

SPOTTING HIGH-RISK DEAF SYNDROMES IN COCHLEAR IMPLANT CANDIDATES: A CLINICAL STUDY

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Abstract

Background: Cochlear implantation has revolutionized auditory rehabilitation in children with severe to profound hearing loss. However, deafness may be a prominent feature of several syndromes associated with multisystem involvement, which can significantly influence surgical safety, anesthesia, rehabilitation, and long-term outcomes.

Objective: To identify the prevalence and spectrum of syndromic deafness in cochlear implant candidates and to highlight high-risk syndromes requiring multidisciplinary evaluation.

Materials and Methods: A clinical observational study was conducted at a tertiary care referral center. Children presenting with hearing loss were evaluated clinically, audiological, radiologically, and genetically where indicated. Syndromic and non-syndromic cases were analyzed, and associated systemic anomalies were documented.

Results: Out of 1078 children with hearing loss, 165 (15.3%) had syndromic deafness. Among these, only 18.18% were clearly labeled syndromes, while 78.78% remained unclassified. Auditory-pigmentary syndromes formed the largest group. Several syndromes posed significant anesthetic, surgical, and rehabilitative challenges, particularly those with craniofacial, cardiac, renal, and airway abnormalities.

Conclusion: Early recognition of syndromic deafness is critical in cochlear implant candidates. Identification of high-risk syndromes can prevent life-threatening complications, guide surgical planning, and improve long-term outcomes through a multidisciplinary approach.

Keywords: Syndromic deafness, cochlear implant, high-risk syndromes, pediatric hearing loss, multidisciplinary care

Introduction

Diagnosing deafness is often straightforward; however, identifying associated syndromes and multisystem involvement requires a higher level of clinical suspicion and expertise. In syndromic deafness, the prominence of hearing loss may obscure other potentially serious anomalies, some of which may be life-threatening if overlooked.

With expanding selection criteria for cochlear implantation, clinicians increasingly encounter children with complex syndromes that pose challenges during surgery, anesthesia, and rehabilitation. This necessitates a comprehensive, multidisciplinary evaluation involving otolaryngologists, audiologists, pediatricians, geneticists, cardiologists, ophthalmologists, and anesthesiologists.

Materials and Methods

This observational study was conducted at the Government ENT Hospital, Hyderabad. Children presenting with hearing loss underwent:

- Detailed clinical examination
- Audiological evaluation (BERA, PTA where applicable)
- Radiological imaging (CT/MRI temporal bone)
- Genetic evaluation when indicated

Syndromes were categorized based on etiology:

- Chromosomal anomalies
- Monogenic (Mendelian) inheritance
- Multifactorial inheritance

Results

Prevalence of Syndromic Deafness

| Total Cases | Syndromic | Non-Syndromic |
|-------------|-------------|---------------|
| 1078 | 165 (15.3%) | 913 (84.7%) |

Labelled vs Unlabelled Syndromes

| Total Syndromes | Labelled | Unlabelled |
|-----------------|-------------|--------------|
| 165 | 30 (18.18%) | 135 (81.82%) |

Distribution of Labelled Syndromes

| Syndrome | Number | Percentage |
|-----------------------|--------|------------|
| Ear pits | 7 | 23.33 |
| Waardenburg | 6 | 20 |
| Usher's | 4 | 13.33 |
| Klippel-Feil | 2 | 6.66 |
| Rubella | 2 | 6.66 |
| CHARGE | 1 | 3.33 |
| Goldenhar | 1 | 3.33 |
| Hurler | 1 | 3.33 |
| Achondroplasia | 1 | 3.33 |
| Jervell-Lange Nielsen | 1 | 3.33 |
| Apert's | 1 | 3.33 |
| HITCH | 1 | 3.33 |

Syndrome Number Percentage

Syndromes Based on Organ/System Involved

| System | Number (%) |
|----------------|------------|
| Ocular | 24.18 |
| Dermatological | 9.63 |
| Cardiovascular | 8.88 |
| Dental | 8.15 |
| Respiratory | 5.18 |
| Skeletal | 5.18 |
| Craniofacial | 3.70 |
| Neurological | 2.96 |

High-Risk Deaf Syndromes Relevant to Cochlear Implantation

Certain syndromes pose significant perioperative risk:

Airway Risk

- Treacher Collins
- Klippel-Feil
- Hurler syndrome

→ Difficult intubation due to micrognathia, short neck, macroglossia

Cardiac Risk

- Jervell-Lange Nielsen
- LEOPARD syndrome
- CHARGE

→ Arrhythmias, prolonged QT, syncopal attacks, sudden death

Renal Risk

- Branchio-oto-renal (BOR) syndrome
- Alport syndrome

→ Progressive renal failure affecting anesthetic clearance

Representative Case Profiles

Branchio-Oto-Renal Syndrome

Six-year-old female with severe hearing loss, ear pits, branchial sinus, microtia, and facial palsy.

Klippel-Feil Syndrome

Eleven-year-old male with short neck, scoliosis, pigeon chest, and bilateral profound hearing loss.

Goldenhar Syndrome

Ten-year-old female with bilateral microtia, facial palsy, cervical vertebral fusion, and internal auditory canal hypoplasia.

Hurler Syndrome

Five-year-old male with mixed hearing loss, coarse facial features, hepatosplenomegaly, macroglossia, and respiratory compromise.

Jervell-Lange Nielsen Syndrome

Fourteen-year-old female with severe SNHL, syncope, prolonged QT interval, and cardiac arrhythmias.

Discussion

More than 400 syndromes associated with hearing loss have been described. The major challenge lies not in identifying inherited deafness but in recognizing syndromic associations with systemic risk. Genetic evaluation is justified not only for diagnosis but also to prevent fatal complications, particularly in chromosomal anomalies such as Trisomy 13 and 18.

Mitochondrial mutations (A1555G) predisposing to aminoglycoside ototoxicity highlight the need for genetic vigilance. Cochlear implant candidates often undergo multiple anesthetic exposures, making preoperative risk assessment essential.

Conclusion

Early identification of syndromic deafness significantly improves survival, surgical safety, and rehabilitation outcomes in cochlear implant candidates. A multidisciplinary approach reduces morbidity, prevents avoidable mortality, and supports informed family counseling and future reproductive decisions.

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