Cochlear Implantation in Children With CHARGE Association

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Objective: To explore the anomalies of the temporal bone found on radiologic examination, technical challenges in cochlear implantation, and audioligic benefit derived from cochlear implantation in a series of children with CHARGE association.

Design: Case series report.

Setting: Tertiary referral children's hospital pediatric cochlear implant program.

Patients: Six children with CHARGE association and sensorineural hearing loss.

Intervention: All patients were evaluated and followed up by the cochlear implant team. Cochlear implantation was attempted in all 6 children.

Main Outcome Measures: Computed tomographic scans and cochlear implantation operative records were reviewed, and their findings were correlated. Audiometric and speech perception data before and after cochlear implantation were compared.

Results: Five children with CHARGE association received implants. A sixth child did not because of an aberrant course to the facial nerve. The 5 children receiving implants obtained varying degrees of measurable benefit from their implants. All 6 children had temporal bone abnormalities seen on their computed tomographic scans and documented at the time of surgery.

Conclusions: Variations in the temporal bone anatomy of patients with CHARGE association can lead to increased technical challenges and risk to the facial nerve during cochlear implantation. Individual outcomes after implantation may vary; our patients receiving implants obtained benefit. Parents should be counseled thoroughly and have appropriate expectations before proceeding with implantation.


THE CRITERIA defining appropriate candidates for pediatric cochlear implantation have expanded significantly as professionals have learned of the benefits implants provide to children with deafness. Although these criteria now include very young children and children with more residual hearing, some controversy remains about whether children with deafness who have significant cognitive, developmental, and physical comorbidities should undergo cochlear implantation. Traditionally, these children have not been considered for cochlear implantation because of questionable benefit from implantation.

One such group of children includes those with CHARGE association, a mnemonic that refers to patients with ocular coloboma and/or choanal atresia and manifestations of at least 4 of the 7 most common findings of the association (coloboma, congenital heart disease, atresia choanae, postnatal retarded growth, retarded development and/or central nervous system anomalies, genital hypoplasia or hypogonadism, and ear anomalies or deafness). Findings that are seen less often include micrognathia, cleft palate, facial palsy, swallowing difficulty, tracheoesophageal fistula, DiGeorge syndrome, renal anomalies, and rib anomalies. These patients may have otologic manifestations involving the external, middle, and inner ear that contribute to profound deafness. The incidence of severe to profound sensorineural hearing loss has been reported to range from 34% to 38%, with as many as 75% of patients with CHARGE association having some degree of sensorineural hearing loss.

Treatment of patients with CHARGE association present multiple challenges. The original description of CHARGE association by Pagon et al includes 54 patients. All but 1 of these patients had some degree of mental retardation or developmental delay. Establishing the diagnosis of mental retardation in children with CHARGE association is complicated by the presence of visual (coloboma) and auditory deficits. It is difficult to predict before implantation how the addition of an auditory stimulus to a child with CHARGE association will...
effect his or her cognitive development and performance.

In considering cochlear implantation for patients with CHARGE association, attention must be given to the varied temporal bone anomalies that may be encountered. Variations in the anatomic course of the facial nerve place these children at increased risk of nerve injury during implantation surgery.

This study explores the anomalies of the temporal bone found on radiologic examination, technical challenges in performing cochlear implantation in these patients, and audiologic benefit that pediatric patients with manifestations of the CHARGE association receive from cochlear implantation.

**PATIENTS AND METHODS**

A query of the St Louis Children’s Hospital Cochlear Implant Program patient database identified 6 patients with manifestations of CHARGE association who had undergone evaluation and surgery for cochlear implantation. The radiologic, medical, and audiologic records of the 6 children were reviewed. The Washington University in St Louis, School of Medicine, Human Studies Committee approved the study.

As part of the standard evaluation for cochlear implantation, patients underwent computed tomography of the temporal bone. High resolution scans were obtained in the axial plane using the following parameters: 0° to −1.5° gantry angulation, slice thickness of 1.0 mm (5 patients) and 2.0 mm (1 patient), 160 to 410 milliamperage seconds, and 120 kV. A neuroradiologist (F.J.W.) who was blinded to the surgical findings reviewed the computed tomographic scans of each patient. Radiologic and intraoperative findings were correlated.

Operative reports were examined to ascertain intraoperative morphologic middle ear and temporal bone findings, degree of difficulty in performing the implantation, and complications during surgery.

Audiometric test results before and after implantation were reviewed to determine the benefit derived from implantation. Data sources included routine audiometric testing, speech perception testing, and parental questionnaires.

Before implantation, ear-specific unaided pure-tone thresholds were obtained. Aided sound-field testing using the patients’ hearing aids was also done. In addition, auditory brainstem response testing was performed for all patients who were unable to complete or had no responses on behavioral tests. After implantation, aided sound-field thresholds were obtained using the patients’ cochlear implants. All speech perception testing was completed at 70-dB sound pressure level using monitored live voice.

The Early Speech Perception test was used to assess closed-set word recognition abilities. Based on the results, a child’s speech perception abilities are categorized as (1) no speech perception, (2) pattern perception, (3) some word identification, or (4) consistent word identification. The chance score for each of the subtests is 25%.

The Glendonald Auditory Screening Procedure (GASP) was used to assess open-set speech perception abilities. The test consists of 12 words (GASP-W) and 10 questions (GASP-S) presented through listening alone. The child is required to repeat the word and answer the questions presented. The chance score is 0%.

The Meaningful Auditory Integration Scale (MAIS) or Infant Toddler–MAIS (IT-MAIS) was used to evaluate the child’s ability to make meaningful use of sound in everyday situations. The parental questionnaires assess 3 major areas: (1) how well the child has adjusted to his or her amplification device, (2) how well the child responds to sound, and (3) the ability of the child to derive meaning from auditory phenomena. A score of 0 indicates the inability of the child to make use of sounds in his or her everyday environment, and a score of 40 indicates that the child can consistently make use of and comprehend sounds.

**RESULTS**

The clinical manifestations of CHARGE association differed among the patients. Individual patient characteristics are outlined in Table 1. Data from the most recent evaluation, including age at time of implantation, duration of implant use, ear receiving the implant, device implanted, electrode insertion depth, communication mode, and years of implant use, are detailed in Table 2.

All 6 patients had abnormalities on review of their temporal bone computed tomography scans (Table 3, Figure 1, and Figure 2). The internal auditory canals were small bilaterally in 3 patients and unilaterally in 1 patient. Four patients had abnormally large vestibular aqueducts. All patients had malformed or absent semicircular canals and bilaterally malformed vestibules. The apical turn of the cochlea was hypoplastic or dysplastic in 3 patients. Middle ear anomalies were less prevalent. One patient had bilaterally partially fused ossicles. Unilateral effusions were seen in 2 patients. In all patients, the facial nerves coursed over the promontory bilaterally.

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Table 1. Occurrence of CHARGE Anomalies*

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Coloboma</td>
<td></td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Heart disease (congenital)</td>
<td></td>
<td>+</td>
<td></td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Atriaia choanae</td>
<td>+</td>
<td>-</td>
<td>+</td>
<td></td>
<td></td>
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<tr>
<td>Retarded growth (postnatal)</td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Retarded development or central nervous system anomalies</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<td>+</td>
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<tr>
<td>Genital hypoplasia (hypogonadism)</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>+</td>
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<td>+</td>
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<tr>
<td>Ear anomalies or deafness</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
<td>+</td>
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<tr>
<td>Tracheoesophageal fistula</td>
<td>-</td>
<td>+</td>
<td>-</td>
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<td>+</td>
<td>-</td>
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<tr>
<td>Cleft lip and palate</td>
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<td>-</td>
<td>-</td>
<td>+</td>
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<tr>
<td>Renal anomalies</td>
<td>-</td>
<td>+</td>
<td>-</td>
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<td>+</td>
<td>+</td>
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<tr>
<td>Rib anomalies</td>
<td>-</td>
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</tbody>
</table>

*Minus sign indicates not present; plus sign, present.
A review of the operative records revealed no abnormal findings documented intraoperatively in patient 1. In patient 2, the facial nerve passed lateral to the oval window. In patient 3, the lateral semicircular canal was absent, the incus appeared hypoplastic, and the cochlear promontory appeared flattened. In patient 4, the incus was hypoplastic, there was a hypoplastic remnant of the stapes superstructure, and the cochlear promontory appeared flattened. In patient 5, the lateral semicircular canal was absent, the incus was hypoplastic with a remnant of the stapes superstructure not attached to the footplate, and the facial nerve was dehiscent for almost 360° as it passed lateral to the oval window. In addition, a perilymphatic gusher was encountered after drilling the cochleostomy, and a 4 × 6-mm area of temporal parietal skull was dehiscent with exposed dura. In patient 6, the lateral semicircular canal was absent, the incus was hypoplastic with a remnant of the stapes superstructure not attached to the footplate, and the stapedial tendon attached directly to the long process of the incus. The stapedial muscle was lateral to the facial nerve. His facial nerve appeared to have a bifid course, with the superior branch passing lateral to the oval window and the inferior branch passing directly over the cochlear promontory. Intraoperative facial nerve stimulation confirmed these observations. Because of the location of his facial nerve, a cochleostomy could not be safely created, and the procedure was aborted. A full insertion of all active electrodes was achieved in patients 1 through 5.

According to the audiometric data before implantation, a pure-tone threshold response was obtained only at 0.5 kHz for patients 1, 2, and 4; no response was obtained at the other pure-tone frequencies. Patients 3 and 5 had no measurable responses at the maximum output limits of the audiometer to speech or tones. After implantation, patients 1 through 4 had thresholds in the mild hearing loss range. Frequency-specific thresholds after implantation were not consistently obtained for patient 5; however, a speech awareness threshold was obtained through behavioral observation at the 45-dB hearing level (Figure 3). Patient 6 did not receive an implant; therefore, the audiometric data are not included.

Speech perception scores after implantation, defined using the Early Speech Perception test categories, improved for 3 of the 5 patients receiving implants. Because of the variation in follow-up duration for the patients, results of the most recent examinations after implantation are reported (Figure 4).

Speech perception scores on the GASP after implantation were available for patient 1. Before implantation, patient 1 scored 0% correct on the GASP-W and the GASP-S. One year after implantation, the scores were 8% correct on the GASP-W and 0% correct on the GASP-S; at 2 years, 17% correct on the GASP-W and 20% correct on the GASP-S; and at 4 years, 50% correct on the GASP-W and 50% correct on the GASP-S. Results were not obtained on patients 3 and 4 because of their young age and were incomplete on patients 2 and 5 because of insufficient language development.

Parental questionnaires revealed that patient 3 scored 3 on the IT-MAIS before implantation and 31 at 6 months after implantation. Meaningful Auditory Integration Scale scores for patient 5 were 3 before implantation and 6 at 6 months after implantation. Meaningful Auditory Integration Scale and IT-MAIS data are not reported for patients 1, 2, and 4 because this questionnaire was not used as part of the St Louis Children’s Hospital standard evaluation battery until after these patients had received their implants.
Most of our patients with CHARGE association benefited from successful implantation. The decision to perform implantation in these children remains controversial. Some implant programs choose not to perform implantation in children with deafness who have multiple comorbidities because of potentially poor results. Other programs perform implantation in these children in anticipation of some benefit from long-term acoustic input.

Imaging provides a useful tool for preoperative evaluation of these patients. The internal cochlear structure, vestibular aqueduct, and internal auditory canals are especially well suited for imaging analysis. Although current high-resolution scans provide exquisite detail, fine structures such as the insertion point of the stapedial tendon may not be reliably demonstrated. Coronal imaging may improve imaging accuracy.

Approaching the middle ear through the mastoid and facial recess can be technically challenging in performing cochlear implantation. The absence of the lateral semicircular canal and ossicular anomalies can deprive the surgeon of important anatomic landmarks for identifying the facial nerve. In patients 5 and 6, we removed the incus buttress to improve exposure to the epi tympanum and provide a wider field of view, through which we were able to follow the facial nerve from the cochle-
tion of these procedures. Careful consideration should be given to the timing of cochlear implantation relative to other potential surgical procedures a child may need.

Results on speech perception tests improved in patients 1, 2, and 4. Patient 3 has significantly improved in responsiveness and use of sound, as measured by the IT-MAIS. Although patient 5 has shown no measurable benefit with the test battery used in this study, his parents report that he has an improved quality of life and is more curious about the world around him. This is an important point. Benefit denotes something that promotes well-being (ie, a useful aid). Our current battery of audiologic and perceptual testing provides the basis to demonstrate objective benefit from cochlear implantation. However, in children with significant cognitive, developmental, and physical comorbidities, our current standard test battery limits our ability to demonstrate objective benefit in these children. More subjective measures reported by parents and other caregivers often serve as early indicators of benefit.

Follow-up for most of our patients was short, limiting demonstration of objective benefit to our patients. Prior studies\textsuperscript{10,11} show that children with motor or cognitive delays develop improved speech perception more slowly than other children receiving implants; however, they progress in their speech and auditory skills over time. In support of this observation, patient 1 developed no open-set speech recognition after 1 year and only minimal recognition after 18 months; patient 3 at 6 months; patient 4 at 1 year (did not cooperated during the ESP test but completed 9 of 20 key words on the Mr. Potato Head Test); and patient 5 at 6 months.

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The opinions and assertions contained herein are the private views of the authors and are not to be construed as official or as reflecting the views of the US Department of Defense.

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CONCLUSIONS

Cochlear implantation in patients with CHARGE association is feasible. Variations in temporal bone anatomy can lead to increased technical challenges. Surgeons planning cochlear implantation in these patients should be knowledgeable about the potential variations to avoid inadvertent facial nerve injury. Parents need to be aware of the increased risks, particularly to the facial nerve, and the possibility of no or partial electrode implantation. Although individual outcomes may vary, all but 1 of the children in our series obtained measurable benefit. Because of variations in the degree of benefit these patients achieved after implantation, it is important to ensure that parents are counseled thoroughly and have appropriate expectations before proceeding with implantation.

REFERENCES