Shaping Tomorrow's Nephrology: Insight-Driven Kidney Care
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Branchio-oto-renal syndrome

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Case Study : Branchio-oto-renal (BOR) syndrome is a rare autosomal dominant disorder caused by pathogenic variants in EYA1, SIX1, or SIX5, which affect the branchial arches, auditory system, and kidneys. Diagnosis integrates clinical criteria that require major criteria such as branchial anomalies, hearing loss, or preauricular pits, as supported by genetic testing. Gene-targeted testing confirms typical BOR syndrome phenotypes, whereas comprehensive genomic testing identifies atypical cases. Management is tailored to the individual phenotypes. Kidney anomalies, the most important prognostic factor, necessitate early nephrology referral and interventions such as pyeloplasty for ureteropelvic junction obstruction or kidney replacement therapy for kidney failure. Hearing loss is addressed using hearing aids, surgical correction, or cochlear implantation, depending on the type and severity of hearing loss. Branchial anomalies including cysts and sinuses can be managed conservatively or surgically when symptomatic. Genetic counselling plays a critical role, given the 50% inheritance risk and the implications for family planning. Prenatal ultrasonography is recommended for detecting renal anomalies during at-risk pregnancies. Management of BOR syndrome requires a multidisciplinary approach that integrates otolaryngology, nephrology, and genetics. Advances in genomic technologies and a deeper understanding of phenotypic variability are essential for improving the outcomes of patients with BOR. Herein, we describe a 29-month-old child who presented with preauricular pits, branchial fistulas, branchial cysts, malformation of inner, meddle, and outer ear, hearing loss, and kidney abnormalities, and confirmed by pathogenic variant in EYA1 gene.