Analisis Asosiasi Single Nucleotide Polymorphism (SNP) rs2476601 Gen Protein Tyrosine Phosphatase Non-Receptor Type 22 (PTPN22) dengan Alopecia Areata pada Populasi Indonesia = Analysis of Association between Single Nucleotide Polymorphism (SNP) rs2476601 Protein Tyrosine Phosphatase Non-Receptor Type 22 (PTPN22) Gene and Alopecia Areata on Indonesian Population

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Abstrak

Alopecia areata (AA) merupakan jenis kerontokan rambut pada manusia yang disebabkan oleh serangan sel-sel imun tubuh terhadap folikel rambut di fase anagen. Single nucleotide polymorphism (SNP) rs2476601 adalah salah satu varian gen yang terletak di gen protein tyrosine phosphatase non-receptor type 22 (PTPN22) dan diketahui berasosiasi dengan kemunculan AA berdasarkan penelitian yang dilakukan di berbagai negara. Perubahan basa sitosin menjadi timin pada posisi ke-1858 diprediksi menyebabkan penurunan kemampuan protein lymphoid-specific tyrosine phosphatase (Lyp) untuk menghambat aktivasi sel T. Penelitian ini bertujuan untuk mengetahui keberadaan asosiasi SNP rs2476601 dengan kemunculan fenotipe AA pada populasi Indonesia. Pengambilan sampel buccal swab dilakukan terhadap 50 pasien penderita AA dan 50 individu normal sebagai subjek kontrol. Penentuan genotipe subjek dilakukan dengan teknik PCR-RFLP menggunakan enzim restriksi RsaI, lalu analisis statistik dilakukan dengan uji Fisher’s exact. Dari seluruh subjek yang diteliti, sebanyak 99% genotipe merupakan genotipe CC yang ditandai dengan 4 pita dan 1% genotipe merupakan genotipe CT yang ditandai dengan 5 pita. Tidak ditemukan adanya genotipe homozigot TT pada kedua kelompok studi. Populasi subjek yang terlibat dalam penelitian berada dalam keseimbangan Hardy-Weinberg. Nilai p yang ditemukan dari hasil uji Fisher’s exact tidak menunjukkan adanya asosiasi yang bermakna antara genotipe CT/TT dan kondisi AA (p>0,05). Penelitian ini memberikan kesimpulan bahwa rs2476601 gen PTPN22 tidak memiliki asosiasi terhadap kemunculan AA pada populasi di Indonesia.

.....Alopecia areata (AA) is a type of hair loss in human caused by the body's immune cells attacking hair follicles in the anagen phase. Single nucleotide polymorphism (SNP) rs2476601 is a gene variant located in the protein tyrosine phosphatase non-receptor type 22 (PTPN22) gene and associated with the emergence of AA based on studies conducted in various countries. The substitution of cytosine to thymine base at position 1858 decreases the ability of lymphoid-specific tyrosine phosphatase (Lyp) protein to inhibit T cell activation. This study aimed to determine the association of SNP rs2476601 with the appearance of the AA phenotype in the Indonesian population. Buccal swabs were extracted from 50 patients with AA and 50 normal individuals as control subjects. PCR RFLP technique were used to determine the subject’s genotype using the RsaI restriction enzyme, then the statistical analysis were conducted using Fisher’s exact test calculations. From all the subjects involved, 99% of the genotypes belonged to CC genotype indicated by four bands and 1% belonged to CT genotype indicated by five bands. No homozygous TT genotype was detected in both study groups. This study implies that the subject population involed is at Hardy-Weinberg equilibrium. However, the p-value generated from the Fisher’s exact test shows that there was no association between the CT/TT genotype and AA conditions (p>0.05). In conclusion, this study found that rs2476601

from the PTPN22 gene is not associated with the emergence of AA in the Indonesian population.