Cochlear Implantation in Siblings with ANSD
A Case Series and Review of the Literature

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Background

- Auditory neuropathy spectrum disorder (ANSD) is a hearing disorder characterized by absent or severely abnormal auditory brainstem responses (ABRs) and intact cochlear outer hair cell function
- Cochlear implantation (CI) has become the standard of care for patients with severe to profound hearing loss, however, its use in ANSD is considered controversial due to variable outcomes
- Here, we present a current review of the literature to evaluate the effect of CI on hearing sensitivity and speech perception among patients with syndromic and non-syndromic ANSD
- Additionally, we present a case series of three sets of siblings with non-syndromic ANSD who received CI and have long-term pure tone audiometry and speech perception scores available

Case Series

- Patient 1A:
  - Diagnosed with auditory neuropathy with mild SNHL and speech delay at 3-years-old
  - Right-sided Advanced Bionic CI at 9-years-old
  - Postoperatively, demonstrated good outcome, enjoyed implant, and hearing loss remained constant with slow progression
  - Contralateral left-sided Advanced Bionic CI at 14-years-old for severe SNHL
  - To date, demonstrates improved hearing thresholds by pure tone audiometry (PTA) at 15 years since first CI and improved speech perception with **AzBio scores of 98% and 100%** at 12 years since CI; still reports some difficulty understanding rapid speech in conversation
- Patient 1B (younger half-brother):
  - Diagnosed with auditory neuropathy with bilateral SNHL at 33-months
  - Left-sided Advanced Bionic CI at 3-years-old
  - Postoperatively, demonstrated good outcome
  - Contralateral left-sided Advanced Bionics CI at 6-years-old
  - To date, demonstrates improved hearing thresholds by PTA at 15 years since first CI, but continues to use ASL alongside speech for communication and follows an IEP in high school
- Patient 2A:
  - Diagnosed with auditory neuropathy with bilateral SNHL at 33-months-old
  - Left-sided Advanced Bionic CI at 3-years-old and right-sided HA
  - Decline in right-ear hearing thresholds from moderate to the severe-to-profound range, with minimal benefit provided by his HA at 7-years-old; elected to continue with right-sided HA
  - To date, demonstrates improved hearing thresholds by PTA at 9 years since CI, but continues to receive speech services and use ASL, with **HINT scores demonstrating 83% accuracy in the left-ear**
- Patient 2B (younger sister):
  - Diagnosed with bilateral auditory neuropathy at 2-months-old
  - OtoSCOPE genetic hearing loss testing revealed **homozygous MYO15A mutations**
  - Left-sided Advanced Bionic CI at 3-years-old
  - To date, demonstrates improved hearing thresholds by PTA at 6 years since CI, and continues to receive speech services and use ASL, with **HINT scores demonstrating 95% accuracy in the left-ear**
- Patient 2C (youngest sister):
  - Diagnosed with bilateral auditory neuropathy at 2-months-old
  - Left-sided Advanced Bionic CI at 2-years-old
  - To date, demonstrates improved hearing thresholds by PTA at 3 years since CI, and continues to receive speech services, primarily uses ASL, has demonstrated decreased **IT-MAIS scores of 4/40 from 13/40 preoperatively;** continues to exhibit decreased language comprehension and expression on the Rosetti Infant-Toddler Language Scale

Results

Fig 1. Effect of CI on Pure Tone Averages in Syndromic & Non-Syndromic ANSD

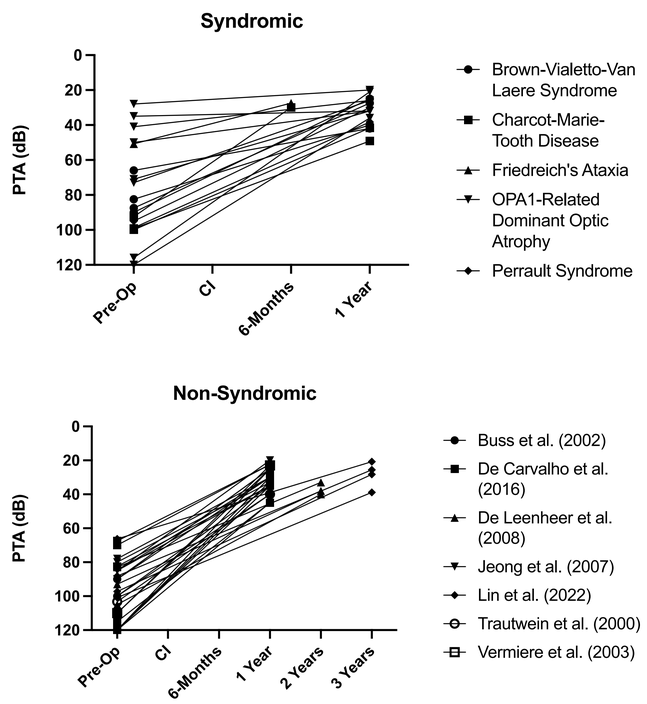


Fig 2. Effect of CI on Speech Perception in Syndromic & Non-Syndromic ANSD

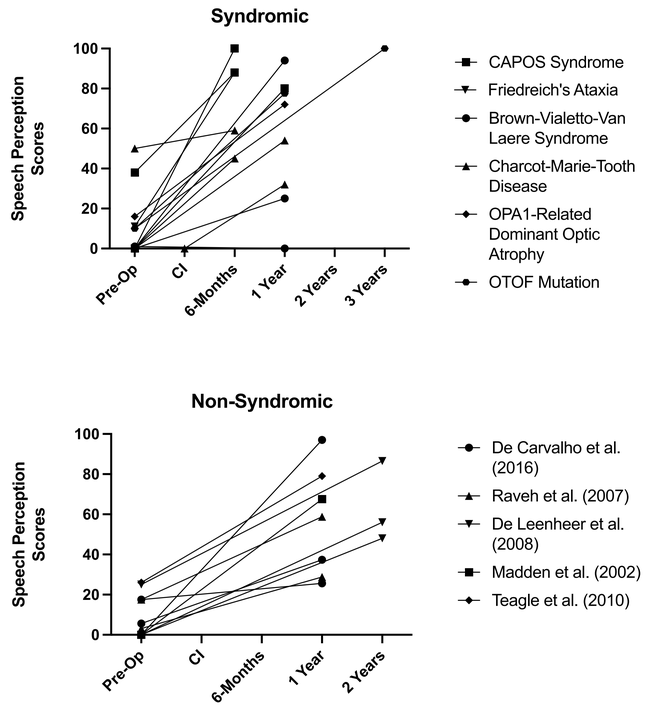
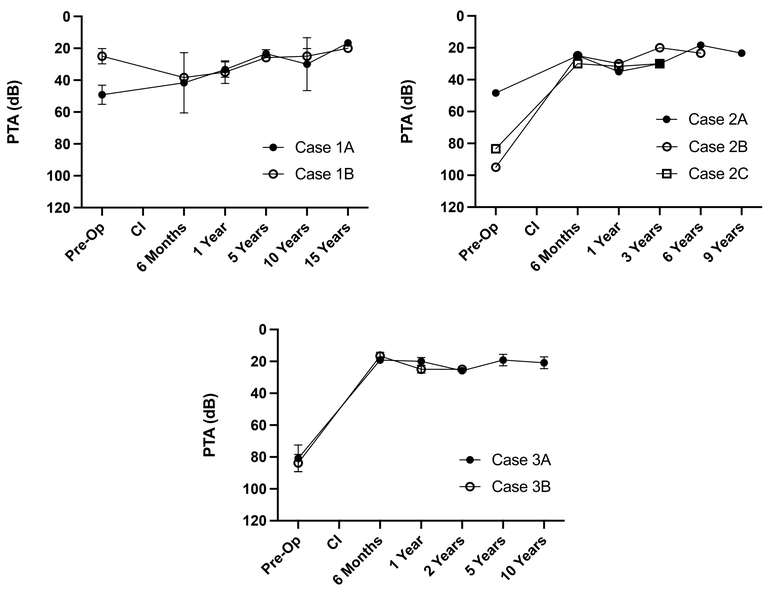


Fig 3. Effect of CI on Pure Tone Averages in Siblings with ANSD



Case Series (cont.)

- Patient 3A:
  - Diagnosed with bilateral auditory neuropathy at 5-months-old
  - OtoGenome Hearing Loss genetic testing demonstrated **heterozygous variants in GJB2, DFNB31, DIAPH1, EYA4, and MYO1A**
  - Bilateral Cochlear CIs at 12-months-old
  - To date, demonstrates improved hearing thresholds by PTA at 10 years since CI and continues to receive speech services but endorses good performance in school and achieved **HINT and AzBio scores with 91% and 94% accuracy, respectively**
- Patient 3B (younger sister):
  - Diagnosed with bilateral auditory neuropathy at 2-months-old
  - Bilateral Cochlear CIs at 11-months-old
  - To date, follow up audiometry demonstrates improved hearing thresholds by PTA at 2 years since CI with **84% accuracy in speech perception on the NU-CHIPS test;** formal speech and language evaluation continues to indicate delayed auditory skills, language expression and comprehension, and speech development

Discussion

- Our current review of the literature demonstrates that although outcomes may vary for CI in ANSD, all patients with either syndromic or non-syndromic ANSD demonstrated improvement in hearing sensitivity and speech perception following CI
- Similarly, our case series of three sets of siblings with ANSD demonstrates that CI improves hearing thresholds with long-term improvement in various speech performance measures
- Future investigation should explore the implementation of standardized language assessments to evaluate the long-term effects of CI in ANSD

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