CHARGE Syndrome: Characteristics and Outcomes in Pediatric Cochlear Implantation

Nancy Young, MD, Beth Tournis, AuD, Jenelle Sandy, AuD
Ann & Robert H. Lurie Children’s Hospital of Chicago
Disclosures

Cochlear Americas Audiology Advisory Board Member
Lurie Children’s CI Team

Helping Children To Achieve Their Full Potential
CHARGE

• Complex disorder usually affecting hearing, balance and vision
• Life threatening heart and airway problems often present at birth
• Malformed cochlea present in 90%
• Prediction of cognitive potential is not possible early in life:
  – Intelligence in normal hearing cannot be accurately predicted
  – Underestimation of intelligence likely in deaf children due to low language level
  – Present of impaired vision and motor function further increases difficulty of evaluation and likelihood of underestimation of cognitive potential
Study Design

• Retrospective chart review
• Subject inclusion criteria
  – Diagnosis of CHARGE Syndrome
  – 12 month minimum of device use
• Outcomes Post-CI
  – Auditory level & device use
  – Communication mode(s) for receptive & expressive language
Auditory Level

- **Level 1**
  - No benefit, minimal detection
- **Level 2**
  - Improved detection (SAT < 30dB)
- **Level 3**
  - Closed-set speech perception (ESP 2,3,4)
- **Level 4**
  - Open-set speech perception
Subject Characteristics (N=12)

- Average age at CI: 3.5 years (range 1.7 - 8.2)
- Implanted unilaterally with full electrode array
- Gender: 7 males and 5 females
- Average length of follow up: 4.8 years (range 2.0 – 10.1)
- All children received long-term aural habilitation
# Auditory Level At Last Follow Up: No Detection/Detection only

<table>
<thead>
<tr>
<th>Patient</th>
<th>Pre CI Auditory Level &amp; aided SAT (dB)</th>
<th>Post CI Auditory Level &amp; aided SAT (dB)</th>
<th>Follow Up Interval (yrs)</th>
<th>CND</th>
<th>Expressive Communication Mode Post CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>1</td>
<td>1</td>
<td>2.2</td>
<td>-</td>
<td>Sign</td>
</tr>
<tr>
<td>4</td>
<td>1</td>
<td>2</td>
<td>4.7</td>
<td>-</td>
<td>Sign/oral</td>
</tr>
<tr>
<td>5</td>
<td>1</td>
<td>2</td>
<td>2.8</td>
<td>-</td>
<td>Sign/oral</td>
</tr>
<tr>
<td>7</td>
<td>1</td>
<td>1</td>
<td>10.1*</td>
<td>+</td>
<td>Sign/AC^</td>
</tr>
<tr>
<td>8</td>
<td>1</td>
<td>2</td>
<td>3.9</td>
<td>+</td>
<td>Sign/AC^</td>
</tr>
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<td>11</td>
<td>1</td>
<td>2</td>
<td>3.7</td>
<td>-</td>
<td>Sign/oral</td>
</tr>
</tbody>
</table>

Level 1= no benefit; Level 2= SAT< 30dB; Level 3= closed-set; Level 4= open-set

*Inconsistent use
^AC=augmentative communication
## Auditory Level At Last Follow Up: Closed-set

<table>
<thead>
<tr>
<th>Patient</th>
<th>Pre CI Auditory Level &amp; Perception Category</th>
<th>Post CI Auditory Level</th>
<th>Follow Up Interval (yrs)</th>
<th>CND</th>
<th>Expressive Communication Mode Post CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1</td>
<td>3</td>
<td>6.2</td>
<td>+</td>
<td>Sign/oral/AC^</td>
</tr>
<tr>
<td>9</td>
<td>1</td>
<td>3</td>
<td>5.2</td>
<td>+</td>
<td>Sign/oral</td>
</tr>
<tr>
<td>10</td>
<td>3</td>
<td>ESP 2*</td>
<td>2.5</td>
<td>+</td>
<td>Oral*</td>
</tr>
</tbody>
</table>

Level 1= no benefit; Level 2= SAT< 30dB; Level 3= closed-set; Level 4= open-set

^inconsistent CI use, open-set in non-CI ear

^AC=augmentative communication
# Auditory Level At Last Follow Up: Open-set

<table>
<thead>
<tr>
<th>Patient</th>
<th>Pre CI Auditory Level &amp; Perception Category</th>
<th>Post CI Auditory Level</th>
<th>Follow Up Interval (yrs)</th>
<th>CND</th>
<th>Expressive Communication Mode Post CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>3</td>
<td>1 -</td>
<td>4 Open-set</td>
<td>5.0</td>
<td>-</td>
<td>Sign/oral</td>
</tr>
<tr>
<td>6</td>
<td>1 -</td>
<td>4 Open-set</td>
<td>9.3</td>
<td>-</td>
<td>Sign/oral/AC</td>
</tr>
<tr>
<td>12</td>
<td>2 ESP 2</td>
<td>4 Open-set</td>
<td>2.0</td>
<td>+</td>
<td>Sign/oral</td>
</tr>
</tbody>
</table>

Level 1 = no benefit; Level 2 = SAT < 30dB; Level 3 = closed-set; Level 4 = open-set

^AC=augmentative communication
## Time Course For Development Of Speech Perception

<table>
<thead>
<tr>
<th>Patient</th>
<th>ESP 2 (years)</th>
<th>ESP 3 (years)</th>
<th>ESP 4 (years)</th>
<th>Open-set (years)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>2</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>1</td>
<td>1.5</td>
<td>2.5</td>
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<tr>
<td>6</td>
<td></td>
<td>1.5</td>
<td>1.5</td>
<td>3.0</td>
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<tr>
<td>9</td>
<td>6</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>10*</td>
<td>1.5</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>12</td>
<td></td>
<td>0.5</td>
<td>0.8</td>
<td></td>
</tr>
</tbody>
</table>

*ESP 3 pre-implant; Limited use at school only, has open-set in non-CI ear
Ethan: Age 8, 5 Years Post CI
(implanted age 36 months)

• Tracheotomy
• Pre CI
  – Bilateral severe to profound
  – Aided SAT 55dB
  – Receptive: none measurable
  – Expressive: natural gesture
  – Implanted in ear with normal 8th nerve
• Post CI
  – Open-set achieved: 36 months
  – Receptive: oral with sign
  – Expressive language:
    • Sign
    • Augmentative communication
Johan: Age 6 Years
(Implanted 4 years, 4 months)

- Pre-implant: MAIS=70%
  - Right: mild sloping to moderately-severe unaided thresholds
    - Aided SAT 20dB
  - Left: moderately-severe sloping to profound unaided thresholds
    - Aided SAT 25dB
  - Sign and Spoken language (poor intelligibility)
  - Imaging: **left eighth nerve hypoplasia** in addition to bilateral vestibular malformation

- Post CI Left Ear
  - Open-set achieved by 9 months
  - MLNT 42% in aided and CI ear at 2 years
  - Improved speech intelligibility
Summary

- Majority (10 of 12)
  - Achieved at least significant improvement detection
  - Consistent device use
- Measurable improvements in auditory skill usually occurred over a long time course
- Improved detection and speech perception may occur despite cochlear nerve deficiency as defined by MRI
- Multiple communication strategies were used
Conclusion

- Children with CHARGE syndrome are viable cochlear implant candidates although outcomes are difficult to predict
- Benefits range from improved detection to open-set speech perception and spoken language
- Family counseling should include:
  - Expected longer time course for auditory skills to emerge
  - Need for aural habilitation despite initial lack of progress
  - Sign language & augmentative communication are likely to be integral to communication
Everybody is a genius. But if you judge a fish by its ability to climb a tree, it will spend its whole life thinking it's stupid.

- Albert Einstein