OP-49

Genetic variants of BMP4/HpHI and IRF6 /MboI genes in two families with non syndromic cleft lip and palate patients

Ani Melani Maskoen[1*], Saskia L. Nasroen[2], Eky S. Soeria Soemantri[3]

[1]Oral Biology Department, Faculty of Dentistry, Universitas Padjadjaran, Bandung, Indonesia
[2]Dentistry Study Program Department, Faculty of Medicine, Universitas Jenderal Achmad Yani Cimahi, Bandung Indonesia
[3]Orthodontic Department, Faculty of Dentistry, Universitas Padjadjaran, Bandung, Indonesia

amelani@yahoo.com

Abstract

Non syndromic cleft lip and palate (NS CL/P) is the most common craniofacial malformation in humans. *Bone Morphogenetic Protein 4* (*BMP4*) and *Interferon Regulatory Factor 6* (*IRF6*) have been consistently shown to be associated in NS CL/P from some human populations in the world. The aim of this study was to know the role of *BMP4*/*HpH*I and *IRF6 /MboI* gene polymorphisms in 2 families with NS CL/P and get to know whether there would be a risk factor for the rehearsal occurence of NS CL/P in the subsequent offspring through the probability analysis of the mutant genotypes. The study was laboratory descriptive design and the examination was performed in the form of pedigrees from 2 families from 3 generations with NS CL/P by using PCR-RFLP with *HpH*I and *Mbo*I restriction enzymes. The study results showed that the probability of TC mutant genotype of *BMP4*/*HpH*I gene polymorphism was 1/6 to be inherited in third generation of NS CL/P patients and the probablity of GA mutant genotype of *IRF6/MboI* gene polymorphism was 1/8 to be inherited in third generation of NS CL/P patients. The probability of the children with *BMP4*/*HpH*I and *IRF6/MboI* gene polymorphisms are greater when their grandparents or parents were also recognize to have *BMP4*/*HpH*I and *IRF6 /MboI* gene polymorphisms.

Keywords: nonsyndromic CL/P; genetic variants; inherited pattern; *BMP4*/*HpH*I; *IRF6 /MboI*

*Author for Correspondence