End-stage renal disease with deafness in an adolescent child

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A 15-year-old male child was referred to the nephrology clinic for persistently elevated serum creatinine (11.2 mg/dl). He had initially presented with pallor, fatigue and difficulty in hearing. At one year of age he had undergone bilateral end uretersotomy for bilateral hydronephrosis with Grade IV refluxing ectopic ureters draining into posterior urethra. There was also a history of neck surgery at eight years of age for an infected cystic neck lesion. Family history was unremarkable. On examination he was noted to have bilateral microtia, right accessory auricle, right branchial fistula and left lower motor neuron facial palsy [Figure 1]. Investigations confirmed severe chronic kidney disease with acidosis, anemia and metabolic bone disease. Audiological investigation revealed moderate sensorineural deafness with normal internal auditory canals and bony labyrinth as per computed tomography. Ultrasound showed a small right kidney with moderate hydronephrosis. Left kidney was not visualized.

A diagnosis of Branchio-Oto Renal (BOR) syndrome was made as he satisfied the clinical criteria [Table 1] postulated by Chang *et al*.[1]

Branchio-oto renal (aka Melnick-Fraser syndrome) is a rare autosomal dominant condition with variable phenotypic expression due to incomplete penetrance seen in about one in 40,000 population.[2,3] The human homolog of the Drosophila eye absent (EYA-1) gene

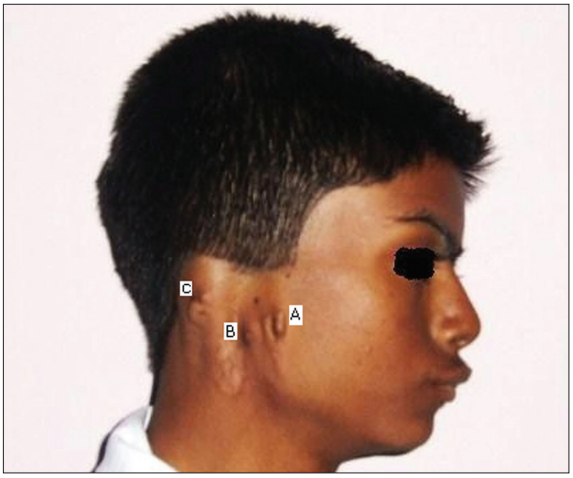

Figure 1: Image of the child showing (A) preauricular tag, (B) branchial fistula and scar from previous operation and (C) right microtia

present on 8Q 13.3 chromosomes plays an important role in the development of the BOR syndrome.[4] EYA-1 mutations are seen in 40% of cases meeting the criteria of BOR.[1] Renal and urological anomalies in BOR can be varied and include renal agenesis, hypoplasia, dysplasia, ureteropelvic junction obstruction, calyceal cysts and diverticula, pelviectasis, hydronephrosis and vesicoureteric reflux and or bifid kidney with double ureter.[1,3,4] Six percent of affected individuals can progress to end-stage renal disease. Deafness is the most common presenting symptom and could be conductive (30%), sensorineural (20%) or mixed (50%). Other ear anomalies include preauricular pits, microtia, middle ear anomalies which include ossicular malformation, facial nerve dehiscence, absence of oval window and reduction in the size of the middle ear cleft. Inner ear anomalies include cochlear hypoplasia or dysplasia. Branchial fistula are common and are related to abnormalities of the second, third and fourth branchial arches.

Children with Alport syndrome can also present with chronic kidney disease but in contrast to BOR, deafness manifests at a later age and they can also have eye anomalies like anterior lenticonus or perimacular flecks in the retina.

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Table 1: Criteria for diagnosis of Branchio oto renal syndrome (BOR)

Major criteria	Minor criteria
Deafness*	External ear anomalies*
Preauricular pits*	Middle ear anomalies
Branchial anomalies* (i.e. cysts and clefts)	Inner ear anomalies
Renal abnormalities*	Preauricular tags*
	Facial asymmetry*, high arched palate

Note: (a) Three major or two major and two minor criteria are required to diagnose BOR syndrome or one major criterion and affected first-degree relative meeting criteria for BOR syndrome, (b) *Clinical features present in our case.

As BOR syndrome can have varied presentation a high index of suspicion is required and the physician should be aware of the association of renal anomalies, particularly in patients with branchial fistula with or without ear anomalies and or history of hearing loss.

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