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Molecular basis of non-syndromic tooth agenesis: Analysis of PAX9 gene in Slovak family

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Tooth agenesis is a common development anomaly in humanswhich includes two types, syndromic or non-syndromic. Genetic and environmental factors may be of etiologic importance considered that complex interactions between genetic, epigenetic and environmental factors caused dental anomalies. Several candidate genes associated with non-syndromic hypodontia have been described. In the current study we performed analyses of PAX9 gene in Slovak family with severe tooth agenesis. Genomic DNA was extracted from buccal swabs (Relia-Prep®, Promega). Sequence analysis was carried by ABI 3500xL Genetic Analyzer with terminator chemistry Big Dye3v1 (Applied Biosystems). Five exons, exon/intron junctions and UTRs of PAX9 gene were sequenced. Mutation detection was performed using reference sequence for PAX9 (GenBank NG_013357. 1) available in NCBI database. In the analysed family we identified six polymorphismsof PAX9 gene. In 5UTR´ region cytosine insertion 99insC was detected in proband (III/1), brother (III/2) and father (II/1). In family members with cytosine insertion C272G substitution (rs4904155) was found. In coding region we detected the substitution of bases G1444C causes a change of amino acid alanine to proline (Ala240Pro) in three affected and one non-affected member in the analysed family. The silent mutation CAC→CAT (His239His) was observed in exon 4. In analysed members of the family we identified variants in 3´UTR region: a new variant G2452A and a rare variant G2307C (rs72679753). Identified variantsdetected in our study were not causal. The identification of causal mutations in hypodontia candidate genes should improve understanding of genetic basis of tooth development.

Biography

Iveta Boroňová is a specialist in human genetics with sixteen years experiences in laboratory practice. She has completed her Ph. D. from University of Prešov in 2006 and habilitation in Anthropology from the same university in 2010. Her actual position is the Head of Department of Biology, Faculty of Humanities and Natural Sciences, University in Prešov, Slovakia. She is a member of European Cytogenetic Association and Anthropological companies. She is the author of monographs, textbooks and a wide range of original scientific papers in national and international journals and currently the field of her research activities is molecular genetics.

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