

Distribusi genotip dan alel polimorfisme genetik
Methylenetetrahydrofolate Reductase (MTHFR) C677T pada penderita
Orofacial Cleft = Genotype and allotype frequencies
Methylenetetrahydrofolate Reductase (MTHFR) C677T polymorphism
in Orofacial Cleft

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Abstrak

Polimorfisme genetik MTHFR C677T adalah faktor resiko orofacial cleft (OFC). Dalam mengetahui distribusi genotip dan hubungan polimorfisme MTHFR C677T dengan orofacial cleft, maka diikutsertakan grup OFC dan grup kontrol sebanyak 24 dan 47 sampel ekstraksi DNA dari Laboratorium Biologi Oral FKG UI. Identifikasi molekuler dengan teknik PCR-RFLP. Kedua grup berada dalam Hardy-Weinberg equilibrium. Grup OFC, frekuensi alel C 89.6% dan alel T 10.4% serta tidak ditemukan genotip homozigot variant (TT). Grup kontrol ditemukan 2 individu bergenotip TT, frekuensi alel C 75.5 % dan alel T 24.4 % . Hasil uji Chi-square, polimorfisme MTHFR C677T berhubungan dengan orofacial cleft ($p < 0.05$).

.....MTHFR C677T polymorphism is a possible risk factor for orofacial cleft. In assessing the genotype distribution of MTHFR C677T polymorphism and its relation to orofacial cleft, 24 OFC samples and 47 control samples were included and provided by Oral Biology Laboratory Faculty of Dentistry University of Indonesia. PCR-RFLP technique was used for genotyping. Allelic frequencies in OFC group were 89.6% (C allele) and 10.4% (T allele) while in control group were 75.5% (C allele) and 24.4% (T allele). Genotyping showed two individuals of TT genotype in control group. Polymorphism MTHFR C677T in two groups were in Hardy-Weinberg equilibrium and showed a significant association with orofacial cleft ($p < 0.05$).