

Branchio-Oto-Renal (BOR) Syndrome-an uncommon form of congenital deafness

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Case report: Children with congenital deafness are common referrals to the Pediatric Otolaryngology unit of the Teaching Hospital. Branchio-Oto-Renal (BOR) Syndrome, an autosomal dominant syndromic form of deafness presents variably with the presence of auricular or preauricular pits in association with hearing loss, branchial sinuses and renal abnormalities ranging from renal hypoplasia to agenesis. Renal manifestations are least common and mostly missed. The present case report highlights a case of Branchio-Oto-Renal syndrome detected at the age of 5 years with unilateral renal agenesis and normal renal function.

Key words: Branchio-Oto-Renal syndrome, hearing loss, renal agenesis

Introduction

Branchio-Oto-Renal (BOR) Syndrome is an autosomal dominant form of congenital deafness associated with various combinations of pre-auricular sinus, branchial sinus and renal abnormalities. This is a rare condition with the birth prevalence of around 1:40,000 live births. In spite of many researches, exact genetic etiology is obscure till now. The Gene EYA-1 on Chromosome 8q13 has been put forward as the associated gene ¹. We are reporting this case as this is so far the first case report in the Nepalese literature.

Case Presentation

Five years old female child from Panchthar district of Nepal was brought to the ENT Outpatient department of our hospital by her father with the chief complaints of multiple openings on bilateral pre-auricular region and bilateral neck region with deformed left pinna since birth. Father also reported that she had decreased hearing noticed after one year of life which was non progressive. There were no complaints of ear discharge, earache, facial abnormalities or trauma of any kind in the past.

The child had no exanthemas in the past and was not admitted to hospital till date for any cause. She was the only child of that family. There are no similarly affected

known individuals in the family in both maternal and paternal side.

The mother had taken two doses of injections of tetanus toxoid during pregnancy. She had not taken any toxic or injurious drugs and she was not exposed to radiation in the prenatal period, either.

The child was full term vaginal delivery in the home. There were no immediate postnatal problems. The child is not allergic to medication. There were no pets at home.

Complete ENT and Head and Neck examination was performed. The child had bilateral pre-auricular sinuses and skin tags. Left pinna was cupped but the right was normal looking. Both the tympanic membranes were retracted and pinkish with decreased mobility on Seigelization. Tuning fork tests were difficult to carry out. The Facial nerve was intact. There was a pit over the right cheek of 5 mm by 5 mm in size. She also had bilateral branchial sinuses which were not infected. Rest of ENT-Head & Neck examination was found to be normal.

On systemic examination she was found to have prenatal cleft and macule over right buttock which was 3 by 3cm in size. Rest of the systemic examination was found to be normal.

Branchio-Oto-Renal (BOR) Syndrome

The child was investigated which showed normal hematological and biochemical parameters. Play audiometry revealed hearing loss of 45 dB at low frequencies, 75 dB at mid frequencies and 75 dB at High frequencies. Tympanogram showed bilateral "B" type curve without reflex. Fistulogram of all the sinuses was performed and found to have blind endings with no dye at the depth. Renal function tests were normal. Computerized Tomography Scan of abdomen and pelvis showed absent right kidney with malrotated left kidney.

Fig. 1: CT scan of Abdomen and pelvis showing absent right kidney and malrotated left kidney

The final diagnosis was made of Branchio-Oto-Renal (BOR) Syndrome.

Surgery was planned. She underwent pre anaesthetic check up and was found to be fit for surgery.

The child underwent excision of bilateral pre-auricular sinus, bilateral branchial sinus and pre-auricular skin tags with myringotomy and ventilation tube insertion on both the ears under general anesthesia.

There were bilateral pre-auricular sinus 2cm deep involving ascending crus of helix. Branchial sinus on the right side had extended 3cm. from outside going in between external and internal carotid arteries and inner extent was difficult to identify. The left was superficial. On Myringotomy, there was thick glue present on the right middle ear and thin serous fluid in the left side.

The postoperative period was uneventful. Stitches were

removed on seventh post operative day and the child was sent home. Both the ventilation tubes were in situ and patent at the time of discharge.

Fig 2: Post-operative photograph of the child

There was no hearing improvement after surgery and in the follow up. She was advised for high gain hearing aid which was found to be beneficial for her.

Discussion

Branchio-Oto-Renal syndrome (BOR) is an autosomal dominant disorder with variable clinical manifestations affecting branchial, renal and auditory development.¹ Mutations in the EYA1 gene on the chromosome band 8q13.3, the human homologue of the *Drosophila* eyes absent (EYA) gene, have been identified to be the underlying genetic defects of the syndrome.²

Genetic deafness is a common cause of congenital deafness. Most commonly it presents as sensorineural (50%) but Conductive and Mixed loss are not uncommon. Usually it presents as a single gene disorder. The cumulative incidence is 1:2000. BOR syndrome is an uncommon form of congenital deafness with variable penetration.

The symptom complex can be divided as Branchial arch abnormalities, Otologic and Renal abnormalities.³

Branchial arch abnormalities present as branchial cleft/cervical fistulae that occur in 50% of patients.

Otologic abnormalities present as Hearing loss of conductive, sensorineural and mixed varieties. Abnormalities of pinna present as pre-auricular sinus, lop ear deformity and pre-auricular tags. Atresia /stenosis of external auditory canal can occur. There might be malformation, malposition, dislocation or fixation of the ossicles. Cochlear hypoplasia (Mondini type), enlargement of cochlear and vestibular aqueducts and hypoplasia of lateral semicircular canal occur as the inner ear abnormalities. High gain hearing aid and speech therapy are the treatment for hearing loss.

Renal abnormalities occur in unilateral, bilateral or in combination forms. Renal agenesis, hypoplasia, dysplasia, ureteropelvic junction obstruction are common. Calyectasis, pelviectasis, hydronephrosis are the uncommon ones.

Severe malformations can cause pregnancy loss and neonatal death. Renal abnormalities are the least reported and mostly missed.

Conclusion

A rare case of BOR syndrome is presented. Management of such a rare case is highlighted.

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