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Original Article

No Evidence for Association between Amelogenesis Imperfecta and Candidate Genes

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Abstract:

Background: Amelogenesis imperfecta (AI) is an inherited tooth disorder. Despite the fact that up to now, several gene mutations in *MMP20, ENAM, AMELX* and *KLK4* genes have been reported to be associated with AI, many other genes suggested to be involved. The main objective of this study was to find the mutations in three major candidate genes including *MMP20, ENAM* and *KLK4* responsible for AI from three Iranian families with generalized hypoplastic phenotype in all teeth.

Methods: All exon/intron boundaries of subjected genes were amplified by polymerase chain reaction and subjected to direct sequencing.

Results: One polymorphisms was identified in KLK4 exon 2, in one family a homozygous mutation was found in the third base of codon 22 for serine (TCG>TCT), but not in other families. Although these base substitutions have been occurred in the signaling domain, they do not seem to influence the activity of KLK4 protein.

Conclusion: Our results might support the further evidence for genetic heterogeneity; at least, in some AI cases are not caused by a gene in these reported candidate genes.

Keywords:

Amelogenesis Imperfecta ، *KLK4* ، *Polymorphism* ، *Consanguineous marriages*

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