Melnick-Fraser Syndrome

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The branchio-otorenal (BOR) syndrome is an autosomal dominant disorder characterized by preauricular pits or branchial fistulas, hearing loss and renal anomalies.

Case Report

An 8-year-old girl had small bilateral pits on the concha of her auricle at birth (fig. 1). When she was 5 years old, her parents noticed a progressive hearing loss, and at the age of 7 years the audiological examination showed moderate sensorineural hearing loss in the right ear and severe hearing loss of 25%, especially for high-frequency sounds, in the left ear. Hypoplasia of the left kidney was identified by an intravenous pyelogram (fig-2).

Rachischisis at the level of the 5 lumbar vertebrae was present. The girl had a normal 46 XX karyotype, normal stature and a she has a palatum ogivale. There between the parents; no similar

normal intelligence index; was consanguinity case was known in the family.

Fig. 1. Small pit on the concha of the patient's left auricle.

Fig. 2. Melnick-Fraser syndrome. Hypoplasia of the left kidney identified by intravenous pyelogram.

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The BOR syndrome was reported by Melnick in 1975 [1]. It is characterized by three major anomalies: structural abnormalities of the external, middle and inner ear, ear pits and preauricular fistulas; hearing loss; renal abnormalities including agenesis, aplasia, hypoplasia and ureteral anomalies.

Other anomalies can also be present: facial asymmetry, lacrymal duct stenosis, palate abnormality; small, low-set ears bilaterally or unilaterally. The hearing loss may be sensorineural in 25% , mixed in 50% or conductive in 25% [2].

Recent reports suggest at least three separate syndromes that include abnormalities of the branchial arches and hearing impairment together with renal abnormalities: (1) the BO syndrome with no renal involvement; (2) the BOR syndrome with abnormalities of all three systems; (3) the

BOU syndrome in which the renal anomalies are limited to duplication of the collecting system and bifid renal pelvis [3, 4].

Heimler and Lieber [5] suggested that the BOU and BO syndromes represented variable manifestations of the BOR gene. The prevalence of the BOR syndrome is approximately 1/40,000 [6].

The BOR syndrome seems more frequent as indicated in textbooks and the current literature as it may affect about 2% of all deaf-born children. Deafness appears in 80% , ear pits and preauricular fistulas in 75% and renal involvement is found in 20% of the cases. Today all authors accept that the BOR syndrome is an autosomal dominant disorder [7, 8] manifested by various combinations of preauricular pits or lacrymal duct stenosis, but this disorder has a reduced penetrance and variable expressivity [5].

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