Cochlear implantation in branchio-oto-renal syndrome syndrome: Our experience with four patients

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Abstract

There still is inadequate literature regarding the surgical indications and postoperative outcomes of cochlear implantation (CI) in patients with Branchio-oto-renal syndrome; therefore, the aim of this study was to investigate whether CI surgery improved the hearing and speech performance of children with BOR syndrome-related severe hearing loss. Clinical manifestations, genetic results,audiological features, intraoperative findings and post-CI outcomes of four children were ascertained and analysed. All patients had bilateral profound HI before CI). The surgery of one patient with aural atresia and complex ear deformity was rather challenging, but the outcome after CI was satisfactory. However,auditory and speech performance of other three patients wasn't good enough, which was probably related to cochlear nerve deficiency.

Key Points

* BOR syndrome is a rare hereditary disease characterized by several congenital defects in multiple organs, including hearing impairment.

* Diagnostic criteria of BOR syndrome includes major and minor phenotypic.

* Cochlear implantation (CI) can potentially benefit BOR syndrome patients with profound hearing loss.

* The CI outcomes in BOR syndrome patients are influenced by dysplasia of cochlear nerve.

* Oblique sagittal MRI of internal auditory canal is of prognostic value in the management of BOR syndrome.

1 . INTRODUCTION

Branchio-oto-renal (BOR, OMIM 113650)syndrome is a rare autosomal dominant condition, first reported in 1975. BOR is also referred to as branchiootic syndrome(BO) when lacking renal malformation phenotype. It accounts for 2% of childhood inner ear deafness, affecting one in 40,000. The clinical features of BOR are variable. Chang et al. developed diagnostic criteria to including major and minor phenotypic features(1). There still is inadequate literature regarding the surgical indications and postoperative outcomes of cochlear implantation (CI) in patients with BOR syndrome. Therefore, the aim of this study was to investigate whether CI surgery improved the hearing and speech performance of four children with BOR syndrome related severe hearing loss.

2 . MATERIALS AND METHODS

Four children with BOR syndrome underwent CI were included in this study. Clinical manifestations, genetic results, audiological features and post-CI outcomes were ascertained and analyzed. Written informed consents were obtained from all the subjects and/or their parents. This study was approved by the research ethics committees of both the hospital and was conducted in accordance with the guidelines of the Declaration of Helsinki.

3. RESULTS

3.1 Clinical manifestations and genetic results

The clinical manifestations, genetic results in the four patients are shown in Table 1. The patients had no mental retardation or growth delay. All patients failed newborn hearing screening and were diagnosed with bilateral profound HI soon after birth. They all presented typical BOR syndrome with at least two major and two minor items according to the criteria by Chang et al [(1)]. Physical examination revealed preauricular pits in all cases but branchial anomalies just existed in Case 2 and 4 , who also had family history of hearing loss. Bone imaging studies using high-resolution computerized tomography revealed abnormal inner ear structures in all three patients. Three cases showed cochlear nerve dysplasia. Additionally, genetic examination was performed in all cases, case 2 using whole-exome sequencing, case 1,3,4 using Hereditary Hearing Loss panel assay (Mygenostics Corporation, Beijing, PRC). Pathogenic variants in the *EYA1* gene were identified only in case 4, which had been reported to be causally linked to BOR syndrome.

Early intervention with hearing aids was provided, and all the patient benefitted little from intensive aural rehabilitation.

3.2 Surgery findings

Five ears underwent CI using traditional retroauricular straight incision 3 3.5cm in length. Four ears were performed using conventional facial recess and round window approach(Table 2). Case 3 underwent simultaneous bilateral CI surgery. Left ear used retrofacial approach due to anteromedially displaced mastoid segment FN.

Individually, full electrode array insertion and excellent intraoperative electrically Evoked Compound action potential was achieved in right ear of Case 1. Case 2 had right aural atresia but normal cochlear nerve dimensions in the operation ear (Figure 1). Since the mastoid was completely sclerotic, and the semicircular canal was aplasia, only the dura and sigmoid sinus were used as anatomical landmarks during mastoidectomy. Drilling out of the superficial mastoid bone exposed a strip-like fibrous tissue resembling the vertical segment of the facial nerve, but facial nerve monitoring ruled out the possibility (Figure 1). The mastoid segment of the facial nerve was exposed gradually by removing the overlying bone with a fine diamond drill in the deep part of the mastoid, with the assistance of facial nerve monitoring. Covert round window niche was identified in front of the nerve and after removing the overhanging bone and fibrous tissue, the round window was exposed. 7 of the 12 electrodes were inserted through the enlarged round window. Congenital dehiscent facial canal of the tympanic and pyramid segment were confirmed in the right ear, which had facial paralysis postoperatively. The patient underwent a secondary exploratory operation 7 days later, and the main trunk of facial nerve demonstrated no reaction to monitoring probe. facial paralysis recovered to Brackmann Level I about 1 year later. Only partial insertion was achieved in left ear of case 4.

3.3 Audiological cochlear implantation outcomes

All four patients had bilateral profound HI. Early intervention with hearing aids was provided, but poor hearing was observed in all patients(Figure 2). Averaged auditory speech scores were rather low, Categories of Auditory Performance (CAP) = 1.5 , speech intelligibility rating (SIR) = 1.5, The (Infant Toddler)-Meaningful Auditory Integration Scale (IT-MAIS)= 8.25, Meaningful Use of Speech Scale (MUSS)=6.5 before surgery. Postoperatively, all patients received regular aural rehabilitation. CAP, SIR, IT-MIAS and MUSS scores were assessed to determine auditory and speech performances, which revealed improvements in all patients. Case 2 showed most satisfactory improvement (Figure 2). However, regarding SIR, and MUSS, cases 1, 3 and 4 showed relatively low scores even at almost 5 years after the surgery.

4 DISCUSSION

4.1 Synopsis of key findings

In this study, we detailed the clinical manifestations, genetic results and audiological features of four patients with BOR syndrome who accepted CI. Auditory and speech performance after CI may vary postoperatively.

In addition, the surgery of case 2 was most challenging as with aural atresia.

4.2 Comparisons with other studies

CI outcomes in patients with ROR syndrome were scarcely documented in the literature, because it accounts for only about 2% profound hearing loss[(1)]. Most surgical process reported in the literature seemed normal, no further details were provided[(2-5)]. A perilymphatic gusher was reported in one adolescent case with bilateral dilated vestibule, enlarged vestibular aqueduct, ossicular anomalies, and bilaterally deviated facial nerves. However, this was easily controlled by conventional methods[(3)].

But there were challenging case report due to ear deformity. Absent temporal bone structures including the lateral semicircular canal, incus and stapes, and oval windows were mentioned in previous case reports, then it took more time to expose the cochlea[(6)]. Patient with aural atresia is most challenging, because few anatomical landmarks were present and the dura might get exposed during mastoidectomy as intraoperative complications. Previous imaging study had showed that non-pneumatized mastoid, antrum and low lying tegmen were very common in BOR. Thus, restricted transmastoid access should be anticipated [(7)].

Furthermore, aberrant courses of facial nerve definitely makes the operation more difficult[(6)]. Residual soft tissue resembling the nerve definitely increased operating time, such as in case 2. Thus facial nerve monitoring were essential. It is important to not cut any soft tissue too hastily. Finally, malformed cochlea also made precise location of the scala tympany and insertion difficult. Full insertion was unattainable. Cochleostomy was sited more anterosuperior to the round window niche than usual as a result of rotated cochlea, such as in case 3[(6)]. Intraoperative navigation CT may be helpful with cochlear location in such patients.

Most previous studies reported promising outcomes no matter in pediatric[(4, 8)], adolescent[(3)] or adult patients[(5)]. Morisada reported 4 in 11 patients who didn't show good effect without details(8). In this study, we observe unfavorable CI outcomes in three patients. Dysplasia of cochlear nerve is the main reason [(9)], which was rarely mentioned before. Cochlear implant in children with CND, especially those with CN aplasia, has been controversial. Oblique sagittal high-resolution MRI is routinely performed to screen CND in most CI centers, although it's not the golden standard test due to limited resolution. Audiological effective cases indicate that certain children do benefit from CI despite of radiographic CND. Thus preoperative communication with CI candidates and their families are essential to help them establish reasonable expectations.

4.3 Strengths and limitations of the study

This study is the first to describe the correlation between CND and CI outcomes in patients with BOR syndrome. However, because of the limited number of cases (only four patients) in this study, our speculation is not conclusive. As BOR syndrome is a disease with extremely low prevalence, large-scale meta-analyses are essential to clearly delineate the correlation between CND, audiological features and CI outcomes.

5 . CONCLUSION

BOR syndrome patients with profound hearing loss could benefit from cochlear implantation. the surgery is challenging in the presentence of complex ear malformations. CI outcomes may be affected by dysplasia of cochlear nerve.

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