

## Distribusi genotip dan alel polimorfisme gen bmp4 t538c pada penderita orofacial cleft = Distribution of genotype and allele polymorphism bmp4 t538c gene in orofacial cleft patients

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### Abstrak

#### **ABSTRACT**

Latar Belakang: Orofacial cleft merupakan ruang abnormal kongenital yang terjadi pada bibir atas, tulang alveolar, dan palatum. Orofacial cleft terdiri dari berbagai jenis yaitu, celah bibir, celah palatum, dan celah bibir dan palatum. Orofacial cleft adalah anomali kongenital yang dapat disebabkan oleh faktor genetik maupun faktor lingkungan. Belakangan ini penelitian menunjukkan bahwa beberapa gen terlibat dalam penyebab terjadinya orofacial cleft, salah satunya adalah gen BMP4. Terdapat penelitian yang menunjukkan hubungan antara polimorfisme nukleotida tunggal gen BMP4 T538C dengan terjadinya orofacial cleft di populasi Cina. Tujuan: Mengetahui hubungan antara polimorfisme gen BMP4 T538C dengan penderita orofacial cleft di Indonesia. Metode: Analisis polimorfisme gen BMP4 T538C dilakukan dengan metode PCR-RFLP dengan enzim restriksi Hph1. Hasil: Dari 100 sampel yaitu, 25 sampel orofacial cleft dan 75 sampel non orofacial cleft, ditemukan 25 sampel orofacial cleft memiliki genotip CC (100%) sedangkan pada kelompok non orofacial cleft ditemukan 11 sampel memiliki genotip CC (14.7%), 55 sampel memiliki genotip CT (73.3%), dan 9 sampel memiliki genotip TT (12%). Seluruh sampel orofacial cleft memiliki alel C sedangkan pada kelompok non orofacial cleft 77 sampel memiliki alel C (51.3%) dan 73 sampel memiliki alel T (48.7%). Kesimpulan: Terdapat perbedaan bermakna pada distribusi genotip dan alel gen BMP4 T538C antara penderita orofacial cleft dan non orofacial cleft (p value genotip dan alel = 0.001).

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"ackground: Orofacial cleft is a congenital abnormal space or gap in the upper lip," "lveolar bone, or palate. There are many types of orofacial cleft, such as, cleft lip, cleft alate, and cleft lip and palate. Orofacial cleft is congenital anomalies that often appened. Orofacial cleft caused by many factor, such as, genetic and environment. ecent studies showed that several genes could affect orofacial cleft, such as, BMP4 ene. There was study that showed association between single nucleotide polymorphism f BMP4 gene T538C with orofacial cleft in Chinese population. Objective: To escribe the correlation between BMP4 T538C polymorphism and orofacial cleft in" "ndonesia. Methods: Analysis of BMP4 T538C gene polymorphism was performed by CR-RFLP methods with Hph1 restriction enzyme. Results: From 100 samples that onstist of 25 orofacial cleft samples and 75 non orofacial cleft samples, 25 samples of rofacial cleft showed genotype CC (100%) while in non orofacial cleft samples, 11 amples showed genotype CC (14.7%), 55 samples showed genotype CT (73.3%), and" "9 samples showed genotype TT (12%). All of orofacial cleft samples showed C allele hile in non orofacial cleft samples were found 77 allel C (51.3%) and 73 allele T" "48.7%). Conclusion: There were significance differences between orofacial cleft and on orofacial cleft samples, both in genotype and allele distribution of BMP4 T538C ene polymorphism (p value for both genotype and allele = 0.001)."