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Enamel Structure Defect, the Rare Dental Anomaly: Isolated or Syndromic

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Abstract : Enamel, the outermost layer of the tooth crown, is the hardest dental tissue and serves as a protective barrier. Amelogenesis, the process of enamel formation, is regulated by multiple genes to ensure normal, defect-free enamel. Defective enamel manifests as hypoplasia or as amelogenesis imperfecta (AI), which may occur in isolation or as part of a syndrome. This study presents 29 patients from 18 unrelated families (16 females and 13 males) who exhibited distinctive enamel abnormalities. We conducted thorough clinical examinations and requested laboratory and radiological investigations. Blood samples were collected for molecular analysis, utilizing a targeted panel for known AI variants and whole exome sequencing for unknown variants. Eleven variants linked to enamel anomalies were identified: four genes associated with isolated AI (WDR72, ACP4, SLC24A4, and FAM83H) and seven associated with syndromic forms, including enamel renal syndrome (FAM20A), tricho-dento-osseous syndrome (DLX3), Jalili syndrome (CNNM4), and others linked to neurological and mitochondrial disorders, skeletal dysplasia, and peroxisome disorders. Abnormal oral and dental phenotypes in individuals may indicate serious inherited disorders. Enamel defects have significant implications for aesthetics, function, and patients' psychological well-being. Dental examination, alongside clinical and molecular investigations, is crucial for the accurate diagnosis and prediction of inherited conditions.

Keywords : amelogenesis imperfecta, enamel defect, Enamel renal syndrome, DLX3, Jalili syndrome, WDR72, FAM83H, whole exome sequencing
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