Branchio-Oto-Renal Syndrome: Detection of EYA1 and SIX1 mutations in five out of six Danish families by combining linkage, sequencing and MLPA analyses

The branchio-oto-renal (BOR) syndrome is an autosomal-dominant disorder characterized by hearing loss, branchial and renal anomalies. BOR is genetically heterogeneous and caused by mutations in EYA1 (8q13.3), SIX1 (14q23.1), SIX5 (19q13.3) and in an unidentified gene on 1q31. We examined six Danish families with BOR syndrome by assessing linkage to BOR loci, by performing EYA1 multiplex ligation-dependent probe amplification (MLPA) analysis for deletions and duplications and by sequencing of EYA1, SIX1 and SIX5. We identified four EYA1 mutations (c.920delG, IVS10-1G>A, IVS12+4A>G and p.Y591X) and one SIX1 mutation (p.W122R), providing a molecular diagnosis in five out of the six families (83%). The present, yet preliminary, observation that renal and temporal bone malformations are less frequent in SIX1-related disease suggests a slightly different clinical profile compared to EYA1-related disease. Unidentified mutations impairing mRNA expression or further genetic heterogeneity may explain the lack of mutation finding in one family despite LOD score indications of EYA1 involvement.
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