Studi Pilot Genome-Wide Association Untuk Parameter Pigmentasi Kulit Wajah Pada Wanita Di Jakarta = Genome-Wide Association Study Of Skin Pigmentation Parameter In Women: A Pilot Study In Jakarta

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Abstrak

Pigmentasi merupakan proses fisiologis yang ditandai dengan perubahan warna kulit yang terjadi secara kompleks dan dapat dipengaruhi oleh profil genetik. Hingga saat ini, belum terdapat penelitian serta publikasi yang membahas bagaimana profil genetik dapat mempengaruhi karakteristik pigmentasi di Indonesia. Analisis Genome-Wide Association Study (GWAS) pada penelitian ini dilakukan terhadap 94 subjek wanita pada populasi di Jakarta. Penelitian meliputi pengambilan dan pengolahan data, uji asosiasi, dan analisis pengayaan menggunakan berbagai basis data. Uji asosiasi menunjukkan terdapat dua SNP yang memiliki asosiasi terhadap indeks melanin dengan nilai P <1x10-5 yaitu rs10031234 (nilai P: 3,229x10-6) dan rs11548325 (nilai P: 7,808x10-6). Analisis pengayaan menunjukkan SNP rs10031234 terletak pada region gen SORCS2, sementara SNP rs11548325 terletak pada region gen PSAPL1. Gen SORCS2 terekspresi pada jaringan kulit khususnya pada sel yang berkaitan dengan akar rambut dan fibroblast. Variasi rs11548325 terjadi pada domain SapA diketahui berkaitan dengan metabolisme sphingolipid. Analisis pengayaan fungsi menunjukkan kaitan gen-gen yang terasosiasi dengan proses keratinisasi. Namun demikian, perlu dilakukan penelitian lebih lanjut menggunakan sampel yang lebih besar agar dapat diperoleh nilai signifikansi yang lebih bermakna serta analisis korelasi antar kedua SNP rs10031234 dan rs11548325 dengan fungsi molekuler.

.....Skin pigmentation is a physiological process characterized by changes in the color of the skin and can be influenced by genetic profiles. To date, there have been no studies and publications that discuss how genetic profiles can affect skin pigmentation in Indonesia. Genome-Wide Association Study (GWAS) was conducted on 94 female subjects who live in Jakarta. The research procedure includes data collection and processing, association testing, and enrichment analysis using various databases. The association test showed that there were two SNPs associated with the melanin index with a P value <1x10-5, which are rs10031234 (P value: 3.229x10-6) and rs11548325 (P value: 7.808x10-6). Enrichment analysis showed that SNP rs10031234 was located in the SORCS2 gene region, while SNP rs11548325 was located in the PSAPL1 and SORCS2 gene regions. SORCS2 gene is expressed in skin tissue, especially in cells associated with hair roots and fibroblasts. The rs11548325 variation located in the SapA domain which is known to be related to glycosphingolipid metabolism. Functional enrichment analysis showed the association of genes associated with the keratinization process. However, it is necessary to carry out further research using a larger sample to obtain a higher significance value, and also conducting correlation analysis between SNP rs10031234 and rs11548325 with molecular.