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# GENETIC ETIOLOGY AND COCHLEAR IMPLANT OUTCOMES IN CHILDREN: A 10-YEAR RETROSPECTIVE COHORT

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Background:
Severe-to-profound sensorineural hearing loss (SNHL) in pediatric patients has multiple etiologies, including perinatal complications, inner ear malformations, and genetic causes. Cochlear implantation (CI) is the most common intervention, yet outcomes can vary widely. Given that CIs bypass the membranous labyrinth but depend on a functional spiral ganglion, we hypothesized that outcomes may differ based on the site of cochlear dysfunction.
Objective:
To assess auditory outcomes in children with genetically determined deafness who received CIs at our center and compare results with the literature.
Setting:
Unidade Local de Saúde de Coimbra and Faculty of Medicine, University of Coimbra, Portugal.
Design:
Retrospective, observational cohort study.
Population:
Children up to 18 years of age with bilateral severe-to-profound SNHL of genetic origin who underwent CI surgery between August 2013 and July 2023.
Method:
Data were collected from medical records. Mutations present in only one patient were grouped under "Other." Outcomes were evaluated using pure-tone average (PTA) thresholds (Good: PTA ≤30dB, Moderate: 30< PTA ≤50dB, Poor: PTA >50dB) and auditory performance scales (CAP and SIR) across three rehabilitation intervals: 4–8, 10–14, and 22–26 months. Statistical analysis was conducted using SPSS.
Results:
A genetic cause was identified in 49% (N=77) of the cases. Twenty-four genes were implicated, with GJB2 (N=38) and GJB2/GJB6 compound heterozygosity (N=4) being most common in non-syndromic deafness; MYO7A (N=5, Usher syndrome) and BSND (N=4, Bartter syndrome) in syndromic cases. Mean age at diagnosis was 8.6 months, but in 17.5% who passed newborn screening, it rose to 17.7 months. No significant differences were found across mutation types (p=0.086). Most patients (81.8%) had bilateral profound SNHL, and 76.6% were implanted between 13–35 months. Only 3.9% were implanted within the first year. Audiometric outcomes were good in 62.7% and poor in only 3%. Though not statistically significant, MYO7A and "Other" mutations had the worst outcomes (66.7% and 69% moderate outcomes, respectively). CAP and SIR outcomes were not significantly associated with mutation type (p=0.579, p=0.475) or age at implantation (p=0.489, p=0.376). However, children with neurocognitive impairment showed significantly worse CAP and SIR results (p=0.006, p=0.002), regardless of mutation (p=0.620).
Conclusions:

Contrary to some literature, mutations affecting the spiral ganglion did not significantly predict CI outcome in our cohort. Methodological limitations, such as small sample sizes and mutation heterogeneity, may explain these findings.