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FGF3-Related Phenotypes: A Study of LAMM Syndrome and Otodental Dysplasia Patients with Two Novel Mutations in *FGF3* Gene

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ABSTRACT The fibroblast growth factor (FGF) signaling pathway regulates the intracellular events which are involved in the proper formation of the internal organs and limbs during the earliest stages of embryonic development. Here, the researchers performed a detailed examination of clinical and radiological findings from syndromic cases with sensorineural hearing loss and determined their molecular genetic etiology. Family history, physical examination, laboratory and radiological examinations for three Turkish families displaying congenital sensorineural hearing loss, microtia, dental anomalies and neuromotor developmental delay were evaluated and molecular analysis of the *FGF3* gene was performed. The researchers detected a heterozygous deletion of a 2.4 Megabase (Mb) segment in the region spanning 68,734,891 to 71,538,481 bases in the chromosome 11q13.3-q13.4. Interestingly, this region included the *FGF3* gene in case 1, whereas two novel mutations (NM_005247: c.8T>G, p.Leu3Arg, NM_005247: c.324+2T>C) were identified in case 2 and case 3 respectively. From this study, the researchers conclude that in the absence of inner ear structures in cases of syndromic hearing loss, *FGF3* gene related phenotypes should be considered and the *FGF3* gene should be examined by sequence analysis and array-CGH methods for both point mutations and gross deletions.