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Identification Of *PAX9* Single Nucleotide Polymorphism In Class III Malocclusion Patients With Mandibular Prognatism

Noraini Abu Bakar^a | Khairani Idah Mokhtar^b | Azrul Fazwan Kharuddin^c

^aDepartment of Paediatric Dentistry, Orthodontics and Dental Public Health, Kulliyyah of Dentistry, IIUM | ^bDepartment of Fundamental Dental-Medical Sciences, Kulliyyah of Dentistry, IIUM | ^cDepartment of Computational and Theoretical Sciences, Kulliyah of Science, IIUM

Introduction: PAX9 (Paired box 9) gene is one of the genes which play significant role during craniofacial development. Single nucleotide polymorphism (SNP) in PAX9 has been associated with Class II/Division 2 malocclusion (with or without hypodontia). However, the relationship between PAX9 SNP marker (rs8004560) with mandibular prognathism (MP) has not been analysed, at least in our local population. This study aimed to detect the presence of PAX9 (rs8004560) SNP in Class III malocclusion patients (with MP) in the local population. Materials and Methods: Genomic DNA were extracted from unstimulated saliva of 31 class I malocclusion (control samples) and 30 patients from Class III malocclusion (MP). Cephalometric measurements were performed prior to saliva samples collection. The DNA was amplified using the specific primers for the marker rs8004560 and the genotyping was done by sequencing. Chi-square test was used to determine the overrepresentation of marker allele (p<0.05). Results: Presence of PAX9 SNP (rs8004560) was detected in local population analysed and the distribution of its genotype and allele could be observed. There were significant differences between allele (p=0.000) and genotype (p=0.000) frequency within control (Class I) and Class III malocclusion. Conclusion(s): The most common allele of a marker flanking PAX9 (rs8004560) was over-represented in the mandibular prognathism (MP) subjects indicating the genetic association of PAX9 (rs8004560) SNP in the incidence of MP. Further studies involving larger number of samples should be developed in order to understand the exact role and mechanism of PAX9 in different classes of malocclusions.

KEYWORDS: malocclusion, mandibular prognathism, gene polymorphism, *Pax9*