

## Branchio-oto-renal (BOR) 증후군

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### A Case of Branchio-oto-renal (BOR) Syndrome

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**Introduction:** Branchio-oto-renal (BOR) syndrome is a rare autosomal dominant disorder, associated with branchial fistulae or cysts, preauricular pits, ear malformations, hearing impairment, and renal anomalies. The incidence of BOR syndrome is approximately 1:40,000 and may account for 2% of children with profound hearing loss. 30–40% of patients with a BOR phenotype were observed to have a pathogenic mutation in EYA1 gene on chromosome 8. Approximately 2% of cases of BOR syndrome are due to mutations in SIX1 and SIX5. Phenotypic presentation of BOR syndrome is extremely variable. 12% to 20% patients have renal anomalies. Renal anomalies can be unilateral or bilateral and occur in any combination. The following anomalies include renal agenesis, hypoplasia, dysplasia, ureteropelvic junction obstruction, calyceal cyst, pelviectasis, hydronephrosis and vesicoureteral reflux. We report a case of BOR syndrome.

**Case:** A 18-year old woman presented to ENT outpatient clinic of our hospital with skin pit and swelling on right anterior neck from birth. Physical examination showed bilateral preauricular pits. Pure tone audiometry (PTA) showed conductive hearing loss. Neck computed tomography (CT) showed about 2cm sized cystic lesion in the right middle neck. Temporal bone CT showed hypoplasia in the both cochlea, vestibule, lateral semicircular canal and both short and wide internal auditory canal. Abdominopelvic CT revealed right ureteropelvic junction stricture and marked hydronephrosis of right kidney. Her blood urea nitrogen and creatinine levels were normal but urinalysis showed 1+ proteinuria. Family history revealed that her grandmother, father and uncle had jugular pit and the uncle had severe hearing loss. The patient underwent surgical excision for branchial cyst on the right side of anterior neck. Her blood chemistry including renal function was normal at 2 days after operation. She was discharged from the hospital without any complication.

**Key Words:** Branchio-oto-renal (BOR) 증후군  
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