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

Ani Melani Maskoen1*, Saskia L. Nasroen2, Eky S. Soeria Soemantri3

1Oral Biology Department, Faculty of Dentistry, Universitas Padjadjaran, Bandung, Indonesia
2Dentistry Study Program Department, Faculty of Medicine, Universitas Jenderal Achmad Yani Cimahi, Bandung, Indonesia
3Orthodontic 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/HpHI 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 occurrence 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 HpHI and MboI restriction enzymes. The study results showed that the probability of TC mutant genotype of BMP4/HpHI gene polymorphism was 1/6 to be inherited in third generation of NS CL/P patients and the probability 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/HpHI and IRF6/MboI gene polymorphisms are greater when their grandparents or parents were also recognize to have BMP4/HpHI and IRF6/MboI gene polymorphisms.

Keywords: nonsyndromic CL/P; genetic variants; inherited pattern; BMP4/HpHI; IRF6/MboI

*Author for Correspondence