Cochlear Implantation in Patients with Osteogenesis Imperfecta: A Scoping Review of Reported Cases and Outcomes

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Introduction

Osteogenesis Imperfecta (OI), also known as brittle bone disease, is a genetic bone disorder most commonly caused by mutations in the COL1A1 and COL1A2 genes, which are responsible for encoding type I collagen.[1]

- Up to 70% of patients with OI develop hearing loss (conductive, sensorineural, or mixed).[2]
- Patients may benefit from hearing assistive devices, which can improve access to sound and daily function.[3]
- Cochlear Implantation (CI) may address profound/mixed loss but faces unique surgical challenges (brittle bones, distorted anatomy, risk of facial nerve stimulation).[3]

Objective

To map the existing literature on cochlear implantation in patients diagnosed with osteogenesis imperfecta and summarize demographics, CI feasibility, complications, and outcomes.

Methods

- PubMed, Scopus, and Embase were searched using *"osteogenesis imperfecta" AND ("cochlear implant" OR "cochlear implantation")* under PRISMA-ScR guidelines. Reports and studies with ≥1 confirmed OI patient receiving CI and reporting surgical or audiologic outcomes were included in this study.
- After duplicates were removed, 9 studies were included.
- Data including patient demographics, surgical outcomes, and audiologic performance post-surgery, were collected.

Results

- 9 reports described 12 patients with osteogenesis imperfecta who received 13 cochlear implants (one bilateral).
- Insertion was successful in 12 of 13 ears, with only one electrode misplacement leading to nonuse.
- Facial nerve stimulation (electrical current from a cochlear implant spreads and unintentionally activates the nearby facial nerve) occurred in a minority of cases, generally manageable with programming changes.
- Excluding the nonuser, hearing outcomes were consistent with typical CI expectations.

Table 1. *Summary of patient characteristics, surgical outcomes, and device arrays in OI cochlear implant reports*

Study (Year)	Patient (Age/Sex)	Side	Insertion Complications	Adverse Effects	Follow-up Length	Device Array
Szilvássy et al., 1998	50 F	Unilateral (ear ns)	None	FNS - mid-array electrodes deactivated	7 days	Nucleus 22 (straight lateral wall)
Huang et al., 1998	42 F	Left ear	None	None	3 mo	Nucleus 22 (straight lateral wall)
Migirov et al., 2003	6 M	Right ear	None	None	6 mo	Nucleus 24 Contour (perimodiolar)
Streubel & Lustig, 2005 – Case 1	31 F	Left ear	None	FNS - basal electrodes turned off during mapping	12 mo	Med-El Combi 40+ (straight lateral)
Streubel & Lustig, 2005 – Case 2	46 F	Left ear	None	None	12 mo	Nucleus 24 Contour (perimodiolar)
Rotteveel et al., 2008 – Case 1	45 F	Unilateral (ear ns)	None	FNS - several electrodes reduced/off	12 mo	Nucleus 24 (straight lateral wall)
Rotteveel et al., 2008 – Case 2	51 F	Unilateral (ear ns)	None	FNS - 5 electrodes switched off	12 mo	Nucleus 24 (straight lateral wall)
Rotteveel et al., 2008 – Case 3	54 M	Unilateral (ear ns)	Electrode misplacement	FNS - all electrodes off	3 mo	Clarion C1 (lateral wall)
Makizumi et al., 2013	54 F	Right ear	None	None	6 mo	Nucleus CI24R "Contour Advance" (perimodiolar)
Marfatia et al., 2020	14 F	Unilateral (ear ns)	None	None	12 mo	Nucleus Contour Advance (perimodiolar)
Takatsu, 2023	2 F	Bilateral	None	None	na	na
Mariani et al., 2025	50 F	Right ear	None	None	6 mo	Nucleus Contour Advance (perimodiolar)

Abbreviations: **FNS**, facial nerve stimulation; **ns**, not specified; **na**, not available.

- Most reported CI recipients were female adults, with unilateral implantation being far more common than bilateral.

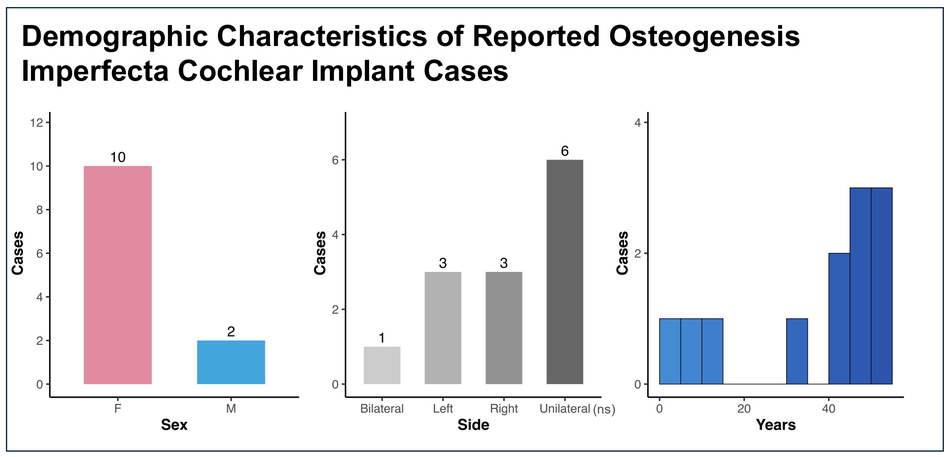

Figure 1. *Distribution of sex, side (laterality) of implantation, and patient age at surgery among published reports (ns, laterality not specified)*

Results (continued)

- Across the limited number of cases (3-5) that reported standardized audiologic outcomes (Figure 2), speech perception scores improved post-implantation, with consistent gains for words, sentences, and phonemes.

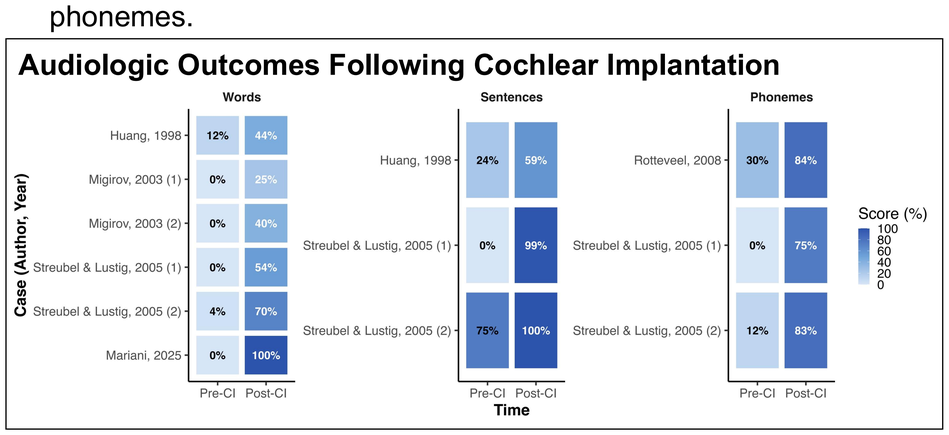

Figure 2. *Pre- and post-implant performance scores for words, sentences, and phonemes in individual reported cases*

Discussion

- Indication for amplification devices (hearing aids) and cochlear implantation generally mirrors standard practice, but is influenced by unique anatomic and surgical considerations in OI patients, such as increased fracture risk and indistinct surgical landmarks due to temporal bone alterations.
- Facial nerve stimulation (FNS) may be a more common adverse effect amongst OI patients as a result of altered temporal bone and otic capsule structure and thickness.
- Cochlear implantation provides meaningful hearing benefits to patients when carefully planned.
- Current literature is limited to very few case reports/series with inconsistent reporting of pre- and post-surgical outcomes.

Conclusions

- Despite the challenges, cochlear implantation in patients with OI is technically feasible in carefully selected patients.
- In the 12 patients that were studied, both children and adults benefited from CI, though it is believed that early intervention and evaluation may optimize outcomes.
- Larger scale studies are needed to improve guidelines and strengthen the evidence of implantation in this rare population.

References

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