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Case Report

Cochlear implantation in Klippel Feil syndrome: pitfalls and outcomes

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ABSTRACT

Cochlear implantation surgery involves multidisciplinary teamwork aiming for electrode insertion, which is as atraumatic as possible with an optimal intracochlear electrode position. Congenital inner ear anomalies often co-exist with musculoskeletal anomalies in syndromic children and can challenge cochlear implant surgery. The authors present a case of Klippel-Feil syndrome with congenital torticollis with bilateral inner ear anomalies who underwent cochlear implantation with multiple intra-operative surgical challenges. Multidisciplinary cooperation between the anesthetists, auditory-verbal therapy team, and the developmental pediatrics team is essential for a good outcome.

Keywords: Klippel-Feil syndrome, Cochlear implantation, Inner ear malformations, Cochlear anomaly

INTRODUCTION

Klippel-Feil syndrome (KFS) is a genetic disorder of segmentation of cervical vertebrae. It affects approximately one in 40,000 live births and is considered one of the most common congenital anomalies of the cervical spine.[1] The classical phenotype consists of a webbed neck, a low posterior hairline, and a limited range of neck movements. Otolaryngologic manifestations are critical extra-skeletal manifestations of KFS, including bilateral or unilateral external, middle, and inner ear anomalies and airway challenges due to limited neck extension.[2] When cochlear implantation is planned for bilateral congenital profound sensorineural hearing loss, KFS presents significant challenges. A preoperative thorough evaluation includes an audiological workup and assessment by a developmental pediatrician to assess candidacy and milestones. The intraoperative period presents unique challenges, which are presented here.

CASE REPORT

A six-year-old girl was referred with complaints of speech delay and not responding to sounds since birth. The parents also noticed her neck turned to the left since birth. On examination, she had a high-arched palate, webbed neck, low posterior hairline, and gross scapular asymmetry (Figure 1). She had a neck tilt to the left with no palpable mass in the sternocleidomastoid. The spine showed kyphoscoliosis. She had no limb weakness and no neurological deficits.

Her perinatal and past history was unremarkable except for an episode of pneumonia at 8 months of age, for which the medication history was unknown. A repeat evaluation at our center revealed no replicable wave V peak, even at 90 dBHL in both ears, suggesting profound sensorineural hearing loss. Distortion product otoacoustic emissions were absent bilaterally. Tympanometry revealed a C curve on the right and a B curve on the left. She was treated medically for the same and had a normal middle ear function before surgery. A hearing aid trial using a GN Resound Magna 490 aid showed bilateral responses out of the speech spectrum. A speech assessment on her first visit revealed expressive and receptive language acquisition at three years. A preliminary evaluation by the developmental pediatrics team concluded that the child was cognitively able and would benefit from auditory rehabilitation, with sign

language being used in the interim under the care of a special educator. An X-ray of the neck and chest depicted multiple fusion segmentation anomalies involving the cervicothoracic ribs and vertebrae with distortion of lung fields and a rudimentary C4 with partial block C5-T1 and block T2-T3 vertebra. An MRI and CT of the brain with temporal bones showed, on the right side, a high jugular bulb reaching up to the round window, cochlear nerve hypoplasia, cochlear aperture stenosis, duplication of the internal auditory canal, dilated vestibule, and absent superior and posterior semicircular canal. The basal turn of the cochlea with a cochlear duct length (CDL) of 28.46 mm was visualized (Figure 1). The left side showed hypoplastic left temporal bone with the non-visualized cochlea, vestibule, superior semicircular canal, and vestibular aqueduct with an atretic internal auditory canal.

She was evaluated by the pediatric respiratory team and the infectious diseases team for preoperative fitness and vaccination, respectively. The pediatric orthopedic team advised surgical correction however, the parents declined it in the present admission.

After a thorough discussion with the radiologist, audiologist, speech therapist, and pediatrician, it was decided to proceed with a right cochlear implantation with a guarded prognosis for speech acquisition. Written informed consent was obtained for anesthesia risks, including the position for intubation and the need for backup airway measures during the lengthy procedure. Right cochlear implantation was performed with facial nerve monitoring. There were significant difficulties in positioning the patient because of the limited neck mobility.

Cervical fusion precluded adequate head position in a ring holder with suboptimal angulation during the initial bone work for cortical mastoidectomy and well for the receiver-stimulator. Ergonomics during routine cochlear implant surgery was modified, and the patient was strapped securely to the operating table to permit tilting of the table for adequate visualization during the posterior tympanotomy. Cochlear nucleus CI422 electrode was implanted via a mastoidectomy-posterior tympanotomy approach, and complete insertion of all electrodes was achieved through the extended round window approach. A high jugular bulb on the right impeded visualization.

However, meticulous drilling with facial nerve monitoring proved effective. Intraoperative neural monitoring revealed normal impedance (Figure 2). However, neural response telemetry detected responses only in the first electrode (Figure 3). The implant was switched on after 14 days of the procedure. The child underwent postoperative auditory verbal therapy for three months at our center. Follow-up telemetry at our institute showed responses in the first seven electrodes (Figure 3). She had no baseline auditory skills during that period, but auditory awareness was present. She improved to associating meaning to words, communicated kinship terms, comprehended and expressed body parts, and could follow one-step commands. She started expressing basic needs through single words and developed a vocabulary of around seven words. Speech intelligibility was 45%. She could inconsistently imitate heard words. On the categories of auditory perception scales (Revised 12-point CAP scale), she scored a 4. On the categories of auditory performance scale (CAP), she scored 4 out of 7 points. The parents declined genetic testing and workup and are on follow-up with speech therapy by video consultation. Her score on the APCEI (Acceptation, Perception, Comprehension, Expression, Intelligibility) profile was 5-3-1-1-2. At three months, her postoperative mapping responses were within the speech spectrum in the implanted ear.

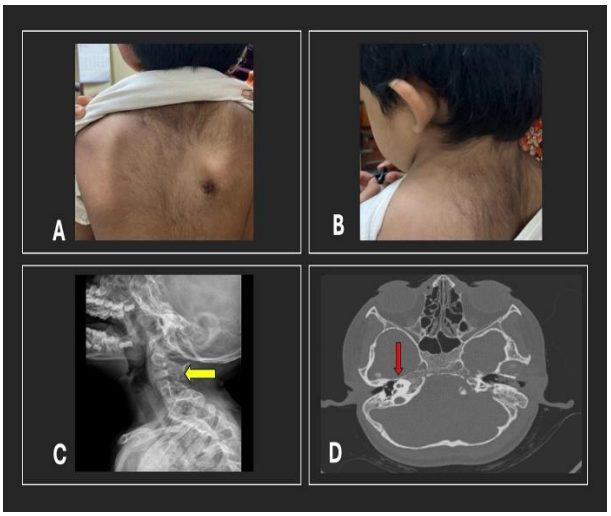

Figure 1: Clinical and radiological features of the case. (A) Scapular asymmetry in the patient. (B) Low posterior hairline and webbed neck suggestive of Klippel-Feil syndrome. (C) Lateral neck radiograph showing block cervical and thoracic vertebral fusion anomaly (Yellow arrow). (D) Axial section of high-resolution computed tomography of the temporal bone showing cochlear aperture stenosis and dilated vestibule (Red arrow).

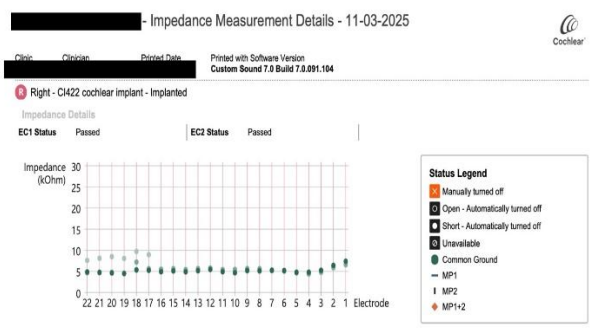

Figure 2: Intraoperative impedance obtained in all electrodes.

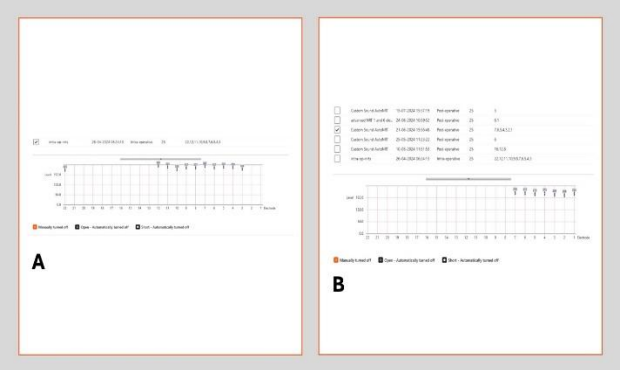

Figure 3: Neural response telemetry (NRT) data. The measured electrodes are mentioned. (A) Intraoperative NRT; (B) postoperative NRT at 2 months after electrode insertion.

DISCUSSION

Klippel-Feil syndrome is a challenge for otolaryngologists to rehabilitate surgically. First described in 1912, current classifications for the subtypes of KFS are as per cervical vertebral fusion. Associated described otolaryngologic manifestations include an absent or very short neck, malformed laryngeal cartilage with an anatomically difficult airway, and deafness. SNHL is most commonly reported, followed by mixed hearing losses.[3] The most familiar otologic procedure performed has been reported to be grommet insertion. Very few reports exist of cochlear implantation in KFS, which involves multiple anesthetic and surgical challenges.

Otologic surgery usually involves turning the operated ear away from the surgeon. In KFS, modifications included by the authors included turning the entire table to orient the microscopic axis and using an ergonomically designed chair to avoid surgeon fatigue. A high jugular bulb on the right with a rotated cochlea proved to be a challenge in the approach to the extended round window, and considerable caution must be exercised in an excessively tilted patient to judge the axis of drilling close to the RW.

The CI422 slim straight electrode with 22 half-banded electrode contact points spread across 20 mm was used, and complete insertion was obtained. The slim, straight electrode has a soft tip with an apical diameter of 0.3 mm and a basal diameter of 0.6 mm. Hypoplastic/aplastic cochlear nerves carry a poor prognosis regarding hearing perception and speech acquisition. Aplasia of the left side made surgery on the left untenable. Increased chances of a perilymph gusher are encountered in patients with dilated vestibules with an absent modiolus. A trans-mastoid labyrinthectomy is used for electrode insertion in common cavity malformations.[4] Various inner ear anomalies in KFS are reported in the literature: absence of the SCC, cochlea, absent or enlarged vestibule, and cochlear anomalies such as Mondini's dysplasia and Michel's aplasia.[5] However, cochlear implantation is rarely reported. In our case, a complete cochlea was radiologically identified on the right side. A normal CDL indicated a suitable neural substrate at least in one ear. Despite the complicated anatomy of the approach, the mastoidectomy posterior tympanotomy approach was considered more appropriate.

In the case of duplicated IAC, cochlear implantation is usually contraindicated due to often coexistent cochlear nerve aplasia.[6] Duplication occurs due to abnormal migration of the brain stem, hindbrain, and otic vesicles. However, no hindbrain anomalies were noted in our patient. There have been isolated reports of good cochlear implant outcomes in patients with KFS and Arnold Chiari malformations with perilymph leak4 and KFS with cholesteatomas.[7] No reports of cochlear implantation in a reasonably normal cochlea with other inner ear anomalies exist in KS.

CONCLUSION

CI in KFS is challenging and risky on many fronts. Each case has to be evaluated individually and hearing rehabilitation optimized. Surgical correction of the neck deformity may precede CI if it precludes safe surgery. To the best of the authors' knowledge, this is the third report on CI being successfully done in the case of KFS.

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