Managing the Complexities of Cochlear Implantation in Children with CHARGE Syndrome: Case Study

chrisanda Sanchez, AuD
University of Miami, Department of Otolaryngology
Children’s Hearing Program

CI2019 Miami
Disclosures

- Employed by the University of Miami
CHARGE Syndrome

- Disorder affecting several areas of the body
- Cluster diagnosis:
  - Colobomas
  - Heart Defects
  - Atresia of the Choanae
  - Retardation of Growth
  - Genital Abnormalities
  - Ear Anomalies

How is it diagnosed?

- Clinical diagnosis, with having at least two of the major findings.
- CHD7 genetic testing is recommended for all patients who have suspected CHARGE syndrome

What are the challenges?

- Features overlap with other syndromes:
  - 22q11.2 deletion syndrome, Kabuki syndrome, & VACTERL
- The characteristics can vary from severe to none; there is not one specific feature found on every single child with CHARGE
- CHD7 testing is not perfect; 10-20% of people with CHARGE do not have an identifiable pathogenic CHD7 variant
Meet J

- 2 year old male
- Born full-term in Jamaica
- Admitted to the NICU in the US shortly after birth

**Medical History:**
- Vision loss: *uses glasses*
- Respiratory problems: *tracheostomy tube*
- Heart & GI problems: *has G tube*
- Facial nerve weakness: *2/6; right sided*
- Global delays: *has 24-hour nurse care*
- Genetics: *heterozygote for truncating mutation in CHD7*
Established care with UM Children’s Hearing Program at 6 weeks old

- **Severe to profound SNHL bilaterally, via two diagnostic ABRs,** negative for ANSD
- Subsequently fit with bilateral amplification (UP hearing aids) at 10 weeks old

**Otoscopy:** Low set, cupped ears; **bilateral microtia**

**Tympanometry:** Chronic ME pathology

**Audiometry:** Severe to profound SNHL via BO/VRA
Otology History

Imaging:
- Possible CN deficiency but still present; R < L
- Absent SCC bilaterally
- Cochlea appear OK, large vestibulae
To Implant or Not To Implant?

Extensive counseling regarding realistic expectations

- Not only due to imaging, but regarding the dual sensory impairment, and the diagnosis of CHARGE as more of a *spectrum; we cannot predict outcomes!*

**Parental acceptance: realistic expectations**

- Mother is heavily involved in a parental support group for parents of children with CHARGE
- Attends sensory days at local Miami Children’s Museum
- Receiving services from Early Steps, Auditory Verbal Therapy, trialing AAC devices, and getting resources from Lighthouse for the Blind
- Now serves as a mentor for other families with children who have CHARGE syndrome
**Left Cochlear Implant**

15 **months**: underwent left ear first, better anatomical findings

Implanted with: *Advanced Bionics HiRes Ultra Mid-Scala electrode*
- Full insertion was achieved via independent cochleostomy

**Intraoperative NRI revealed no measurable response up to 600 CU**
CI Programming

**Subjective Methods:**
- Visual Reinforcement Audiometry
- Behavioral Observation

**Objective Methods:**
- Impedance Check
- Evoked Cortical Action Potential (AB – NRI)
- Electrical Stapedial Reflex Threshold (ESRT)

**Creative Methods:**
- Use assists
- Use therapists
- Use parents
- Network with multi-disciplinary team
- Don’t give up!
About Not Giving Up…

**Intraoperative NRI revealed no measurable response up to 600 CU**

**Neural Response Telemetry:**

- ECAP is a synchronous response from electrically stimulated auditory nerve fibers and is essentially the electrical version of the Wave I of an ABR.

- Advanced Bionics' NRI is built with an algorithm that marks negative and positive peaks of the ECAP, calculates the peak-to-peak amplitude difference, and then plots that difference as a function of current level.

- Continuous attempts at running NRI up to the same levels (400-450CU) began to yield responses which were previously absent.
Real Responses?

- Assessment of benefit is going to vary from child to child
- Quality of life improvements are the strongest motivating factors
- Progress is going to be slow, **patience is a must**
- There is no checklist that can be made for children with CHARGE
  - Each child has their own needs that are unique to them
- Don’t give up!
Where are we now?

Bilaterally implanted
Beginning to walk unassisted
Using multiple modalities to communicate

Remember! When you’ve met one patient with CHARGE, you have simply met one patient with CHARGE syndrome.

Each and every child is unique and special in their own way.
Come back and see us in Miami, Florida in December!

7th International Conference on Bone Conduction and Related Technologies

cmsanchez@med.miami.edu