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Sequence Variants in Exon 1 of *MSX1* Gene Associated With Nonsyndromic Cleft Lip/Palate (NS CL/P) Among Indonesian Patients

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Abstract : *Background:*Orofacial cleft, especially nonsyndromic cleft lip/palate (NS CL/P), is a common congenital abnormalities that affect between 1 in 700 - 1000 live births worldwide. The etiology of NS CL/P is multifactorial that involves the interaction of both genetic and environmental factors. Drosophila muscle segment homeobox homolog-1 (*MSX1*) gene has been proposed as one of strong candidate genes associated with NS CL/P. The patterns of *MSX1* gene mutation may vary according to race and geographical region. The purpose of this study was to detect and analyze the mutation in exon 1 of *MSX1* gene in the form of sequence variants as the risk factor that being an etiological role in the development of NS CL/P in Indonesian patients.
Methods: This study was case control design using samples from 48 NS CL/P subjects and 43 control subjects. PCR and Sanger Sequencing Technique were used to resolved the aim of the study. Statistical analysis which was used to determine significantly of differences from sequence variants frequency among NS CL/P subject and control subject was $\div^2$. The odds ratio was used to determine a risk factor of NS CL/P.
Results: The study results showed that 2 sequence variants in exon 1 of *MSX1* gene were identified as C101G/rs36059701 and C330T/rs34165410. In C101G, the frequency of G mutant allele was 50% in both NS CL/P subject and control subject. This difference was not significant ($\div^2$=0,019; p > 0,05). In C330T, the frequency of T mutant allele frequency was 48.1% in NS CL/P subject and 51.9% in control subject and this difference was not significant ($\div^2$=0,326; p > 0,05). The odds ratio from all mutant alleles did not show significant result statistically means that the risk factor can not be determined.
Results: The study results showed that 2 sequence variants in exon 1 of *MSX1* gene were identified as C101G/rs36059701 and C330T/rs34165410. In C101G, the frequency of G mutant allele was 50% in both NS CL/P subject and control subject. This difference was not significant ($\div^2$=0,019; p > 0,05). In C330T, the frequency of T mutant allele frequency was 48.1% in NS CL/P subject and 51.9% in control subject and this difference was not significant ($\div^2$=0,326; p > 0,05). The odds ratio from all mutant alleles did not show significant result statistically means that the risk factor can not be determined.
Keywords : nonsyndromic cleft lip/palate, exon 1 of *MSX1* gene, Indonesian.

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