Asosiasi antara genotip gen SCGB3A2 Rs1368408 dan kekambuhan penyakit graves pada populasi Jakarta = Association between genotype of SCGB3A2 gene Rs1368408 and relapse of graves' disease in Jakarta's population.

Aisya Sita Marcha, author

Abstrak

Latar belakang: Sekitar 50% pasien penyakit Graves mengalami kekambuhan setelah obat antitiroid diberhentikan. Padahal penyakit Graves yang kambuh umumnya bersifat lebih berat, sehingga membutuhkan terapi yang lebih agresif. Dengan demikian penting untuk mengetahui faktor untuk memprediksi kekambuhan penyakit Graves. Faktor risiko kekambuhan penyakit Graves di antaranya adalah usia, tanda klinis, faktor lingkungan, serta faktor genetik. Gen SCGB3A2 merupakan salah satu gen yang berkaitan dengan penyakit Graves. Genotip AA dan GA, serta alel A gen SCGB3A2 rs1368408 diketahui merupakan faktor risiko timbulnya penyakit Graves. Namun, gen SCGB3A2 rs1368408 belum diketahui asosiasinya dengan kekambuhan penyakit Graves.
Tujuan: Mengetahui hubungan polimorfisme gen SCGB3A2 rs1368408 dengan kekambuhan penyakit Graves.
Metode: Desain penelitian ini adalah penelitian potong lintang dengan total 77 sampel. Sampel yang digunakan adalah DNA yang telah diisolasi dari pasien yang telah menjalani terapi obat antitiroid selama 12 bulan. Sampel dipilih dengan metode consecutive sampling. Data yang diambil yaitu genotip dan alotip gen SCGB3A2 rs1368408 melalui teknik tetra-primer ARMS-PCR. Sampel dikelompokkan sesuai dengan kekambuhannya dan dilihat frekuensi genotip dan alotip dari masing-masing kelompok. Data juga dianalisis dengan uji chi-square atau uji Fisher untuk melihat asosiasi genotip dan alotip gen SCGB3A2 rs1368408 dengan kekambuhan penyakit Graves.
Hasil: Frekuensi genotip pada keseluruhan sampel pasien penyakit Graves didapatkan 98,7% GG dan 1,3% GA. Pada kelompok kambuh didapatkan genotip 97,8% GG dan 2,2% GA, serta alotip 98,9% alel G dan 1,1% alel A. Sedangkan pada kelompok tidak kambuh 100% memiliki genotip GG, sehingga didapatkan frekuensi alotip 100% alel G. Dari analisis menggunakan uji Fisher, tidak didapatkan perbedaan signifikan antara genotip gen SCGB3A2 rs 1368408 dan kekambuhan penyakit Graves (p=1.000). Analisis uji Fisher pada alotip gen SCGB3A2 rs1368408 juga menunjukkan tidak adanya perbedaan signifikan dengan kekambuhan penyakit Graves (p=1.000).
Simpulan: Tidak terdapat asosiasi genotip atau alotip gen SCGB3A2 rs1368408 dengan kekambuhan penyakit Graves.
.....
Background: Approximately 50% of Graves' disease patients experience a relapse after discontinuation of antithyroid drugs. Relapsed Graves' disease patients are generally more severe and require more aggressive therapy. Thus, it is important to know the factors that can predict the relapse of Graves' disease. Age, clinical signs, environment, and genetic are some of the risk factors for Graves' disease relapse. SCGB3A2 gene is one of the genes associated with Graves' disease. Genotype GA and AA, as well as alelle A of SCGB3A2 gene rs1368408 are known to be risk factors for Graves' disease. However, the association

between SCGB3A2 gene rs1368408 and relape of Graves’ disease is not yet known.
Objective: To determine the correlation between the polymorphism of SCGB3A2 gene rs1368408 and Graves' disease relapse.
Methods: Study design of this study was cross-sectional with a total of 77 samples. The sample used in this study was DNA that had been isolated from patients who have received antithyroid drug treatment for 12 months. Samples were selected through consecutive sampling method. The data taken were genotypes and allotypes of SCGB3A2 gene rs1368408 using tetra-primer ARMS-PCR technique. Samples were grouped according to the incidence of relapse. The genotype and allotype frequency of each group was observed. Data were also analyzed using chi-square test or Fisher’s exact test to determine the association of genotype and allotype of SCGB3A2 gene rs1368408 with Graves’ disease relapse.
Results: The genotype frequency in the entire sample of patients with Graves’ disease was found to be 98.7% GG and 1.3% GA. In the relapse group, the genotype observed was 97.8% GG and 2.2% GA whereas the allotype frequency in this group was 98.9% G allele and 1.1% A allele. All samples in non-relapse group had GG genotype (100%), thus the allotype frequency was 100% G allele. Analysis using Fisher’s exact test showed no significant difference between genotype of SCGB3A2 gene rs1368408 and Graves’ disease relapse ($p=1.000$). Fisher's exact analysis of SCGB3A2 gene rs1368408 allotype also found no significant difference with Graves' disease recurrence ($p=1.000$).
Conclusions: There was no association between genotype or allotype of SCGB3A2 gene rs1368408 and Graves' disease relapse.