FAM20A Mutation in a Patient with Enamel-Renal-Gingival Syndrome: A Case Report

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Abstract:

Objectives: Amelogenesis imperfecta with Gingival fibromatosis syndrome (AIGFS), amelogenesis imperfecta with nephrocalcinosis or Enamel-Renal syndrome (ERS), and Enamel-Renal-Gingival syndrome have been associated with mutations in the *FAM20A* gene. A number of cases in the literature have described patients with three important findings, including amelogenesis imperfecta (AI), gingival fibromatosis and nephrocalcinosis. This study was aimed to identify *FAM20A* mutations in an 11-year-old Turkish male affected with enamel-renal-gingival syndrome.

Methods: Clinical and radiographic examinations and mutational analysis of the coding exons of *FAM20A* gene were performed.

Results: The patient was the first child of non-consanguineous parents. Oral examination revealed AI and generalized gingival fibromatosis. A panoramic radiograph showed generalized absence of enamel, delayed eruption of permanent teeth, intrapulpal calcification and multiple unerupted teeth. No calcification was observed with renal ultrasound. Mutation analysis of *FAM20A* revealed a novel missense mutation in exon 10 (NM_017565.3: c.1307G>A; g.61999G>A; p.Gly436Glu). This is notable because G^{436} is highly conserved among *FAM20A* homologues.

Conclusions: Our study reports a novel *FAM20A* mutation and confirms that AIGFS and ERS actually are the same entity with different manifestations. Patients with AI, hypoplastic type with unerupted teeth and gingival fibromatosis are advocated to have renal ultrasonography to rule out nephrocalcinosis or nephrolithiasis.

Keywords: *FAM20A*, Amelogenesis imperfecta, Gingival hyperplasia, Nephrocalcinosis

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