A review on non-syndromic tooth agenesis associated with PAX9 mutations

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Abstract

Tooth agenesis in the reduction of tooth number which includes hypodontia, oligodontia and anodontia is caused by disturbances and gene mutations that occur during odontogenesis. To date, several genetic mutations that unlock the causes of non-syndromic tooth agenesis are being discovered; these have been associated with certain illnesses because tooth development involves the interaction of several genes for tooth epithelium and mesenchyme odontogenesis. Mutation of candidate genes PAX9 and MSX1 have been identified as the main causes of hypodontia and oligodontia; meanwhile, AXIN2 mutation is associated with anodontia. Previous study using animal models reported that PAX9-deficient knockout mice exhibit missing molars due to an arrest of tooth development at the bud stage. PAX9 frameshift, missense and nonsense mutations are reported to be responsible; however, the most severe condition showed by the phenotype is caused by haploinsufficiency. This suggests that PAX9 is dosage-sensitive. Understanding the mechanism of genetic mutations will benefit clinicians and human geneticists in future alternative treatment investigations. (C) 2017 The Authors. Published by Elsevier Ltd.

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