Outcomes of Cochlear Implantation for Pediatric Patients with CHARGE Syndrome: Beyond Surgery

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What is CHARGE Syndrome?

- Coloboma of the eye
- Heart defect
- Atretic choanae
- Retarded growth or development
- Genital hypoplasia
- Ear anomalies
Diagnostic Criteria for CHARGE:
All four major characteristics (the four Cs) or three major and three minor

Challenges to CHARGE and CI

- Life Threatening Medical Conditions
- Abnormal Ear/ Facial Nerve Anatomy
- Neurologically Complex
- Behavioral Issues
<table>
<thead>
<tr>
<th></th>
<th>Ocular Coloboma</th>
<th>Choanal Atresia or Stenosis</th>
<th>Cranial Nerve Dysfunction</th>
<th>Characteristic CHARGE Ear</th>
<th>Genital Hypoplasia</th>
<th>Developmental Delay</th>
<th>Cardiovascular Malformation</th>
<th>Growth Deficiency</th>
<th>Orofacial Cleft</th>
<th>Tracheoesophageal Fistula</th>
<th>Distinctive Facial Features</th>
<th>GENETIC TESTING</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patient A, Female, 21 yrs</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Patient B, Male, 8 yrs</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>Patient C, Female, 7 yrs</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
</tbody>
</table>
**Pre-Activation Patient Info**

<table>
<thead>
<tr>
<th>Pt</th>
<th>Age of HL dx</th>
<th>Age of CHARGE dx</th>
<th>Degree of HL</th>
<th>Age of CI</th>
<th>Anatomical considerations</th>
<th>Communication mode</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>8 mo</td>
<td>14 yrs</td>
<td>Progressive, profound AU</td>
<td>12 yrs, 19 yrs</td>
<td>Bifid facial nerve</td>
<td>ASL &amp; spoken English</td>
</tr>
<tr>
<td>B</td>
<td>9 mo</td>
<td>4 yrs</td>
<td>Profound R, absent nerve L</td>
<td>16 mo</td>
<td>Abnormal round window niche</td>
<td>ASL</td>
</tr>
<tr>
<td>C</td>
<td>4 mo</td>
<td>neonatal</td>
<td>Severe R, Profound L</td>
<td>28 mo</td>
<td>Abberent facial nerve</td>
<td>ASL &amp; picture exchange</td>
</tr>
</tbody>
</table>

*All patients received right-sided cochlear implants, full insertion of electrode array*
## Post-Activation Patient Info

<table>
<thead>
<tr>
<th>Pt</th>
<th>Internal electrode</th>
<th>Coding strategy</th>
<th>Facial stimulation?</th>
<th>Communication mode</th>
<th>Auditory outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>AB Hi-Res 90K 1J</td>
<td>Hi-Res S</td>
<td>Yes – affected set M levels</td>
<td>Same as pre-op</td>
<td>45-75 dBHL thresholds, ESP Category 1</td>
</tr>
<tr>
<td>B</td>
<td>AB Hi-Res 90K 1J</td>
<td>Hi-Res P w/ Fidelity 120</td>
<td>No</td>
<td>Same as pre-op</td>
<td>15-25 dBHL thresholds, 40% on NU-CHIPS</td>
</tr>
<tr>
<td>C</td>
<td>Cochlear CI512</td>
<td>ACE</td>
<td>No</td>
<td>Same as pre-op</td>
<td>25-40 dBHL thresholds</td>
</tr>
</tbody>
</table>
Lit Review – Audiological Outcomes

Bauer et al. (2002):
- 5 CI pts w/CHARGE
- ESP: 1 achieved Category 4; others Categories 1 & 2
- Mild aided sound field thresholds

Buchman et al. (2004):
- 4 CI pts w/CHARGE
- 1 achieved closed-set speech recognition

Lanson et al. (2007):
- 10 CI pts w/CHARGE
- No formal speech measures
- Improved scores on IT-MAIS post-op
- Mild-moderate aided sound field thresholds

Southwell et al. (2010):
- 3 CI pts w/CHARGE
- 1 scored 89% on HINT
- 2 w/ no speech perception
- Mild-severe aided sound field thresholds
Lit Review - Behavioral Profile

Bernstein & Denno (2005):
• N=29; Compulsive Behavior Checklist
• Majority showed repetitive behaviors
• Self-stim, maladaptive behaviors, tics, obsessive-compulsive behaviors

Smith et al. (2005):
• N=13; parent-report questionnaires (CDI, BASC, SCQ, Vineland ABS)
• Atypical behaviors noted, not “internalizing” or “externalizing”
• Majority showed Sz consistent with ASD. Differential dx? Part of CHARGE?

Vervloed et al. (2006):
• N=27; Behaviors corresponding with medical conditions
• Cerebral deficits assoc. with withdrawn behavior & depressed mood
• Deafblindness assoc. with decreased communication & language

Hartshorne et al. (2005):
• N=160; parent questionnaire (Autism Behavior Checklist)
• CHARGE children scored differently than Autism or Deafblind – unique
Lit Review – Impact on Quality of Life

Hartshorne et al. (2009):
- N=87; **Sleep disturbances** (SDSC used to assess sleep)
- ½ - clinically sig. sleep problems
- Degree of visual impairment & mult. Disabilities → worse sleep

Wulffaert et al. (2009):
- N=22; parent-report (Parenting Stress Index, Dev Beh checklist, Vineland)
- 59% - Reported high **parenting stress**
- Child’s behavior probs, autistic characteristics, self-absorbed & disruptive behaviors → higher stress

Hartshorne et al. (2007):
- N=98; parent-report questionnaires (CHARGE Hx questionnaire, BRIEF, Autism Behv Checklist – ABC)
- **Executive dysfunction** → reduced flexibility, monitoring, inhibiting

Wachtel et al. (2007):
- N=87; **Psychiatric Dx** (Dev Behav Checklist; CHARGE Hx questionnaire)
- High co-morbidity with OCD, PDD, ADHD
- Anti-depressants most common med (SSRIs – treat anx, esp. OCD; antipsychotics for PDD Sx)
Beyond Surgery: Common Outcomes

Use American Sign Language for Communication

Alert to Sounds; Show Environmental Awareness

Improved “Connectedness” with Others
Patient Snapshots

21 Female
- Attended college 1 yr; bullied; transferred
- Dx “Asperger” as child; Forced puberty

8 Male
- Attends Deaf school; tolerates CI ~ 30 min.
- Nearly always 1:1 supervision

7 Female
- Attends deaf-blind program; Uses <5 signs
- Self-injurious (biting, picking); ABA therapy; not wearing CI since 9/14
Quality of Life:
Psychological Outcomes

- Heightened Anxiety
- Sleep Problems
- Rigid, Inflexible or “OCD”
- Immaturity & Behavioral Dysregulation
Recommendations for Professionals
Caring for CHARGE Patients

Coordinated Care

Psychological Monitoring
Flexible Communication
Educate Parents & Patients
Thank you for listening

www.bostonchildrens.org/cochlearimplant

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References


