Branchio-oto-renal (BOR) syndrome is an autosomal dominant disorder with branchial, otologic, and renal manifestations.

The medical management of BOR syndrome requires medical care include audiologic, otologic, head and neck, urologic, and genetic evaluation. The recognition of hearing impairment is important, and children with the phenotypic characteristics of BOR syndrome should undergo thorough audiologic testing as early as possible.

**Introduction**

BOR syndrome is characterized by the following 3 essential clinical features: (1) hearing loss with structural defects of the external, middle and/or inner ear; (2) second branchial arch defects; (3) renal anomalies, ranging from mild hypoplasia to aplasia, which can lead to varying degrees of renal failure. The general prevalence of BOR syndrome is 1 in 40,000 people, and it occurs in 2% of profoundly deaf children. The branchiogenic origin of BOR syndrome can cause a wide range of anatomic malformations.

**Methods:**
Retrospective analysis of patients with BOR syndrome.

**Results**

We present 3 cases with BOR syndrome. In first case, we report a family, with five generations, with different phenotype expression of BOR syndrome.

**Table 1:** Clinical features in family with BOR. R – right, L – left, plus sign – present, minus sign – not present, NA – not available.

The disease is represented by various phenotypic expressions. Three causative genes for BOR syndrome have been reported: EYA1, SIX1 and SIX5, but the causative genes for approximately half of all BOR patients remain unknown. Presented patients did not have genetic testing; it was not available.