Risiko kekambuhan penyakit graves ditinjau dari genotip gen PTPN22 rs2476601 pada populasi di Jakarta = Recurrence risk of graves disease attributed to the genotype of PTPN22 rs2476601 gene in Jakarta's population

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Abstrak

Latar belakang: Penyakit Graves merupakan penyakit autoimun yang memiliki kemungkinan untuk kambuh setelah remisi. Sebanyak 50% pasien berisiko mengalami kekambuhan setelah menyelesaikan terapi obat antitiroid (OAT). Salah satu prediktor kekambuhan penyakit Graves adalah faktor genetik. Gen PTPN22 rs2476601 merupakan gen yang berperan dalam persinyalan sel T. Polimorfisme pada gen ini dapat memicu proses autoimun pada penyakit Graves.

Tujuan: Mengetahui hubungan polimorfisme gen PTPN22 rs2476601 terhadap risiko kekambuhan penyakit Graves.

Metode: Penelitian ini adalah penelitian observasional dengan metode cross sectional. Subjek penelitian merupakan 80 sampel DNA pasien penyakit Graves yang telah mendapatkan terapi OAT selama >12 bulan. Kekambuhan pada pasien dinilai dalam 12 bulan pasca pasien berhenti menggunakan OAT. Pemeriksaan genotip gen PTPN22 rs2476601 dilakukan menggunakan teknik Tetra-primer amplification refractory mutation system-polymerase chain reaction (T-ARMS-PCR).

Hasil: Pada pasien penyakit Graves di Jakarta, frekuensi genotip GA ditemukan paling tinggi (77.5%) dibandingkan GG (18.75%) dan AA (3.75%). Genotip AA lebih sering ditemukan pada kelompok kambuh (6%) dibandingkan tidak kambuh (0%). Sementara itu, Genotip GA dan GG lebih sering ditemukan pada kelompok tidak kambuh (79% dan 21%) dibandingkan kambuh (77% dan 17%). Namun, hasil uji fisher tidak menunjukkan adanya perbedaan yang bermakna antara genotip gen PTPN22 rs2476601 dengan kekambuhan penyakit Graves (p=0.264). Frekuensi alotip alel A ditemukan lebih tinggi pada kelompok kambuh (45%) dibandingkan tidak kambuh (39%). Sedangkan, frekuensi alotip alel G ditemukan lebih tinggi pada kelompok tidak kambuh (61%) dibandingkan kambuh (53%). Namun, hasil uji chi-square juga tidak menunjukkan adanya perbedaan yang bermakna antara alotip gen PTPN22 rs2476601 dengan kekambuhan penyakit Graves (p=0.505).

Kesimpulan: Polimorfisme gen PTPN22 rs2476601 tidak berhubungan dengan kekambuhan penyakit Graves pada populasi di Jakarta

.....Background: Graves' disease is an autoimmune disease that has a chance to recur after remission. 50% of patients have the risk of recurrence after completed anti-thyroid drugs (ATD) therapy. One of the recurrence predictors of Graves' disease is the genetic factor. The PTPN22 rs2476601 gene is a gene involved in T cell signaling. Polymorphism in this gene may trigger the autoimmune process in Graves' disease.

Aim: Determine the association between PTPN22 rs2476601 gene polymorphism and the recurrence risk of Graves' disease.

Methods: This study was an observational study with a cross-sectional design. The subjects of this study were 80 DNA samples from patients with Graves' disease who received ATD therapy for >12 months. Recurrence of the disease was assessed within 12 months after patients completed ATD therapy. Genotype examination of PTPN22 rs2476601 gene was performed using Tetra-primer amplification refractory mutation system-polymerase chain reaction (T-ARMS-PCR) technique.

Results: Patients with Graves' disease in Jakarta had the highest genotype frequency of GA (77.5%) compared to GG (18.75%) and AA (3.75%). AA genotype was more frequent in the recurrence group (6%) compared to non-recurrence (0%). In contrast, GA and GG genotype were more frequent in the non-recurrence group (79% and 21%) compared to recurrence (77% and 17%). However, the result of fisher test showed there was no significant difference between the genotype of PTPN22 rs2476601 gene and recurrence of Graves' disease ($p=0.264$). The allotype frequency of A allele was higher in the recurrence group (45%) compared to non-recurrence (39%). Meanwhile, the allotype frequency of G allele was higher in the non-recurrence group (61%) compared to recurrence (53%). However, the result of chi-square test also showed there was no significant difference between the allotype of PTPN22 rs2476601 gene and recurrence of Graves' disease ($p=0.505$).

Conclusion: PTPN22 rs2476601 gene polymorphism is not associated with the recurrence of Graves' disease in Jakarta's population.