Outcomes of Cochlear Implantation in a Child with Zellweger Syndrome

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Objective

The aim of this case report is to present audiological management and results of unilateral cochlear implantation of a 15-year-old child with Zellweger syndrome (ZS).
In cases of severe to profound sensorineural hearing loss (SNHL) and ZS, a cochlear implant (CI) may be a possible treatment option.

Study Design

This case report describes a 15-year-old male with Zellweger syndrome and profound SNHL. He received a unilateral cochlear implant by age 15 years after progressive hearing loss and increased limitation in benefit from hearing aids. Audiological characteristics and parent-reported benefits via IT-MAIS outcome measure are discussed in the report. (1)

Background

Zellweger syndrome is a rare peroxisome biogenesis disorder (PBD). The major clinical features include:

- Hypotonia
- Feeding problems
- Seizures
- Developmental delay
- Adrenocortical dysfunction
- Issues with organ function
- Other neurological abnormalities
- Vision loss
- Hearing loss

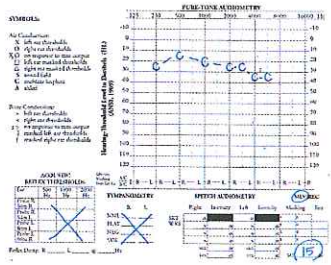
The syndrome is a spectrum disorder as part of a heterogeneous group of autosomal recessive disorders. These disorders include Zellweger syndrome (severe), neonatal adrenoleukodystrophy (intermediate), and infantile Refsum disease (mild). ZS is caused by defects in peroxisome formation and mutations in 1 of 13 PEX genes. (2) There is extreme variability in disorder manifestation. The frequency has been estimated to be 1 in 50,000 births in North America. (3)

Outcomes with CI in ZS are not well reported in the literature. Cochlear implants have been effectively placed in children with ZS when hearing aids provide limited audibility. It is of note that improvements in environmental awareness, and in some circumstances, speech, have been frequently noted in other syndromes with congenital deafness. (4)

Hearing / CI Results

- Passed NBHS, bilaterally.
- Diagnostic ABR at 2 years of age revealed bilateral severe SNHL.
- Fit with binaural hearing aids - increased responsiveness and audibility.
- Hearing loss progressed until diagnostic ABR at 15 years of age revealed profound SNHL.
- Note: Diagnosis of retinitis pigmentosa and optic nerve atrophy.
- CI surgery: 15 years of age
  - Full insertion with Advanced Bionics HiRes 90K MidScala internal device
  - Optima S processing strategy
  - Neptune processor
- Difficult to test behaviorally due to developmental delay and vision loss.
- Reliable CI audiograms obtained using Visual Reinforcement Audiometry in a dark sound booth.

Figure 1. Audiogram age 15 years (6 months post CI)



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Speech / Language Results

Infant-Toddler Meaningful Auditory Integration Scale (IT-MAIS)

Pre-implantation:

- Total Score: 17/40

9 months Post-implantation:

- Total Score: 34/40

Parent Report (Pre- to Post-CI)

1. Increased vocal behavior. "Constantly playing with sound and/or listening."
2. Increased communication. "New words, clearer speech, increased signing."
3. Increased responsiveness. "Responds appropriately and consistently to name called / being spoken to."
4. Increased responsiveness in noise. From Never to Frequently.
5. Increased alertness to environmental sounds. "Always turning and listening."
6. Increased alertness to new sounds. "Searches for source of sounds."
7. Increased recognition of auditory signals. From Never to Always.
8. Similar performance in voice discrimination.
9. Increased speech vs non-speech discrimination. "Much better at understanding the source of sound."
10. Increased vocal tone association. "Better at identifying emotion being conveyed through sound."

Other Parent-Reported Benefits

- Decreased anxiety
- Significant progress in communication methods (total communication through verbal language, hand under hand signing, and object cards on calendar system)
- Overall improved quality of life

Conclusions

Cochlear implantation is an appropriate option for treatment of profound SNHL in children with ZS. It is still unclear how successful he or others with ZS will be with developing spoken language. Cochlear implantation can provide good access to audibility of spoken language for children with ZS and profound SNHL. Early identification and treatment of ZS and the subsequent hearing loss is of great importance to rehabilitate the patient to better meet communication needs.

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