregnancies, we recommend:

- A geneticist should see if the parents have any mild symptoms of CHARGE syndrome (hearing, balance, smell, shape of the ears).
- If a *CHD7* mutation has been found in a child, the parents can also be offered DNA analysis.

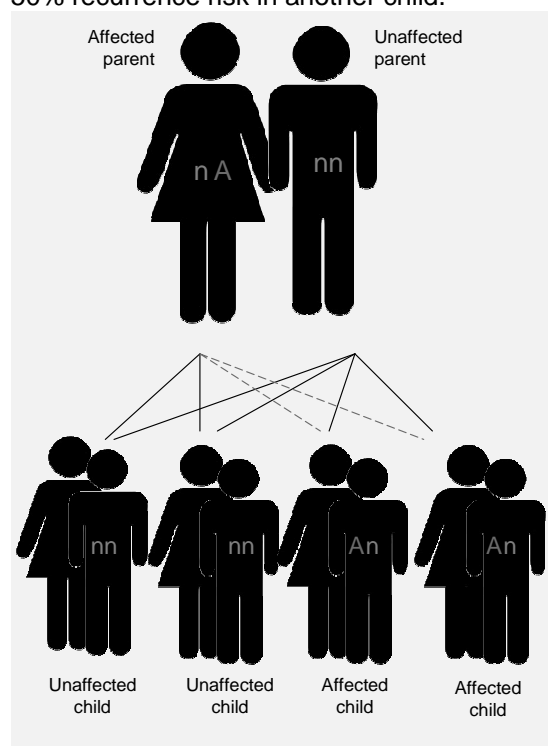
There are three possibilities:

#### 1. Parent does not have CHARGE syndrome or a *CHD7* mutation

If the *CHD7* change is not found in one of the parents (the most common situation), there is still a small risk of carrying a *CHD7* mutation ("germline mosaicism"), so the recurrence risk is not zero, but 1% or 2%.

#### 2. Parent has CHARGE syndrome

If one of the parents has CHARGE syndrome, the recurrence risk varies. Parents pass half of their genetic information on to their children. The parent with CHARGE syndrome can pass on either their normal *CHD7* gene or their gene with a mutation. This means that there is a 50% recurrence risk in another child.



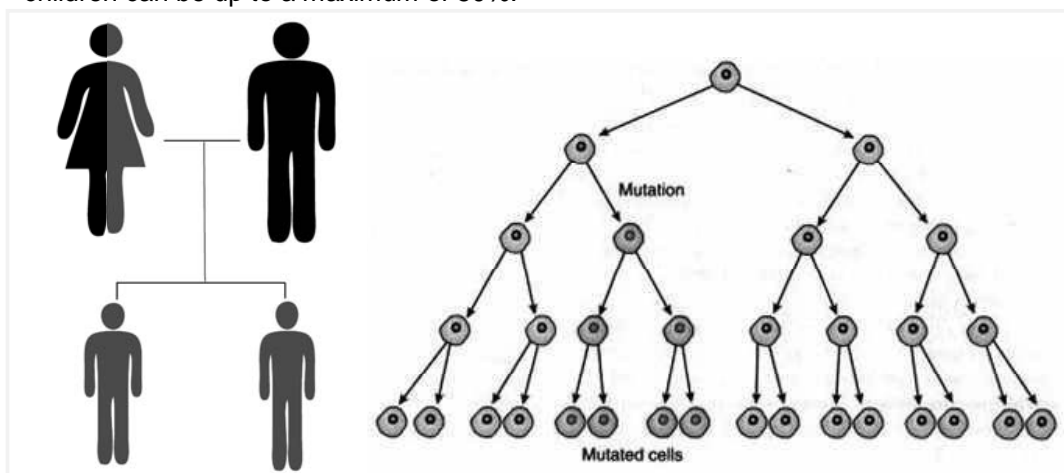
*The parent with mild CHARGE syndrome passes on the normal *CHD7* gene (n) or the abnormal *CHD7* gene (A). The recurrence risk is 50% for each pregnancy.*

#### 3. Parent with a *CHD7* mutation in some of their cells ("mosaicism")

Very rarely, a family may have two affected children with the same *CHD7* change, while the parents do not have any features of CHARGE syndrome. How can this happen? We may find that one of the parents carries a change in the *CHD7* gene in only some of his/her body cells. This is called mosaicism and it can occur when the fertilized egg has a normal *CHD7* gene, but a spontaneous change occurs later in one cell as the cells divide and grow (see

figure). Only the cells coming from that cell with the changed *CHD7* gene will have the mutation. If these cells are also present in the parent's ovaries or testes, egg or sperm cells with the *CHD7* mutation can be formed and lead to the pregnancy of a child with CHARGE syndrome.

If a parent has a mosaic form of the *CHD7* mutation, the recurrence risk for further children can be up to a maximum of 50%.



*In this example of mosaicism in the mother, there is a *CHD7* change (mutation) in some of her body cells. The scheme on the right shows that this can occur when the change (red cells) arises spontaneously during a cell division after fertilization. The mother will not have CHARGE syndrome herself, but she does have an increased risk of having affected children because she can pass on the *CHD7* change via an egg cell. The subsequent child will have the *CHD7* change in all its cells and will therefore have CHARGE syndrome.*

#### Options in a subsequent pregnancy

Parents who have a child with CHARGE syndrome in whom a *CHD7* mutation has been identified, may choose to have prenatal diagnosis performed for a subsequent pregnancy. But please remember that DNA analysis cannot predict the severity of CHARGE syndrome. A fetal ultrasound can provide extra information, like whether the baby has a heart defect, but ultrasound cannot “see” everything – deafness, developmental delay and behavioral problems cannot be detected.

The choice to have prenatal diagnosis is a personal one, and the geneticist must inform the parents well, so that they can make a choice they feel good about.

#### **Is *CHD7* the only gene causing CHARGE syndrome?**

In 5% to 10% of patients with typical CHARGE syndrome, no mutation in the *CHD7* gene can be found. This percentage is higher in patients who do not fulfill the clinical criteria for CHARGE syndrome (atypical presentation). So there may well be other genes that are also responsible for CHARGE or CHARGE-like syndrome. Mutations in a gene called *SEMA3E*, had been identified in two patients. A few other genes, like *CHD8* (a “candidate gene”), have been studied, but no mutations were found in these genes in CHARGE syndrome patients. Recently, we started to search for other genes that might cause CHARGE syndrome using a new technique, called next-generation sequencing or whole exome sequencing. This technique makes it possible to look for mutations in all the known genes in a single test.

**Any questions?** Please come and ask us during the CHARGE conference, or send an email to [n.corsten@umcg.nl](mailto:n.corsten@umcg.nl) or [c.m.a.van.ravenswaaij@umcg.nl](mailto:c.m.a.van.ravenswaaij@umcg.nl). More information on our research projects on CHARGE syndrome is available at [www.rug.nl/research/genetics/research/chargesyndrome/](http://www.rug.nl/research/genetics/research/chargesyndrome/)



## How do you „Follow the child“?

David Brown  
CHARGE conference  
Oberwesel  
Saturday, 05. July 2014

## The changing nature of the population of children with CHARGE syndrome

Have these changes caused problems for us?

- There is slower progress and/or regression
- There is less time for learning (because of illness, absence, or medical procedures)
- There is a need for more one-on-one support
- There is more physical care involved
- There are more agencies & different personnel involved
- Adaptations are needed to assessment procedures, teaching methods & communication systems
- There is a need for more careful preparation for attending & learning

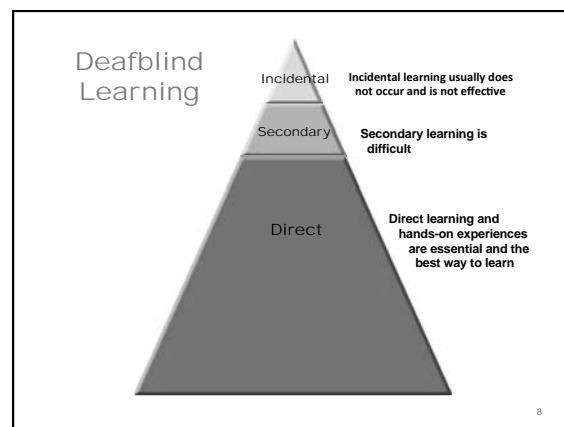
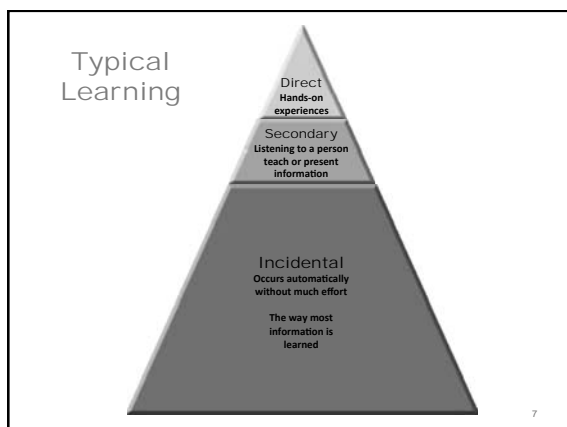
Have these changes offered us any advantages?

- Improved our observation skills
- Challenged our creativity
- Taxed our imaginations
- Increased our openness & honesty
- Encouraged our collaboration and sharing
- Increased our empathy
- Expanded the parameters of what we thought possible

## Tony Best (2003)

Deafblindness is a spectrum disability...but...the neurological involvement of the vast majority of deafblind people under the age of 10 makes it a medical condition as much as a sensory disability

- The eyes and the ears are the communication receivers above all others
- The senses of sight and hearing are the learning avenues above all others
- Distortion or absence of what is conveyed by those channels results in deprivation - emotional as well as intellectual
- Because of the dual impairment neither distance sense can compensate effectively for the impairment of the other



McInnes & Treffry "Deafblind Infants & Children(1982)

A child who is deaf-blind *may*:

- lack the ability to communicate with his or her environment in a meaningful way
- have a distorted perception of the world
- be deprived of the information necessary to anticipate future events or the results of his or her actions
- be deprived of many of the most basic extrinsic motivations

McInnes & Treffry (1982) cont.

- have medical problems that lead to serious developmental lags
- be mislabeled as developmentally disabled or emotionally disturbed
- be forced to develop unique learning styles to compensate for his or her sensory impairments
- have extreme difficulty in establishing and maintaining interpersonal relationships

*Personal conclusion after more than 30 years of scientific and practical work*

"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."

Jan Van Dijk (2001). My Own Evolution.  
<https://nationaldb.org/library/page/1962>

Why do people assess  
children with CHARGE  
syndrome?

### My view of assessment.....

- Is unusual!
- Is positive
- Looks at positive skills & achievements
- Looks at learning styles
- Looks at preferences & interests
- Looks at the whole child
- Credits the child with intelligence

### My view of assessment (2).....

- Seeks to improve my understanding of the child
- Seeks to help me to build a positive relationship with the child
- Seeks to help me to know what to teach and how best to teach it
- Seeks to give me a clear focus for measuring progress

### What might be going wrong with an assessment? (1)

- A focus on deficits rather than skills
- Ignoring the child's motivators
- The pacing is inappropriate for the child, their health, their alertness level, the time of day, the place, the activity, the people present, & the materials used
- The task is too complicated and too challenging
- Too serious - where's the play?
- The expectation that the child will 'Pass or Fail'

### What might be going wrong with an assessment? (2)

- Inappropriate assessment tools
- Mimicry of standardized clinical assessment procedures (timing, positioning, materials, expected outcomes)
- Lack of a whole-child perspective
- The assessor is wearing blinkers ("This is what I want to see, now!")
- The assessor is assessing the wrong things
- What is a response?

### Challenges to Assessment Process

- CHARGE presents a very diverse population of learners
- CHARGE presents a wide variety of idiosyncratic behaviors
- People doing assessments usually only know one type of assessment process
- There are limited resources and assessment tools available
- People doing the assessment often forget "The reason why" of assessment

### Are Standardized Tests Useful?

- Tests not standardized with this population are not valid
- Norming samples must include this population for standardized data to apply
- Conclusions regarding verbal tests must be made with caution
- Results of tests should be interpreted only in the light of observation of the child's performance in other tasks in other environments, with other assessors

- Most tests require an understanding of spoken language, the ability to see or hear materials, a means of responding either verbally or motorically, or all three
- Most tests require a good connection between the brain and the body
- loss of hearing & loss of vision both restrict the quality and quantity of experiences available to the child

If not the usual assessment ways...  
then what can we do?

- Return to “The reason why”
- Why do we assess children with CHARGE syndrome?
- What kind of information can help us to answer these questions?

“Assessing Communication and Learning in Young Children Who are Deafblind or Who Have Multiple Disabilities”

Edited by Charity Rowland, Ph.D.  
and published in 2009 by Design to Learn  
Projects of Oregon Health & Science University.

[http://www.ohsu.edu/oidd/d2l/com\\_pro/db\\_assess\\_ab.cfm](http://www.ohsu.edu/oidd/d2l/com_pro/db_assess_ab.cfm)

Natalie Barraga (1976)

Visual functioning is related in part to the condition of the eye. More explicitly, visual functioning is determined by the experiences, motivations, needs and expectations of each individual in relation to whatever visual capacity is available to satisfy curiosity and accomplish activities for personal satisfaction

Deafblind Education  
July-December 1995

“The natural way of learning the social rules needed in communication for a child is through play. Play is very much initiated and controlled by the child. It is characterised by shared affections, curiosity, and discovery through exploration”



Is this approach cheating?



Jan van Dijk (1966)

In the educational atmosphere I describe, the child holds the central position, the teacher 'follows' the child and, when the child responds, the teacher is present to answer the child's request

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?"

D Brown "Follow the child" reSources Vol 10 No 9 Winter 2001

Assessment Questions

D Brown "Follow the Child" (2001)

- How do you feel?
- What do you like?
- What do you want?
- What do you do?

D Brown 'Follow the Child' (2001)

- Consult those who know the child better than you do
- Identify the child's motivators
- It's okay to match different sensory inputs
- Relax or arouse the child as necessary
- Position the child to facilitate perception and functional skills
- Allow the TIME necessary for the loop of sensory perception, interpretation, and response

Paradoxes we should embrace (because they are unavoidable & because embracing them stimulates creativity)

- Clinical findings - or the child's functional behavior?
- 'Hands on' - or 'hands off'?
- Distraction-free environment - or the comfort of familiar 'distractions'?
- Should the child be energetic and alert - or maybe a bit tired is better?
- Our pacing and structure - or the child's?



[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

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The Van Dijk Approach - Evaluation challenges

- No prescribed protocol
- No specific implementation order
- No set of testing materials
- Each assessment is unique
- No set interpretation scale

The Van Dijk Approach - Quality indicators

- *Respecting* the caregiver
- *Respecting* the child
- *Following* the child’s lead
- *Communicating* with the child
- Utilizing *turn-taking* routines
- Creating of *enjoyable* routines

The Van Dijk Approach - Fidelity

- Utilization of stop-start within routines
- Introducing a mismatch with the child’s expectations
- Returning to established routines in order to examine memory
- Creating situations that allow for problem-solving
- Utilizing varying sensory channels

Robbie Blaha: *Thoughts on the Assessment of the Student with the Most Profound Disabilities* (1996)

- What range of states is exhibited?
- What are the most common states?
- Can the child reach the quiet alert or active alert states?
- Is the child able to maintain these?
- Are there problems shifting states?
- What variables affect state?

Blaha (1996) *Continued*

- Most effective sensory channels for gaining attention?
- Best sensory channels for conveying reliable information to the child?
- How much sensory information, delivered at what pace, helps the child to attend?
- What channels are associated with orienting reflexes?

### Blaha (1996) *Continued*

- What did the child used to notice but doesn't any more?
- Does he stop responding after 2-3 times?
- Does he seem to pair things, events, people together?
- Does he show anticipation? Surprise at changes in familiar routine?
- Does he know familiar from unfamiliar people?

### Blaha (1996) *Continued*

- What specific voluntary movements does the child exhibit (and in what positions)?
- How do different positions affect the child's level of arousal or bio-behavioral states?
- Does transition from one position to another cause significant change in the child's bio-behavioral state?

For any assessment it is important to ensure that results (including scores) are accurate and reflect the information the assessment was intended to collect, and are meaningfully linked to teaching practice

(Zatta & Pullin, 2004)

### Process not Product

- Trial and Error
- This is always a work in progress
- Questions not answers
- Why are you assessing the child?

Wiley D. (ed) 1997. IEP Quality Indicators for Students with deafblindness. TSBVI.  
[http://www.tsbvi.edu/attachments/1800 IEP Indicators.pdf](http://www.tsbvi.edu/attachments/1800%20IEP%20Indicators.pdf)

Brown, D. & Rodriguez-Gil, G. 2010. A Self-Evaluation Guide for Assessing the Quality of Your Interactions with a Student who is Deaf-blind. Fact Sheet #41.  
<http://www.cadbs.org/fact-sheets/>

Taylor E. Stremel K., Steele N. 2006. Classroom Observation Instrument.  
<https://nationaldb.org/library/page/534>




## Using a Person Centred Planning Approach



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

1




## What is Person Centred Planning (PCP)?

- A process of planning around the individual
- A tool to support effective planning for transition to adulthood
- A replacement for more traditional assessment approaches

© Sense 2013

2




## Why use PCP with a younger person?

- To gather information and plan an effective educational programme
- To support clear identification of strengths and needs
- To take a longer term view

© Sense 2013

3




## Why use PCP with a child or young person with CHARGE?

- It encourages a focus on the question:  
  
"What can be done to support greater independence, ownership of learning and self-determination?"

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4




## What information should PCP attempt to gain?

- To identify strengths and needs
- To identify their communication preferences and needs
- Use of vision, hearing and other senses and identification of support needed to enable optimal use of sensory information
- What the young person's likes/dislikes are at home and at school
- Things that can be achieved independently
- Things that help is needed with, and the type of help required
- Hopes for the future

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5



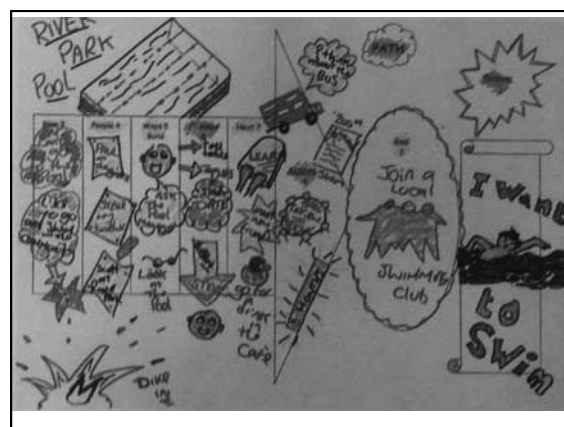
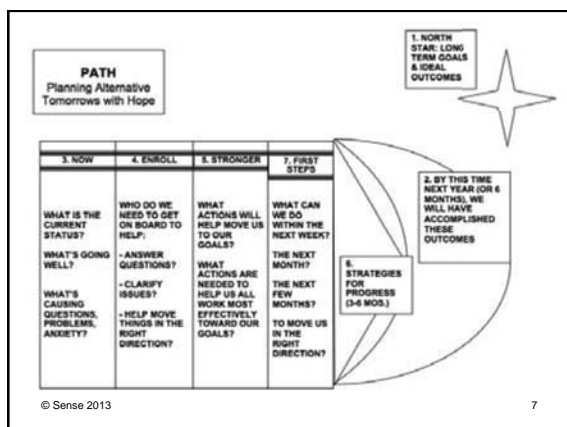
## How to obtain this information

A range of tools:

- Photographs
- Drawing
- Writing on large sheets of paper
- Talking and words being scribed
- Mind maps
- PATH

© Sense 2013

6



What is vital:

- That the child or young person remains at the centre
- It reflects what is important to the child/young person
- It reflects what they are capable of

© Sense 2013

Case study: Hannah

© Sense 2013

The Person Centred Meeting

- Who was to be invited to attend
- Hannah's contribution
- Areas to cover
- Tools
- Ground rules

© Sense 2013

Person centred meeting

The ground rules:

- We are all here for Hannah
- Everything is to be said positively
- Everyone has to take turns
- We are all equal and everyone's opinion matters and is valued
- Confidentiality is essential. What is said here is only shared with others appropriately with others when needed
- 5 minute rule
- Parking lot

© Sense 2013

**Hannah**

**Things people admire about me:**  
 I make people smile  
 My independent free spirit  
 How I can sort out muddles and helping  
 My sense of humour  
 Making my wants known  
 My giggle and cheekiness ☺

**People that are important to me:**  
 Mummy and Daddy  
 Tom and Alice  
 Karen  
 Nicki  
 Annette

**Places I like to go:**  
 Swimming pool  
 School  
 Going on the train  
 Legoland  
 Kid's Club

**Things I am good at:**  
 Gardening  
 Communicating things I have signs/gestures for  
 Trampolining  
 Swimming  
 Asserting myself  
 Shopping  
 Blowing out candles  
 Using the computer

**Things I find difficult:**  
 Change  
 Monday morning  
 Building changes at home  
 Noise  
 Calming down after a stimulating day  
 Cooperating  
 Everything at the moment!

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**What is important to Hannah now?**  
 -Photographs and concrete cues to let me know what is happening  
 -Routines and structure  
 -Consistent key people in her life at home and at school  
 -Being a part of the family  
 -Increasing my ability/opportunity to express myself  
 -Developing choice-making  
 -An effective behaviour plan and building in more positive praise  
 -Physical activity- it puts Hannah in a good mood & helps her sleep better  
 -To think: what is Hannah getting out of this? What is the purpose?

**What is working well:**

- ☺ Developing use of photographs to make a choice
- ☺ Physical activities and going on trips
- ☺ Hannah's persistence in communicating
- ☺ Coping well with the visual timetable that helps structure the day and lets Hannah know what is happening next
- ☺ School is a definite +
- ☺ Food- Likes eating!
- ☺ Music therapy

**What is not working so well:**

- ☹ Going for a walk at home- parents need a backpack like the one used at school for Hannah to use when going out
- ☹ Food- textures rather than taste seem to be important to Hannah
- ☹ Summer holidays, especially the last part of August.

**What is important for Hannah in the future?**

- ☺ To be able to communicate her wants and needs
- ☺ Developing life skills she can use in different settings
- ☺ To be able to cope in the community
- ☺ Increasing her independence
- ☺ Developing her self-advocacy


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**Future Dreams!**  
 A good quality of life and to live independently of her family  
 To have a good social life and friends

© Sense 2013

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Hannah's Year 9 Review

© Sense 2013

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My Year 9 Review

Hannah

What my friends like about me

My giggle when I'm happy

My determination

My independence

Following when someone signs to me

Listening to what is happening

Important people in my life

Family: Mum, Dad, Tom and Alice

School staff: Karen, Linda, Gill B, Gill C

Friends: Raphael

What's important to me now?  
What I like to do at ...

school

activities: sensory activities, drama, riding the tricycle, trampoline, swimming, big swing, music, computer, gardening

Hannah – my learning since my last review ...


- have had a lot of colds and a perforated eardrum (twice)
- a vertical visual timetable
- use photographs
- a qualified Intervenor who gives me greater opportunities
- sometimes I am happy to go out and about
- I'm not sure if I like walking long distances
  - I have been quite angry at times
  - swimming and horse-riding


What's important to me now?  
What I like to do at ...


home


activities: time with the family, watch TV, respite with Annette, nothing much really!


### What's important to me now?

I have food that I like to eat  That I feel healthy and well

I have my drink in a plastic beaker with a lid  That I have plenty of physical activity that supports my vestibular system

I need to wear my hearing-aids everyday  Please sign, use photographs and talk to me all the time

Being left with nothing to do and being ignored  Not being listened to when I am trying to tell you something

Being unexpectedly touched  Walking too far

### Working / not working



	
an Intervenor	being unwell
movement activities	not using my peak cap
photographs	not wearing my hearing aids
new classroom chair	food
going on outings	noise
music therapy	changes
	calming down




### Important to me now ...







- to be safe and comfortable and have my needs met
- to have someone to communicate with
- I like routine and to be cued into activities with speech, sign and photographs
- to stay at home with occasional visits to Castlegate
- to stay at school with continued one-to-one support
- to have a varied curriculum, appropriate activities and equipment
- to have opportunities for social activities
- I like to go to new places, to explore new environments but I don't always like to have to walk
- to be offered choices and do the things that I enjoy and interest me

### My targets ...

1. I need to choose from two objects, items or activities when I am in class.
 

	
--------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------
2. I need to be able to collect whatever I need to help me to do a task.
 

		
-------------------------------------------------------------------------------------	--------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------
3. I need to be able to put photographs in the right order to help me make a sandwich.
 

					
-------------------------------------------------------------------------------------	--------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------	---------------------------------------------------------------------------------------


### Finished!

Thank you for listening.

# Back to the Basics and Beyond

David Brown  
Tim Hartshorne

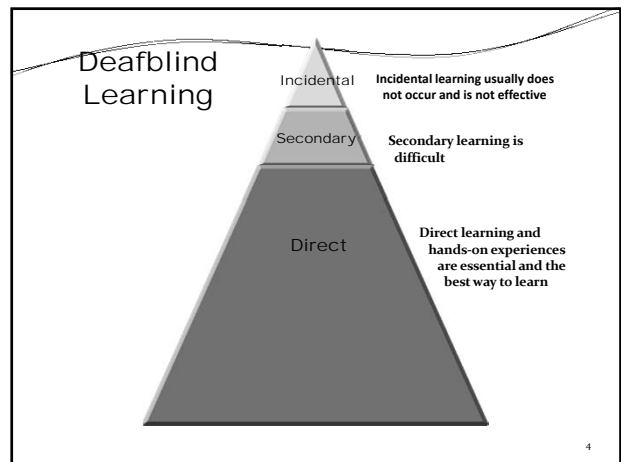
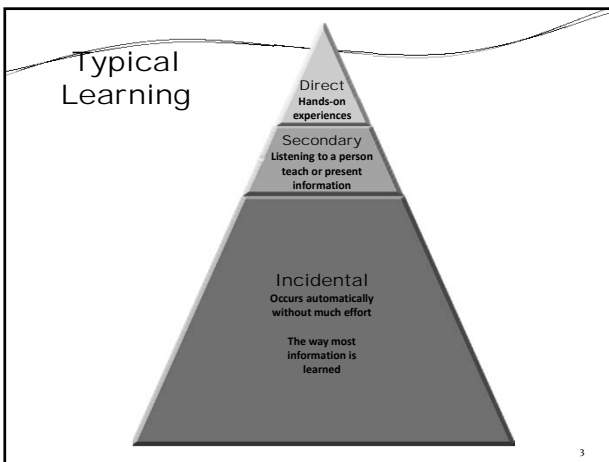
**CHARGE**  
CHARGE Syndrom e.V.



Personal conclusion after more than 30 years of scientific and practical work

**“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.”**

Jan Van Dijk (2001). My Own Evolution.  
<https://nationaldb.org/library/page/1962>



**McInnes & Treffry Deafblind Infants & Children 1982**

Communication can be summed up as our attempts to obtain information from and impose order upon the world around us

- \*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) Alert the child to the coming activity
- 3) Introduce the activity
- 4) Do, *and discuss*, the activity
- 5) On completion, review what you have done (eg. What is different now?)
- 6) Alert the child to your departure

## Communication Options

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

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## COMMUNICATION MODES AND THE SENSORY CHANNELS THROUGH WHICH THEY CAN BE PERCEIVED

✓ SPEECH	HEARING, VISION, TOUCH
✓ WRITING	VISION, TOUCH
✓ FINGER SPELLING	VISION, TOUCH
✓ MANUAL SIGNS	VISION, TOUCH
✓ NATURAL GESTURES	VISION, TOUCH
✓ PICTURES	VISION
✓ SYMBOLIC OBJECTS	VISION, TOUCH, HEARING, SMELL, TASTE
✓ SYMBOLS	VISION, TOUCH
✓ DRAWINGS	VISION, TOUCH

## The way to success?.....

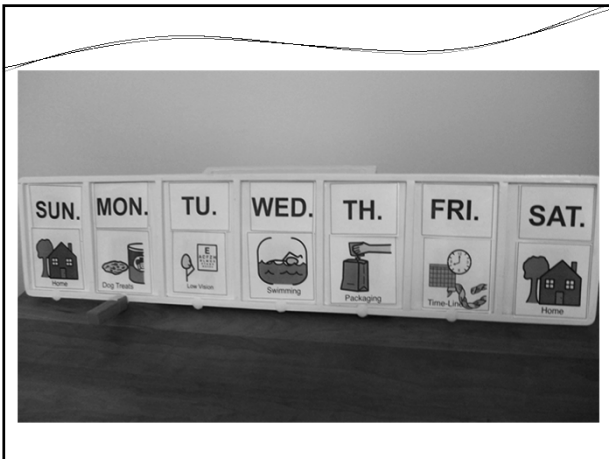
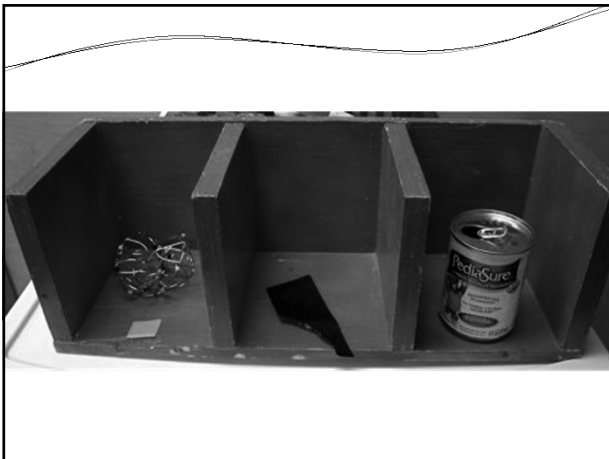
.....the child's preferred mode(s) of communication

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- + physical space – qualities of the room and activity area
- + positioning – where the student, instructor, and materials should be
- + materials – how teaching materials look, sound and feel
- + devices and equipment – adaptive aids used for sensory impairments
- + orientation & mobility – knowing where you are, and getting around
- + communication – getting information from, and giving information to others
- + trust and security – feeling supported, connected, and safe
- + literacy – recording information for future reference, and retrieving recorded information
- + pacing – how quickly the lesson should move forward
- + content of the activity or coursework – adding to, reducing, or changing what is taught.

- 10 Issues to Always Consider When Intervening for Students with Deafblindness
- By David Wiley, Texas Deafblind Outreach





## Personal Passports

Beginning at the beginning - a Personal Passport as an aid to enhancing realistic positive expectations of the individual, increasing the confidence & competence of the family, making assessment data effective and accessible, creating appropriate so successful programs, assisting with transitions, & recording progress.

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- A special way of sorting and recording information about an individual (not everything you know but what others need to know)
- An efficient way of presenting the information and making it accessible (simple, clear, direct, first-person, specific, honest, detailed, lively)

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- An empowering and positive way of presenting information - showing a person as human, individual, and recognizable
- A tool for helping an individual during transitions (meeting new people and starting at new places like nursery, school, hospital, holiday program, workplace)

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- A way of orientating new people quickly
- A tool for working towards consistency in how different people interpret, understand, and approach the individual
- A way of de-mystifying professional knowledge, and of helping people to observe more competently

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- A reminder that behavior is communication and that ALL people communicate
- An approach that gives a specific focus to discussions for parents, other family members, friends, and professionals to achieve a deeper understanding of an individual (and of each other)

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- A way of recognizing and valuing the roles of family and carers, and of giving them more influence

ADAPTED FROM CALL CENTRE  
INFORMATION SHEET 5 (JULY 1997)

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### Initial basic information

- Most important is the child's "likes and dislikes" (or their "want and don't want" behaviors) - and a description of how you know which of these opposite feelings the child is probably demonstrating. Consult with those who know the child best, and if it is not clear then at least have a 'best guess' which will be an important step on the journey to an effective assessment.

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### Teaching Strategies (Brown-2005)



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

## All behavior has a purpose

- Sensory Integration
- Self-Regulation
- Behavior as communication

## Sensory Integration

## The Senses

### Distance Senses

- Vision
- Hearing
- Smell

### Near Senses

- Taste
- Touch
- Vestibular
- Proprioception

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## SENSORY IMPAIRMENT

- Complete or partial loss
- Distortion
- Long processing time
- Variability/unreliability
- Knock-on effect
- Energy goes to adaptation

## Sensory Deficits in CHARGE

- Hearing – sensorineural hearing loss
- Vision – coloboma
- Smell – anosmia
- Taste – prefer strong tastes
- Tactile – defensiveness
- Vestibular – missing semi-circular canals
- Proprioceptive – body awareness

## Common Deafblind Behavior

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

We see all of these in children with CHARGE

## Sensory Issues

- Missing, partial, distorted, fragmented information
- Over-sensitivity or Under-sensitivity
- Processing time may be longer
- Confusion & the need for consistency & predictability
- Fatigue
- Communication issues (receptive & expressive)
- Movement and postural differences
- Idiosyncratic behaviors & misinterpretation
- Developmental delay

## Sensory systems have in common

- Conveying information from the environment to the brain for processing
- Each sense is designed to work simultaneously with all the other senses for maximum efficiency
- If other senses are not working properly or are missing, then the intact sense will become potentially more important but also more challenged
- Obstacles to effective processing can include:
  - malformation or damage of the sensory receptors
  - problems with the nerve pathways
  - brain malformations or brain damage

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We don't see with our eyes, or hear with our ears -  
we see and hear with our brains

- When you are assessing vision - don't think 'eyes', think 'child'
- When you are assessing hearing - don't think 'ears', think 'child'

## Natalie Barraga (1976)

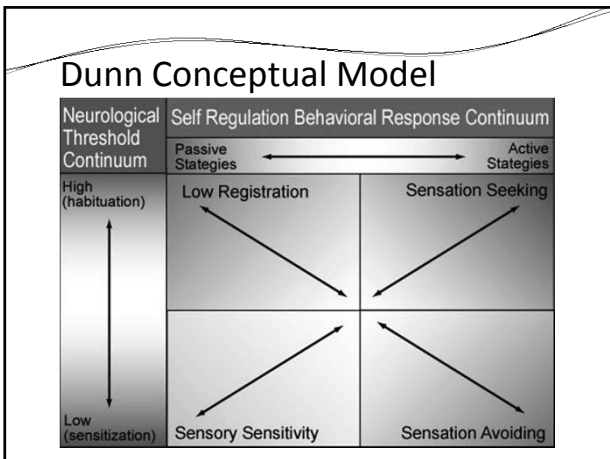
Sensory functioning is determined by the experiences, motivations, needs and expectations of each individual in relation to whatever capacity is available to satisfy curiosity and accomplish activities for personal satisfaction.

## When it fails to coordinate

- When information does not get into the brain properly
- When the brain fails to correctly interpret the information
- When the brain cannot sort out information from different sensory systems
- When the brain over-reacts or under-reacts to sensory input
- THERE IS A PROBLEM

## SENSORY INTEGRATION ISSUES

- Extreme sensitivity (or under reaction) to touch, movement, sights, or sounds
- Distractibility
- Social and/or emotional problems
- Activity level that is unusually high or unusually low
- Physical clumsiness or apparent carelessness
- Impulsivity, or lack of self-control
- Difficulty making transitions from one situation to another
- Inability to unwind or calm one's self
- Delays in speech, language, or motor skills
- Delays in academic achievement





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

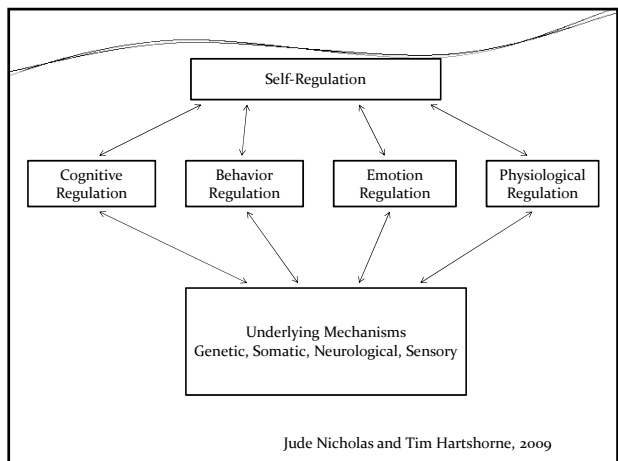
## Self-Regulation

### 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

### Two ends of the spectrum

- Totally regulated 
- Completely dysregulated 



## Cognitive Regulation

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



## 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

- 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

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



## 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.

## Development of Self-regulation

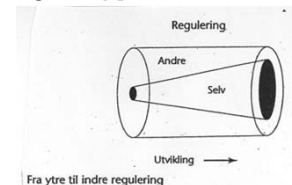
- Parents provide support
- Language allows child to talk or think about experience
- Peer relations provide experience and modeling
- All behavior has a purpose
- Learning to select and pursue goals

## Goals and Self-Regulation

- In order to pass the test I will make sure that I study (cognitive self-regulation)
- In order to get the job I will make sure that I am polite and seem interested (behavioral self-regulation)
- In order to stay friends I will not let my anger show but will talk with her (emotion self-regulation)
- In order to not feel overwhelmed by the crowd I will stay home (physiological self-regulation)

## Scaffolding for self-regulation

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



## 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



## 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-soothe
- Physiological
  - Squeeze technique
  - Hand on arm or leg
  - Brushing and joint compression

## Behavior as Communication

## ALL BEHAVIOR HAS A PURPOSE



## ALL BEHAVIOR IS COMMUNICATION



## Reading Behavior as Communication



## Justin: 9 year old with CHARGE

When Justin is touched he often hits.

- I hate surprises, therefore,...
- I don't like being touched,...
- I wanted to acknowledge your presence,...
- I like you very much,...
- I need more warning before you approach me,...
- I have been touched too much and I cannot tolerate it any more,...
- I was telling you that I'm ready for work,...
- I was asking you to come back later,...

## Many Communication Systems

- Direct behaviors
- Gestures
- Vocalizations and sounds
- Signs
- Speech
- Argumentative



## Communication Hypothesis

Behavior often functions as a primitive form of communication for those who do not yet possess or use more sophisticated forms.

- Does not mean the behavior necessarily has communicative intent
- Does not apply only to those who do not talk
- Does not imply that individuals systematically and intentionally use problem behavior to influence others.
- But we can understand and read the behavior of an individual as communication

## It's not easy!

- Learning to read behaviour can be challenging and takes TIME to get it right
- Recognising and interpreting subtle behavior requires keen powers of observation

## Managing Behaviors through Communication

- Identify the purpose of the behavior – what does it communicate?
- Let the person know that you understand the communication – they have to know that you know what they want
- Teach the person a different way to let you know what they want – word, sign, picture, object, expression
- Respect communication
  - It has to work before you can shape conditions

## Building Communication

- Bonding and attachment
  - Sometimes through touch
- Consistency of response and interaction
  - Including touch and object cues
  - Use of routine
  - Responding to all communication efforts
- Nonverbal conversations
  - Reciprocity
  - Imitating
  - Turn-taking
- Respecting behavioural state
- Acknowledging behaviour as communication

## What do I look for?

- Turning away
- Moving away
- Eyes looking away
- Loud repetitive vocalising
- Picking at skin or fingers
- Flicking of thumb
- Biting fingers
- Etc...

## What does it mean?

- Need for a break
- Fatigue
- Transition time
- Time to process
- Time to put it all together
- Need to regulate or reorganise self
- Sensory integration dysfunction

## Wait.....

- Give time to process
- Give time to self-regulate
- Give time for transition
- Give time for breaks
- Give time to put it all together
- Give time to refresh
  - Always consider fatigue

## Power of Communication

- How much control does a child with little formal communication have over his/her life?
  - What to do and when to do it
  - Where to go and when to go there
  - Time for therapy; time for medication
- Acknowledging behaviour as communication gives back some control

## Remember

- Behavior tells you something. It is a form of communication
- Children with CHARGE may struggle with their ability to self-regulate, and their behavior may be their attempt to manage
- Multi-sensory impairment and sensory integration problems can significantly impact the child's perception and understanding of the world.

## Finally

- Do not attempt to change a child's behavior until you understand what it means and why the child is engaging in it.
- Do not try to eliminate a behavior until you have an alternative to replace it with that will serve the same function.
- Endeavor to make sure that the child knows that you know what the child is communicating.
- Always respect communication

### Bone Health & Puberty – Not a Humorous Issue



Schonburg, Oberwesel, Germany, July 2014



Citadel, Halifax, Nova Scotia, Canada

Dr. Kim Blake, Professor, Dalhousie University  
Halifax, NS, Canada (kblake@dal.ca)

### Objectives

At the end of this presentation you will:

1. Have a better understanding of the risk factors that predispose individuals with CHARGE syndrome
2. Understand the prevention and treatment for good bone health.
3. Identify puberty issues related to your children.

### Osteoporosis Why do I Need to Worry?



Two friends with CHARGE Syndrome

CHARGE Syndrome from Birth to Adulthood: an individual reported on from 0 - 33 years.



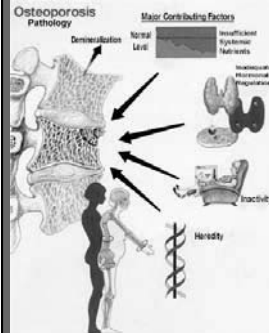
Searle et al American Journal of Medical Genetics  
2005;113A(3), 344-349.

### Adolescent and Adult Medical Issues



- Hormone replacement therapy (14-21 years)
- Thyroid replacement (19 years)
- Gallstones removed
- Reflux oesophagitis, stricture and hiatus hernia
- Osteoporosis

### What is Osteoporosis?



- Bone is a living tissue
- Calcium and Phosphate (CaPo<sub>4</sub>) [Mineral]
- Collagen [Protein]

Demineralization of bone and/or thinning of bone.

### Risk Factors for Osteoporosis in Individuals with CHARGE



Delayed/absent puberty.  
 Poor diet (low Ca<sup>2+</sup> & Vitamin D intake).  
 Inactivity  
 Growth hormone deficiency.

### Risk Factors for Poor Bone Health in Adolescents and Adults with CHARGE Syndrome

Karen E. Forward<sup>1</sup>,  
 Elizabeth A. Cummings<sup>2</sup>,  
 Kim D. Blake<sup>2,\*</sup>



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Great article to share with your Pediatrician or Endocrinologist

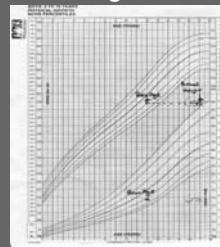
### Methodology

Parents/Caregivers of 45 individuals with CHARGE syndrome (ages 13+) were asked to complete questionnaires examining:

- Characteristics of CHARGE syndrome
- Bone Health
- Puberty
- Growth & Nutrition
- Habitual Activity Estimation Scale (HAES) (normative data has been gathered).

### Results: Bone Age

#### Actual Age 17 Years



#### L wrist & Hand X-ray



Bone Age: 92.3% (13/14) of individuals showed delays in bone age ranging from 2-8 years (assessed by L. wrist x-ray).

### Results : Spine and Fractures

Scoliosis (53.3%)



Kyphosis (16.7%)

Bony Fractures (30%)



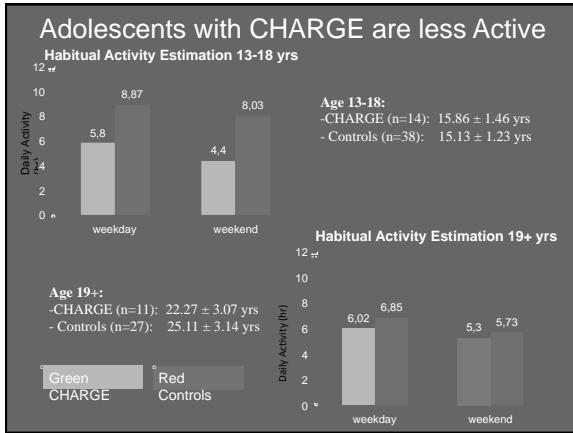
Scoliosis in CHARGE syndrome Doyle C, Blake KD,. AJMG. 133A: 340-343. 2005.

### Results: Nutrition Calcium and Vitamin D Intake is Not Adequate

Calcium:  
 50% of adolescents and adults failed to meet the Recommended Daily Allowance (RDA) for Calcium.

Vitamin D:  
 87% of adolescents and adults failed to meet the RDA for vitamin D.

53% of population used a gastrostomy tube. (mean age removed 8 +/- 6.5 yrs)



### Conclusion

- Approximately 50% of individuals with CHARGE Syndrome do not consume the Recommended Daily Allowed (RDA) for Calcium and most do not consume enough for Vitamin D.

### Puberty

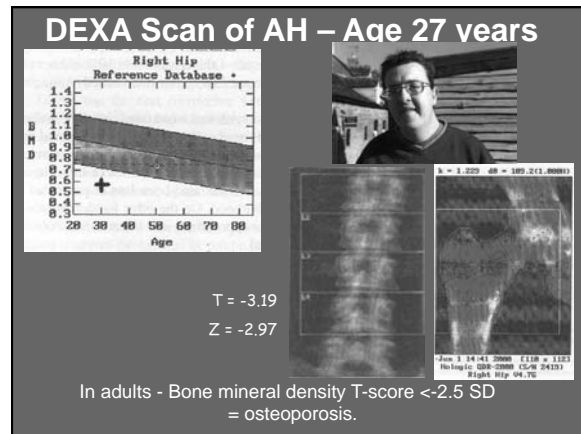
- Many CHARGE individuals do not undergo normal puberty (especially males).

### Activity:

- Teens ages 13 – 18 are significantly less active than controls, particularly during weekends (when there is less 1:1 support).

### Osteoporosis

- 25% of bone mass is laid down during puberty, in the 2 years either side of peak growth rate (average 12 years in girls, 14 years in boys).
- Every 10% increase in bone mass is equivalent to a 50% reduction in fracture rate.
- Dietary factors are also important: a low calcium intake < 500 mg/day reduces bone mass gain.



### To Measure Bone Density

#### Dual Energy X-ray Absorptiometry (DEXA or DXA)

Late 1980's postmenopausal women  
 1990's development of validation software

Different DEXA manufacturers, different modules, different software analysis = different numbers



### Osteoporosis - Prevention

Adequate Calcium in Diet  
 (from all sources diet and supplements)


Pre-pubertal (4-8 years) 800 mg/day  
 Adolescents (9-18 years) 1300 mg/day  
 Adults 1000 mg /day

### Osteoporosis - Prevention

- Adequate Vitamin D
- 800 IU (international Units)\*






Food rich in Vitamin D: sardines, herring, mackerel, salmon and fish oils (halibut and cod liver oils)




This may be an under estimate of vitamin D, especially in Northern climates

### Great for Balance but not for Bone Mineral Density (BMD)

### Exercises

- To increase BMD, exercise must be weight bearing
- Osteogenesis (bone accumulation) occurs under mechanical loading (*Madsen 1998*)
- Elite swimmers have no increase in lumbar spine BMD compared to sedentary individuals (*Bachrach 2000, Madsen Speckes 2001*)



### Prevention of Osteoporosis in CHARGE Syndrome

- Adequate diet and exercise\*
- Regular follow up with an endocrinologist for height, weight and pubertal status
- Sex Hormone replacement therapy
  - Testosterone in boys start at low dosage
  - Low dosage estrogens in females

\*Seek physiotherapy, recreational therapy


### Osteoporosis Treatment

- Recommended Daily Allowance of Calcium 1300 mg
- 800 IU Vitamin D
- Hormone replacement therapy

Bisphosphonates and raloxifene are the first line treatment in postmenopausal females... few studies in children  
Prevention is better!

### Puberty in CHARGE Syndrome

Pediatrician  
Endocrinologist



Family Doctor  
Internist  
Adult  
Endocrinologist

Delays and/or Divergent Puberty seen in >80 % of population.

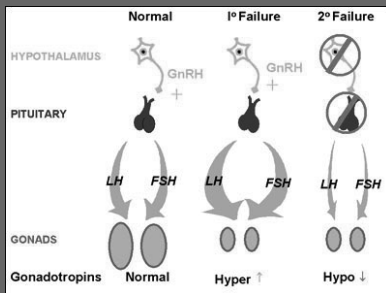
Puberty gets forgotten.

### Tom, 19 Year Old Male

- HCG test at 3 months, no testosterone
- 15 years, sparse pubic hair and axillary hair and perspiration
- 17 years, no real change
- 18 years, stimulation test, no puberty hormones or testosterone.
- 19 years, started testosterone and entered true puberty

### Pointers to puberty problems

- Anosmia (absent sense of smell) in both sexes.
- Micropenis (pinch an inch?) }
- Undescended testicles } in males.
- NB: Remember that pubic hair alone “adrenarche” may be due to sex hormones arising from the adrenal glands.

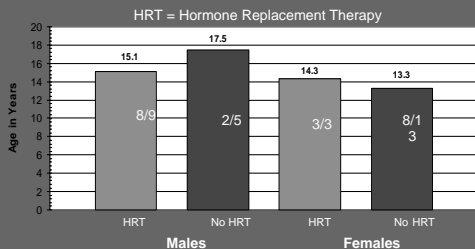


Forms of gonadal failure: central vs. peripheral

### Diagnosis of puberty problems

- Baseline “one-off” bloods (often not useful): LH, FSH, oestrogen (oestradiol) in girls, testosterone in boys.
- Stimulation tests:
  - LHRH tests “top end”: hypothalamus and pituitary.
  - hCG (in boys) tests “bottom end”: testicles.

CHARGE Syndrome: Average Age of Development of Secondary Sex Characteristics. (M = 14 F=16)



No secondary sexual characteristics 4 males (mean 14.2 years) 5 Females (mean 13.8 years).  
Questionnaire Completed by Families (Blake K, Abi Daoud M, 2000 CPS) CPSP 2003

### When to Investigate Delay in Puberty?

If puberty has not started by:

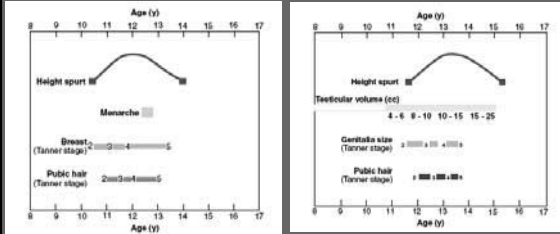
- 13 years in females.
- 14 years in males.
- Or if puberty has not been completed within 4-5 years.

#### Base-line Investigations

- Growth Chart
- Bone Age (left wrist X-Ray)
- Tanner Staging

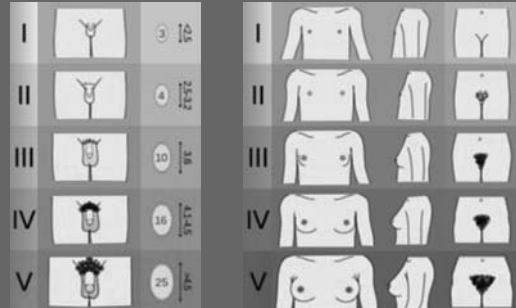


### Physical Maturity



Mention Constitutional Delay  
 Nelson's Essentials of Pediatrics, 5th ed. Philadelphia, 2006, Section XII "Adolescent Medicine"

### The 5 Stages of Tanner Development

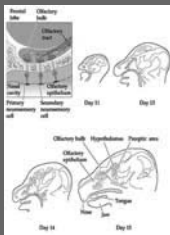


Nelson's Essentials of Pediatrics, 5th ed. Philadelphia, 2006, Section XII "Adolescent Medicine"

### Kallmann syndrome (KS)

An association of:

- Central hypogonadism (hypogonadotropic hypogonadism).
- Absent sense of smell (anosmia).
- Other clinical features eg. absent kidney, mirror movement (synkinesia) are also variably described.





Bergman JEH et al, Anosmia Predicts Hypogonadotropic Hypogonadism in CHARGE Syndrome. Journal of Pediatrics Vol 158, 2011

### Thanks! – Questions?



## Resilient Parenting

Tim Hartshorne, proud parent

## Everyone else seems competent

- AND
  - Happy with their child and family
  - Able to stand up to professionals and the schools
  - On top of the medical issues with a huge vocabulary
  - Able to still have a life



“Tonight I am so very tired of being the person figuring out what’s going on. It’s been awhile since I’ve felt so isolated, scared, on and on. Right now it’s as though nothing is enough to really help. I’m damned tired of this. I would like some help, too. I need it now. My daughter needs it now.”

### Parent experiences

- Parental Guilt: The Courage to be Imperfect

“How do you know if you have the right doctors and therapists doing the right things and enough of them? How do you know if you’re doing all the right things and enough of them? I have been avoiding phone calls and insurance stuff, etc, because I just want us to be normal.”

### Parent experiences

- Need to defend and protect the child

“Our first battle was over getting deafblind consultative services. We fought and fought. Now we are going through it again. Maybe it’s just the looming battle, but I feel like weeping. Yes, it can be necessary to fight like the devil, but that doesn’t make it right.”

### Parent experiences

- Capability of the child

“When she was an infant the hospital sent out evaluators. They sat me down and told me she would never walk, talk, or be independent. After he left I cried and had such terror my world spun. Then I thought “Baloney.” She ended up learning lots. Her performance always goes above and beyond potential. Always.”

### Parent Experiences

- Can I cope?

"Feel defeated today! Laura was diagnosed with oppositional defiant disorder awhile back but is getting more and more defiant on a daily basis which makes it very difficult for her special needs teacher to get any work done with her! I know there is stuff that she can do but refuses to and we can't revise her IEP based off of what Mommy knows! I get her need for control ... what she does is really the only thing she has control over as we take her to Doctor after Doctor! I get home and am explaining to my husband that I am frustrated and just don't know what to do ... I feel like as a parent I am losing the ability to discipline my child properly because she is constantly acting out ... As I am telling him all of this my phone rings and it's the Pulmonary and sleep Doctor she tells me she is concerned about Laura's Ferritin results."

### The View in 1973

The impact of a handicapped child on a family is never negligible, usually damaging, and sometimes catastrophic. A few families with great spiritual strength may be bound more firmly together by the experience, but in most, the stresses imposed far outweigh any benefit. Mitchell, 1973.

### Negatives?

- I don't know enough to do this
- There is no support out there
- Someone will need to stay home
- Getting the right medical help
- Fear for my child
- Guilt that I am not doing enough
- I may spend the rest of my life doing this
- There is not enough time in the day
- There is not enough time for spouse and other children

I can't cope!!!

### The View Today

Families of children with disabilities report positive perceptions in addition to negative perceptions and stress, and there is some data to suggest that positive perceptions are common, and that they assist the family in adapting to having a child with disabilities.

Hastings & Taunt, 2002

### Positives?

- Pleasure/Satisfaction in providing care
- Child is a joy
- Sense of accomplishment
- Strengthened family
- New sense of purpose
- New skills, abilities, even career
- Have become a better person
- Increased personal strength/confidence
- Social networks
- Increased spirituality
- Changed perspective on life
- Making the most of every day

### COURAGE IS NOT DENIAL

**BUT SOMETIMES COURAGE LOOKS LIKE DENIAL TO OTHER PEOPLE**

Hartshorne, T. S. (2002). Mistaking courage for denial: Family resilience after the birth of a child with severe disabilities. *Journal of Individual Psychology, 58*, 263-278.

A SHORT FORM OF THE QUESTIONNAIRE ON  
RESOURCES AND STRESS  
Friedrich, Greenberg, & Crnic (1983)

T\_ 2. Other members of the family have to do without things because of \_\_\_\_.

T\_ 6. \_\_\_\_ is limited in the kind of work he/she can do to make a living.

F\_ 7. I have accepted the fact that \_\_\_\_ might have to live out his/her life in some special setting (e.g. institution or group home).

T\_ 13. It bothers me that \_\_\_\_ will always be this way.

F\_ 18. The family does as many things together now as we ever did.

T\_ 22. \_\_\_\_ doesn't do as much as he/she should be able to do.

T\_ 28. I am disappointed that \_\_\_\_ does not lead a normal life.

T\_ 32. I worry about what will be done with \_\_\_\_ when he/she gets older.

F\_ 38. \_\_\_\_ can ride a bus.

The problem is how to love your child with no need for your child to be any different, AND without giving up hope that your child may progress with time and resources.

## Mothers

- Most research is on mothers
- Burden of care giving may fall on mothers most of all
- Parental roles may be more well-defined
- Mothers may experience the most guilt

## Fathers

- "scared, concerned, anxious, fearful" (30%)
- May not feel they contribute enough
- Find work to be slightly less central to their identity
- Find male-male friendships to be less important
- Slightly less happy and satisfied with life

## Siblings

- Somewhat lonely
- Positive evaluation of having sibling
- Somewhat knowledgeable about CHARGE
- Believe parents to have moderately high levels of stress, moderately due to CHARGE
- Tended to avoid adding to parent stress
- See selves as pretty healthy overall

## Global Meaning

- Enduring beliefs
  - About the world
  - About the self
  - About the self in the world
- Valued goals
  - Purpose in life
- Understanding of past and present
- Expectations regarding the future
- That which is supposed to provide meaning in my life



## Situational Meaning

- When situations happen, we draw on global meaning as we decide what to do
- We try to find meaning in situations that is consistent with our global meaning
- When that fails we may modify the situational meaning
- Ultimately we may shift our global meaning

## The desire for offspring

- My life will only be fulfilled if I have biological offspring
- ["normal" is often implied]
- Infertility creates a situation that challenges global meaning
- Can be resolved by thinking of the situation differently – biology is not everything
- Or resolved by changing the global meaning – Love and family are more important than offspring
- Or maybe I can mentor other people's offspring

## A child with CHARGE is a huge challenge to global meaning

- For most of us
- "I did not sign up for this."
- Even when managing the situation well (modifying situational meaning – "I have learned to appreciate the small things")
- This challenges the intended source of meaning in life – a healthy child
- "It took me a long time to realize I did not need her to be any different."



Viktor Frankl

"Man's search for meaning"

## Life is Learning to Cope with Loss

How much can be taken away before we lose the meaning in our lives?



## Sources of meaning in life

- Love, or experiencing someone deeply
- Doing a deed; accomplishing something
- Unavoidable suffering – suffering can have meaning



### A CHARGEd life

- Love – we are here today because we love our children with CHARGE and what they have taught us about love



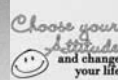
### A CHARGEd Life

- Doing a deed – we have learned so much and accomplished and achieved so much in our raising of our child.



### A CHARGEd Life

- Unavoidable suffering – YES, it is really hard to parent a child with CHARGE. You have given up much, you have sacrificed much, you have cried many tears. But you would do it all over again.
- If you doubt your own strength, look at your child.



### Resilience

- Attitude and shifts in global meaning
  - **Choosing** a social context
  - **Choosing** to bond
  - **Choosing** to learn and gain knowledge
  - **Choosing** one's experiences
- Resilient parents find a positive social support group, come to know their child in a way that creates a bond, learn as much as they can about their child's condition, and quickly acquire as much experience as they can in meeting the needs of their child.

### Resources: resilient parents find the resources they need

- General and specific beliefs
- Problem-solving skills
- Health/Energy/Morale
- Material resources
- Social support
  - Network orientation
  - Seek and find

### SOCIAL SUPPORT

#### POSITIVE AND NEGATIVE EFFECTS

- |                                |                                |
|--------------------------------|--------------------------------|
| □ Reduce worry and uncertainty | □ Create worry and uncertainty |
| □ Set good example             | □ Set bad example              |
| □ Share problems               | □ Create problems              |
| □ Label beneficial             | □ Label negative               |
| □ Give helpful information     | □ Give misleading information  |
| □ Foster independence          | □ Foster dependence            |

## Network Orientation Scale

1. Sometimes it is necessary to talk to someone about your problems.

1 2 3 4

2. Friends often have good advice to give.

1 2 3 4

3. You have to be careful who you tell personal things to.

1 2 3 4

4. I often get useful information from other people.

1 2 3 4

5. People should keep their problems to themselves.

## Two Useful Perspectives

- The courage to be imperfect
- A psychology of use and not possession



## Post-Traumatic Growth

- New coping skills and resources
- Relating to others
- New possibilities
- Personal strength
- Spiritual change
- Appreciation of life
- Positive affect



## Parenting

*It is hard enough to cope with raising a child who does not have obvious disabilities. Why in the world would you think you should know how to raise a child with tremendous needs?*

## Especially the behaviors!

*"I've heard Tim talk about behavior multiple times and I still don't know how to cope with it."*



## Good-Enough Parenting

- Make your parenting errors with confidence
- Recognize there is no manual
- Trust your instincts, but be mindful of results
- Choose your battles wisely
- Be consistent when it matters
- Give as much freedom of choice as possible
- Know that children who feel good, do good
- Understand that misbehaviors may be creative solutions on the part of your child

## Forgive Yourself

- For not being the parent your child deserved
- None of us gets the parents we deserved
- None of us is perfect



## Pat yourself on the back

- You are resilient
- You are present
- You are making a tremendous difference in your child's life



Perfect courage is the courage to be imperfect.



## MARATHON SKILLS

Ann P. Turnbull

- Meet basic needs
- Know your self and your family
- Love your child unconditionally
- Establish relationships
- Experience and benefit from emotions
- Take charge
- Anticipate future/transitional planning
- Establish balance



## Contact information

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