

INTISARI

POLIMORFISME GEN *ADENOSINE TRIPHOSPHATE-BINDING CASSETTE SUBFAMILY-A MEMBER-1* rs2230806 SEBAGAI FAKTOR RISIKO TERJADINYA DISLIPIDEMIA PADA ETNIS BUGIS DI SULAWESI TENGAH

Latar Belakang: Dislipidemia merupakan penyakit multifaktorial yang dipengaruhi oleh interaksi antara faktor genetik dan lingkungan. Polimorfisme gen ABCA1 rs2230806 berkaitan dengan risiko dislipidemia dan penyakit kardiovaskular di beberapa etnis. **Tujuan:** Mengkaji pengaruh polimorfisme gen ABCA1 rs2230806 terhadap profil lipid dan risiko individu mengalami dislipidemia pada laki-laki dan perempuan Masyarakat Bugis di Sulawesi Tengah. **Metode:** Penelitian observasional dengan pendekatan *case control* menggunakan sampel *whole blood*. Sebanyak 172 subjek dibagi menjadi 2 kelompok yakni kelompok kontrol dan kelompok kasus. Subjek dikelompokkan sebagai kasus apabila memenuhi setidaknya dua dari empat kriteria profil lipid berikut: LDL-C ≥ 130 mg/dL, HDL-C < 40 mg/dL (laki-laki)/HDL-C < 50 mg/dL (perempuan), TC ≥ 200 mg/dL dan TG ≥ 150 mg/dL. Subjek juga dikelompokkan sebagai kasus apabila sedang mengonsumsi obat antidislipidemia seperti obat statin. Analisis genotipe dilakukan dengan metode PCR-RFLP. **Hasil:** Genotipe GA, AA dan gabungan genotipe GA+AA polimorfisme rs2230806 meningkatkan risiko menderita dislipidemia pada Masyarakat Etnis Bugis di Sulawesi Tengah dengan nilai OR (95%CI;p) secara berturut-turut adalah 2,018 (1,010-4,034;0,046), 2,355 (1,009-5,429;0,046) dan 2,119 (1,112-3,040;0,021). Kadar trigliserida pada subjek dengan genotipe GA+AA polimorfisme rs2230806 lebih tinggi secara signifikan dibandingkan dengan genotipe GG ($p=0,024$). Ketika dilakukan stratifikasi berdasarkan jenis kelamin, Genotipe GA dan gabungan genotipe GA+AA polimorfisme rs2230806 meningkatkan risiko menderita dislipidemia pada laki-laki (OR=4,071; 95%CI=1,311-12,646; $p=0,013$). Kadar trigliserida dan kolesterol total pada subjek laki-laki pembawa genotipe GA+AA lebih tinggi daripada subjek laki-laki pembawa genotipe GG. Analisis multivariat menunjukkan bahwa polimorfisme rs2230806 gen ABCA1 tetap berkaitan dengan risiko dislipidemia disamping jenis kelamin perempuan dan IMT. **Kesimpulan:** Pembawa genotipe GA dan AA polimorfisme gen ABCA1 rs2230806 berperan dalam meningkatkan kadar trigliserida dan risiko kejadian dislipidemia pada Masyarakat Bugis di Sulawesi Tengah. Perbedaan kadar trigliserida disertai dengan perbedaan kolesterol total serta kasus dislipidemia terlihat pada individu laki-laki namun tidak terlihat pada individu perempuan.

Kata Kunci: Dislipidemia, Gen ABCA1, Kolesterol, Polimorfisme rs2230806, Trigliserida

ABSTRACT

POLYMORPHISM OF ADENOSINE TRIPHOSPHATE-BINDING CASSETTE SUBFAMILY-A MEMBER-1 GENE rs2230806 AS A RISK FACTOR OF DYSLIPIDEMIA FOR BUGINESE PEOPLE IN CENTRAL SULAWESI

Background: Dyslipidemia is a multifactorial disease that is influenced by interactions between genetic and environmental factors. The ABCA1 rs2230806 gene polymorphism is associated with dyslipidemia and cardiovascular disease risk in several ethnic groups. **Objective:** To examine the effect of the ABCA1 rs2230806 gene polymorphism on lipid profiles and the individual risk of developing dyslipidemia in male and female Buginese people in Central Sulawesi. **Method:** Observational study with a case-control approach using whole blood samples. One hundred seventy-two subjects were divided into two groups: the control group and the case group. Subjects were classified as cases if they met at least two of the following four lipid profile criteria: LDL-C ≥ 130 mg/dL, HDL-C < 40 mg/dL (male)/HDL-C < 50 mg/dL (female), TC ≥ 200 mg/dL and TG ≥ 150 mg/dL. Subjects also be classified as cases if they take anti-dyslipidemia drugs. Genotype analysis was performed using the PCR-RFLP method. **Results:** The GA, AA genotype and the combined GA+AA genotype polymorphism rs2230806 increased the risk of suffering from dyslipidemia in the Bugis Ethnic Community in Central Sulawesi with OR values (95%CI;p) respectively 2.018 (1.010-4.034;0.046), 2.355 (1.009-5.429;0.046) and 2.119 (1.112-3.040;0.021). Triglyceride levels in subjects with the GA+AA polymorphism rs2230806 genotype were significantly higher than those with the GG genotype ($p=0.024$). When stratified by sex, the GA genotype and the combined GA+AA genotype polymorphism rs2230806 increased the risk of developing dyslipidemia in males (OR=4.071; 95% CI=1.311-12.646; $p=0.013$). Triglyceride and total cholesterol levels in male subjects carrying the GA+AA genotype were higher than in male subjects carrying the GG genotype. Multivariate analysis showed that the rs2230806 polymorphism of the ABCA1 gene was still associated with the risk of dyslipidemia, apart from female sex and BMI. **Conclusion:** The carriers of the GA and AA genotypes of the ABCA1 rs2230806 gene polymorphism play a role in increasing triglyceride levels and the risk of dyslipidemia in the Buginese People in Central Sulawesi. Differences in triglyceride levels accompanied by differences in total cholesterol and cases of dyslipidemia are seen in males but not in females.

Keywords: Dyslipidemia, ABCA1 Gene, Cholesterol, Polymorphism rs2230806, Triglycerides