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TGFA TaqI Gene Variant and the Risk Factor of Non-Syndromic Cleft Palate only 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 in 1000 - 2500 live births worldwide. The etiopathogenesis of clefts including NS CPO has been widely studied, yet it remains unclear. NS CPO is considered as a genetically complex, multifactorial disease. Transforming growth factor alpha (*TGFA*) is a candidate gene that might contribute to the development of NS CL/P. The objective of this study was to detect *TGFA TaqI* gene variant and and analyze the risk of NS CPO among Indonesian patients. This study was case control design using samples from 33 NS CPO subjects and 31 control subjects. DNA was extracted from venous blood and the *TGFA* gene was amplified using polymerase chain reaction (PCR) technique, then digestion product from *TaqI* restriction enzyme were evaluated. Results showed that the *TGFA TaqI* gene variant was identified. The frequency of C2 mutant allele (odds ratio (OR) = 2.229; 95% CI = 0,746 – 6,654) and C2C2 genotype (OR = 1.935; 95% CI = 0,167 – 2,482) were associated with increased risk of NS CPO. In conclusion, *TGFA TaqI* gene variant can be considered to be the risk factor associated with NS CPO development in Indonesian patients.
Key words: non syndromic cleft palate only, *TGFA TaqI* gene variant.

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