PAX-9 polymorphism may be a risk factor for hypodontia: a meta-analysis

W. Zhang, H.C. Qu and Y. Zhang

Department of Orthodontics, Hospital of Stomatology, China Medical University, Shenyang, Liaoning, China

Corresponding author: Y. Zhang
E-mail: zgykdxzhangyang@126.com

ABSTRACT. To evaluate the association between paired box 9 (PAX9) gene polymorphisms and tooth agenesis in isolated humans, we performed a comprehensive meta-analysis. We examined 6 case-control studies, with a total of 855 hypodontia cases and 1201 healthy controls. The G allele and G carrier (AG + GG) of A1031G were positively associated with hypodontia susceptibility. Similarly, the T allele and T carrier (CT + TT) of C912T and rs12881240 in the PAX9 gene also indicated an increased risk of hypodontia. In addition, the C allele and C carrier (CG + CC) of 718C, IVS2-109, rs4904210, and rs7143727 showed no significant association with oligodontia. The G allele and G carrier (AG + GG) of IVS2-41 in the PAX9 gene were not related factors. Interestingly, the genotype (AG + GG) of IVS2-54 in the PAX9 gene may be a protective factor for oligodontia (odds ratio = 0.21, 95% confidence interval = 0.07-0.63, P = 0.005). However, no significant differences were found in the allele frequency of IVS2-54 in the PAX9 polymorphism between controls and subjects with sporadic tooth agenesis. In conclusion, our meta-analysis results revealed 4 genetic sites of the PAX9 gene involved in hypodontia cases, of which 3 sites may be risk factors and 1 may have a protective role.

Key words: Genetic polymorphism; Hypodontia; Meta-analysis; PAX9