Branchio-oto-renal Syndrome
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Abstract
Branchio-oto-renal (Melnick-Fraser syndrome) is a rare autosomal dominant disorder characterized by syndromic association of branchial cysts or fistulae along with external, middle & inner ear malformations and renal anomalies. Authors are reporting a 19 year male patient, who presented with profound deafness & low set “lop-ear” with right sided preauricular pit. USG abdomen revealed agenesis of the left kidney.

INTRODUCTION
Branchio-oto-renal (BOR) syndrome refers to an autosomal dominant disorder occurring in approximately 1:40,000 new born infants. BOR syndrome is associated with major clinical findings of branchial cysts or fistulae, external ear malformation and/or preauricular sinus, various types of hearing loss and renal anomalies ranging from mild, asymptomatic hypoplasia or dysplasia to complete agenesis of kidney. Phenotypic presentation of BOR syndrome is extremely variable.1

CASE REPORT
A 19 years old male patient presented to E.N.T out door for medical handicap certificate for deafness. Patient had normal speech development. His family & birth history were insignificant. On examination both auricles were low set and “lop-ear” in shape (Fig. 1). Bilateral tympanic membranes were intact on otoscopy. Further examination revealed left sided preauricular sinus (Fig. 2). Facial nerve was intact on both sides on clinical examination. On audiological assessment BERA (Brainstem Evoked Response Audiometry) waves were absent on left side even at 100 db, while on the other side patient responded at 53 db. Our patient had no branchial cyst or sinus. Ultrasonography abdomen revealed right kidney to be normal in shape and size with left renal agenesis. Subsequently the patient was diagnosed as a case of Branchio-oto-renal syndrome. As our patient had major complaint of hearing disability rather than preauricular sinus or renal agenesis, he was advised to use hearing aid on left ear and he responded well to the aid.

DISCUSSION

The Branchio-oto-renal syndrome is an infrequent but well described entity that combines deafness, early onset renal failure together with branchial clefts & pre auricular pits. Among profoundly deaf children 2% are diagnosed to have BOR syndrome.2 EYA 1, the human homologue of the drosophila eye absent gene plays an important role in the development of BOR syndrome. EYA 1 gene maps on 8q13.3 chromosome and expresses very early, between 4th & 6th weeks of human embryogenesis.3 Deafness relates to abnormalities in the three ossicles of the middle ear derived from the first and second branchial arches, while the branchial fistulae relates to second, third and fourth arches. EYA 1 gene is strongly expressed in the human embryonic kidney and in BOR syndrome there is fault between the ureteric bud and metanephric mesenchymal mass as the ureteric bud branches into renal parenchyma, resulting in renal anomalies.4

Deafness is the most common presenting symptom reported in 90% of cases which can be conductive (30%) or sensorineural (20%) but is most often mixed (50%).5 Pre auricular pits are present in 70% to 80% patients and sometimes can be the only external ear finding while 30% to 60% patients with BOR syndrome have external ear anomalies in the form of microtia to small lop or cupped ears with over folded superior helices.6 Middle ear anomalies include ossicular malformations (fusion, displacement, and underdevelopment), facial nerve dehiscence, absence of oval window and reduction in the size of middle ear cleft and inner ear anomalies include cochlear hypoplasia or dysplasia.7 Enlargement of the cochlear or vestibular aqueducts may be seen8 and there may be hypoplasia of the lateral semicircular canal.7 Structural kidney anomalies seen in 12% to 20% patients, includes unilateral renal agenesis with contralateral hypodysplasia or bilateral hypodysplasia that lead to end-stage renal disease (ESRD). Less common findings are preauricular tags (13%), lacrimal duct aplasia(11%), short palate(7%), retrognathia, euthyroid goitre and facial paralysis. BOR syndrome often may be confused with Alport’s syndrome, characterised by

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deafness, chronic renal failure, anterior lenticonus & other eye disorders. Anterior lenticonus, a conical protrusion of the central portion of lens in to the anterior chamber is pathognomonic for Alport’s syndrome. Retinal changes of perimacular flecks are also found in 35% cases of Alport’s syndrome. Other uncommon ocular lesions include recurrent corneal ulceration and corneal endothelial vesicles. Deafness in the Alport’s syndrome manifests at a later age.4

REFERENCES