Understanding Behavioral Issues in CHARGE Syndrome

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A CHARGE Behavioral Phenotype (first draft)

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

- Autism
- ADHD
- OCD
- Tourette
- Etc.
Problems with these Diagnoses

- Create a reality that may not exist
- Lack explanatory value
- Ignore the uniqueness of the behavior
- Lead to multiple drug treatments
The Big Three

- Pain
- Self-Regulation
- Sensory Issues
- Anxiety
Possible sources of pain

- Surgery
- Procedures
- Doctor visits

Syndrome “related” pain
Sources of Pain in CHARGE

- Otitis Media\(^1\)
- Sinus infections
- Migraines\(^1\)
- Abdominal Migraines\(^1,2\)
- Gastroesophageal reflux disease (GERD)\(^2\)
- Gas
- Constipation\(^1,2\)
- Muscle pain\(^2\)
- Tactile defensiveness
- Stoma pain
- Oral pain/teeth
- Surgery\(^1\)
Hearing – sensorineural hearing loss
Vision – coloboma
Smell – anosmia
Taste – prefer strong tastes
Tactile – defensiveness
Vestibular – balance issues
Proprioceptive – awkwardness
This is normal viewing posture...

...when you have no vestibular sense, upper visual field loss, poor tactile & proprioceptive perception, & low muscle tone.
What could our kids possibly have to be anxious about?
1. PAIN
It doesn’t often come with a big sign
2. Sensory overload or underload issues

Wanted to build a castle.

Had to touch sand.
3. Stress

- Life is chaotic.
- I cannot hear/see/balance/predict what’s going to happen next.
- I want to escape the chaos.
We like to know what is going on.

- What are we doing right now?
- What are we going to do next?
- What did we just do?