Abstract

The tooth development (odontogenesis) is a complicated and dynamic process involving many proteins. They interact with each other to create a complex signaling network. Irreversible agenesis of the teeth (hypodontia) may occur by disrupting the necessary balance. This congenital absence of one or more teeth may have a genetic cause and/or may be caused by environmental factors (drugs, trauma etc.). *PAX9*, *MSX1*, *AXIN2*, *EDA*, *EDAR* and *WNT10a* genes are most frequently mentioned in the literature.

This work deals with odontogenesis in biological and molecular aspects in its introduction and describes results of mutations and polymorphisms of selected genes in patients and control subjects of Czech population. Gene regions have been studied by molecular biology techniques – capillary sequencing and next-generation sequencing (NGS).

The results of this study show that the most missing teeth in the patient group were the third molars in both jaws, followed by the second premolars in the lower and upper jaws and lateral incisors in the upper jaw. The most significant genetic results include the g.9527G>T mutation in the splice site of the *PAX9* gene and g.8177G>T mutation in the *MSX1* gene, which leads to a stop codon. Both heterozygous substitutions have been identified in probands suffering from oligodontia, the absence of 6 or more teeth in the dentition. These two mutations, together with the oligodontia signs, were also observed in the families of affected subjects. Other mutations and polymorphisms found in this study were not associated with tooth agenesis on selected groups of patients and control subjects in the Czech population.