Hearing loss in osteogenesis imperfecta: follow-up and treatment

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Follow-up of hearing loss in OI

Is there a need for follow-up of hearing in OI?

How should this follow-up be organized?
Audiologic phenotype in OI

Deafness
Sensorineural
Mixed
Conductive

Age category

Hearing-impaired ears (%)

N=364 ears

< 10 y. (n=32)
10-19 y. (n=88)
20-29 y. (n=78)
30-39 y. (n=48)
40-49 y. (n=66)
50-59 y. (n=32)
60 y. ≤ (n=22)

6.3%
34.1%
38.5%
76.0%
54.5%
78.1%
77.3%
Age-related typical audiograms (ARTAs)

Conductive / mixed hearing loss

Sensorineural hearing loss
# Cross-sectional study

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Conductive/mixed loss (36.3%)</th>
<th>Pure sensorineural loss (11.6%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Onset</strong></td>
<td>2nd to 4th decade (mean 21.3 y.)</td>
<td>Any age (mean 30.2 y.)</td>
</tr>
<tr>
<td><strong>Severity</strong></td>
<td>• Age &lt; 40 y.: mild (15-40 dB HL) to moderate (40-70 dB HL)</td>
<td>Mild (95.2%) or moderate (4.8%)</td>
</tr>
<tr>
<td></td>
<td>• Age ≥40 y.: mild to profound (≥ 95 dB HL)</td>
<td></td>
</tr>
<tr>
<td><strong>Audiometric configuration</strong></td>
<td>• Flat (70.5%)</td>
<td>• Flat (45.2%)</td>
</tr>
<tr>
<td></td>
<td>• Sloping (27.2%)</td>
<td>• Sloping (50.0%)</td>
</tr>
<tr>
<td></td>
<td>• Rising (2.3%)</td>
<td>• Through (4.8%)</td>
</tr>
<tr>
<td><strong>Symmetry</strong></td>
<td>Predominantly symmetric</td>
<td>Predominantly symmetric</td>
</tr>
<tr>
<td><strong>ARTAs</strong></td>
<td>• ABG frequency-specific but age-independent</td>
<td>• ATD: 0.2 dB/y. (0.5-1.0-2.0 kHz) to 1.2 dB/y. (0.8 kHz)</td>
</tr>
<tr>
<td></td>
<td>• Annual threshold deterioration (ATD) 0.6 dB/y. (≤ 2 kHz) to 0.8 dB/y. (8 kHz)</td>
<td></td>
</tr>
</tbody>
</table>
Follow-up study

• Subjects
  o 36 OI-patients
  o Age: 6-79 y.
  o COL1A1 or COL1A2 mutation

• Follow-up audiometry
  o Interval: 4 years
  o Audiologic testing:

<table>
<thead>
<tr>
<th>Initial evaluation</th>
<th>Follow-up evaluation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tympanogram</td>
<td>Tympanogram</td>
</tr>
<tr>
<td>Acoustic Stapedius reflex</td>
<td>Acoustic Stapedius Reflex</td>
</tr>
<tr>
<td>Audiogram</td>
<td>Audiogram</td>
</tr>
<tr>
<td>Oto-acoustic emissions</td>
<td>Oto-acoustic emissions</td>
</tr>
</tbody>
</table>

Follow-up study

- Normal hearing: 66%
- Conductive/mixed: 3%
- Mixed: 10%
- Sensorineural: 21%
Follow-up study

<table>
<thead>
<tr>
<th>Measurement</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Air conduction thresholds*</td>
<td>Significant increase (P&lt;0.001)</td>
</tr>
<tr>
<td>Bone conduction thresholds*</td>
<td>Significant increase (p&lt;0.001)</td>
</tr>
<tr>
<td>Air-bone gap</td>
<td>Stable</td>
</tr>
</tbody>
</table>

* Corrected for age-related hearing deterioration (P95 – ISO 7023)

Hearing loss progression is dominated by sensorineural deterioration

~ cross-sectional studies (Garretsen et al. 1997; Swinnen et al. 2012)
Follow-up of hearing loss in OI

- Regular audiometry should be part of the multidisciplinary follow-up in OI patients
  - Pediatrics: yearly
  - Adults: every 2-5 years, depending on the indication

- Proposed audiometric test battery:

<table>
<thead>
<tr>
<th>Audiometric test</th>
<th>Intended measure</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pure-tone audiometry</td>
<td>Degree and type of hearing loss</td>
</tr>
<tr>
<td>Tympanometry and acoustic stapedius reflexes</td>
<td>Middle ear compliance (thin eardrums!)</td>
</tr>
<tr>
<td>Oto-acoustic emissions</td>
<td>Outer hair cell function (inner ear)</td>
</tr>
<tr>
<td>Speech audiometry (if hearing loss is present)</td>
<td>Functional hearing</td>
</tr>
</tbody>
</table>

Differential diagnosis between conductive versus sensorineural hearing loss, especially in the early stages of hearing impairment.
Follow-up of hearing loss in OI

Hearing loss in OI is variable (multifactorial?)

Recommendations:

• Hearing protection in noisy situations
• Adequate treatment of middle ear infections
Treatment of hearing loss in OI

- Stapes surgery
- Hearing aids
- Implantable hearing devices
1. **Stapes surgery: reduction of air-bone gap**

![Graph showing bone conduction and air conduction](image)

- **Bone conduction** is shown by the dashed line, which is less than the solid line representing **Sensorineural component**.
- **Air-bone gap** is indicated by the gap between the two lines.
- **Conductive component** is represented by the solid line.

A diagram illustrates the dB HL (decibels hearing level) on the y-axis and kHz (kilohertz) on the x-axis, with various dB levels marked for different frequencies.
1. **Stapes surgery**

![Stapes surgery diagram with labeled parts: Footplate, Caput, Crura, Incus, Malleus, Eardrum, Stapedotomy tool.](image-url)
1. **Stapes surgery**

- Controversy benefit of stapes surgery in OI
  - Intraoperative complications (mucosal bleeding, ossicular fragility)
  - Progressive sensorineural hearing loss
- **Large series of stapes surgery in OI:**

<table>
<thead>
<tr>
<th>Authors</th>
<th>Year</th>
<th>Country</th>
<th>No. of operated ears</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shea &amp; Postma</td>
<td>1982</td>
<td>USA</td>
<td>51</td>
</tr>
<tr>
<td>Pedersen</td>
<td>1985</td>
<td>Sweden</td>
<td>42</td>
</tr>
<tr>
<td>Garretsen &amp; Cremers</td>
<td>1990</td>
<td>Netherlands</td>
<td>52</td>
</tr>
<tr>
<td>Kuurila et al.</td>
<td>2004</td>
<td>Finland</td>
<td>43</td>
</tr>
<tr>
<td>Swinnen et al.</td>
<td>2009</td>
<td>Netherlands</td>
<td>15</td>
</tr>
<tr>
<td>Swinnen et al.</td>
<td>2012</td>
<td>Belgium &amp; Netherlands</td>
<td>34</td>
</tr>
</tbody>
</table>


# 1. Stapes surgery

## Results

**Intraoperative findings:**

<table>
<thead>
<tr>
<th></th>
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</tr>
</thead>
<tbody>
<tr>
<td><strong>No. of ears</strong></td>
<td>42*</td>
<td>58</td>
<td>43</td>
<td>62</td>
</tr>
<tr>
<td><strong>a) Stapes footplate</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fixed, No. (%)</td>
<td>42 (100)</td>
<td>54 (93)</td>
<td>43 (100)</td>
<td>62 (100)</td>
</tr>
<tr>
<td>Thick/brittle, No. (%)</td>
<td>26 (62)</td>
<td>32 (55)</td>
<td>21 (49)</td>
<td>NA</td>
</tr>
<tr>
<td><strong>b) Stapes crura</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Fractured, No. (%)</td>
<td>2 (5)</td>
<td>10 (17)</td>
<td>5 (12)</td>
<td>NA</td>
</tr>
<tr>
<td>Atrophic, No. (%)</td>
<td>17 (40)</td>
<td>22 (38)</td>
<td>14 (33)</td>
<td>13 (21)</td>
</tr>
<tr>
<td><strong>c) Mucosa</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Excessive bleeding, No. (%)</td>
<td>8 (19)</td>
<td>12 (21)</td>
<td>22 (51)</td>
<td>18 (29)</td>
</tr>
</tbody>
</table>

* Revision cases excluded
1. **Stapes surgery**

**Results**

**Audiometry:**

![Graph showing preoperative audiogram (n=26)]

- **Mean preoperative audiogram (n=26)**
1. **Stapes surgery**

**Results**

**Audiometry:**

Mean short-term postoperative (mean 1.5 months) audiogram (n=26)
1. **Stapes surgery**

**Results**

Audiometry:

Mean long-term postoperative (mean 11.4 y) audiogram (n=26)
1. **Stapes surgery**

**Conclusion**

- Efficient technique to reduce conductive hearing loss in OI
- Hearing gain for several decades in the majority of the patients
- Experienced otologists since higher rate of intra-operative complications such as atrophic ossicles, bleeding mucosae
- Less benefit should be expected in revision stapes surgery

**Revision surgery (n=7)**
- Intraoperative findings: erosion of incus
- Audiometry
  - Short-term: successful in 7/7 ears
  - Long-term: successful in 3/4 ears
2. **Hearing aids**

- Efficient for mild to moderate hearing loss of both types:
  - Conductive /mixed hearing loss
  - Sensorineural hearing loss

- Mixed hearing losses: stapes surgery may facilitate hearing aid adjustments

- Purpose: improvement of speech understanding
3. **Implantable auditory devices** (1)

- Bone-anchored hearing aids (BAHA)
  
  - General indication: conductive hearing loss
  - **OI-population**: not preferable in OI due to the concomittant progression of the bone conduction threshold
3. Implantable auditory devices (2)

- Cochlear implants
3. Implantable auditory devices (2)

• Cochlear implants
  - General indication: severe to profound hearing loss; no functional hearing with HAs
  - OI-population: higher risk of complications due to:
    • Bony obliteration in the cochlea
    • Hypodense pericochlear bone may result in spread of electrical current and facial nerve stimulation
Treatment of OI-related hearing loss

• Personalised treatment based on:
  – Degree and type of hearing loss
  – Radiological information of the temporal bone
  – Progression rate of the hearing loss

• Experienced otologists!
THANK YOU!