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MSX1 AND *TGFβ3* Gene Mutation and the Risk of Nonsyndromic Cleft Palate Only (NS CPO) Among Indonesian Patients

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Abstract : Non syndromic cleft palate only (NS CPO) is one of the most common congenital malformations that affect between 1 of 1000 - 2500 live births worldwide, which is considered as a genetically complex, multifactorial disease. Based on several association studies among the candidate genes with NS CPO, *MSX1* and *TGFβ3* genes emerged as the strong candidate genes in different populations with NS CPO. The objective of this study was to analyze the relationship between the mutations in *MSX1* gene (exon 1 C101G and exon 2 G817T) and *TGFβ3* gene (intron 3 T>A) and the risk of NS CPO in Indonesian patients.

This study was case control using samples from 22 NS CPO subjects and 43 control subjects. Venous blood samples were collected, then DNA was extracted and the segment of *MSX1* and *TGFβ3* genes were PCR-amplified. DNA sequencing from DNA fragments covering *MSX1* exon 1 C101G was performed by Sanger method. Digestion products containing *MSX1* exon 2 G817T and intron 3 T>A *TGFβ3* were evaluated. Statistical analysis used to determine significance differences from mutations frequency among both subjects was $\chi 2$. The odds ratio was used to determine a risk factor of NS CPO.

The study results showed that mutations in exon 1 C101G of *MSX1* gene were identified yet the differences in both subjects were not significant and also in *MSX1* gene exon 2 G817T as there was no mutation found in G817T. In *TGFβ3* intron 3 T>A, the frequency of A mutant allele was 35.7% in NS CPO and 28.8% in control. This difference was not significant statistically ($\chi 2$=0.748; p > 0,05), but the frequency of A mutant allele (odds ratio (OR) = 1.825; 95% CI = 0,635 - 5,245) and heterozygous mutant of TA genotype (OR = 1.941; 95% CI = 0,700 - 5,384) were associated with increased risk of NS CPO.

In conclusion, the mutations of *MSX1* gene in exon 1 C101G and exon 2 G817T are not considered to be a risk factor in Indonesian patients with NS CPO but *TGFβ3* intron 3 T>A can be considered to be the risk factor associated with NS CPO development in Indonesian patients.

Key words : nonsyndromic cleft palate only, gene mutation, *MSX1* gene, *TGFβ3* gene.

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