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用先天性缺牙编码分析先天性缺牙表型与基因型的相关性

巩宇,冯海兰,何慧莹,葛严军*

北京大学口腔医学院 口腔医院修复科,北京100081

Correlation between the Phenotype and Genotype of Tooth Agenesis Patients by Tooth Agenesis Code

GONG Yu,FENG Hai-lian, HE Hui-ying,GE Yan-jun*

Department of Prosthodontics,Peking University School and Hospital of Stomatology,Beijing 100081,China

摘要 参考文献 相关文章

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摘要 目的 用先天性缺牙编码(TAC)及传统缺失牙位方式比较先天缺牙表型及基因型的相关性。方法 选择截止至2007年5月文献报道的明确为PAX9或MSX1基因突变导致的单纯型先天缺牙病例,将其缺失牙位情况按不同牙位的缺失率和TAC编码形式分别记录,对比两种基因突变导致的缺牙模式异同。结果 除上颌中切牙和侧切牙及下颌尖牙和第一磨牙外,其余各牙位间牙齿缺失率差异均有统计学意义（P<0.05,P<0.001）,MSX1基因突变以上颌第一前磨牙、上颌第二前磨牙和下颌第二前磨牙缺失常见。PAX9基因突变以第一、第二和第三磨牙缺失常见。TAC编码形式的分析结果相似。结论 PAX9或MSX1基因突变导致的先天性缺牙表型有明显差异。TAC编码形式可用于缺牙表型与基因型相关性的分析。

关键词： 先天性缺牙编码 PAX9 MSX1 先天性缺牙表型

Abstract: Objective To analyze the correlation between the phenotype and genotype of tooth agenesis using the tooth agenesis code (TAC) and the traditional descriptor for missing teeth. Methods Patients with isolated hypodontia caused by PAX9 or MSX1 mutation reported before May 2007 were enrolled. The teeth missing rate and TAC code were recorded. The missing teeth patterns caused by the two mutations were compared. Results The teeth missing rates in each teeth positions were significantly different between maxillary and mandibular except maxillary central incisor,lateral incisor and mandibular canine,first molar (P<0.05,P<0.001). MSX1 gene mutation often led to the loss of maxillary first premolar,maxillary second premolar,and mandibular second premolar,while PAX9 gene mutation often led to the loss of the first,second,and third molars. The results were similar when analyzed either by TAC code analysis or by traditional descriptor. Conclusions PAX9 and MSX1 gene mutation can cause different phenotypes of tooth agenesis. The TAC code can be used in the analysis of the correlation between phenotype and genotype of the missing teeth patients.

Keywords: tooth agenesis code PAX9 MSX1 phenotype of tooth agenesis

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Corresponding Authors: 冯海兰 Email: kqfenghl@bjmu.edu.cn

About author: 010-62179977-5232,邮箱:kqfenghl@bjmu.edu.cn

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