

Analysis of MSX1 and PAX9 genes in patients with non-syndromic hypodontia

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Hypodontia is agenesis of one to six teeth, and it represents the most common orofacial deformity in humans. The tooth development is very complicated and complex process. Research of the molecular basis of hypodontia is based on the detection of mutations in genes that affect the process of odontogenesis. Several candidate genes associated with isolated hypodontia have been described. The most common causes of hypodontia are mutations in transcription factors, such as the homeobox gene MSX1 and paired box gene PAX9. In this study, DNA samples of 19 patients, aged 9 to 25, with different types of tooth agenesis were analyzed. The agenesis of lateral incisors in the first and second quadrant has occurred more frequently (80, 43%) than agenesis of second premolars present in the third and fourth quadrant (23, 21%). On average, patients were missing two teeth. In one patient microdontia of lateral incisors in the first quadrant was present. In 52% of patients familial tooth missing (dental agenesis occurred in at least one of the two parents) was confirmed. Exons, exon/intron junctions and UTRs of MSX1 and PAX9 gene were sequenced in genomic DNA. Sequence analysis of candidate genes MSX1 and PAX9 showed the presence of several polymorphisms. Genomic DNA was isolated from buccal swabs. DNA sequence analysis was performed by the 24-capillary 3500xl Genetic Analyzer (Life Technologies). Sequences were evaluated by SeqScapeSoftware and Sequencing Analysis Software (Life Technologies) and compared with reference sequences. The detected variants have been described in relation to the pathogenesis of hypodontia.

Biography

Jarmila Bernasovska studied biology at Pavol Jozef Šafarik University in Košice, Slovakia. She is expert on molecular genetic methods, focusing on clinical genetics. Her research at University of Presov focused on the analyses of mutations in candidate genes of selected genetic diseases in Slovak population including minorities. In the present time she is director of Excellence Centre of Animal and Human Ecology. She has published more than 150 papers in reputed scientific journals and is the author of three monographs. She has long teaching experience and experience in laboratory medicine.

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