INTISARI

STUDI DESKRIPTIF SNP GEN ARMS2 rs10490924 PADA PASIEN AMD DI YOGYAKARTA

Latar Belakang: Degenerasi makula terkait umur (*Age-related macular degeneration*) adalah kondisi gangguan degeneratif yang memengaruhi makula pada mata dan berdampak pada hilangnya penglihatan sentral seseorang di usia lanjut, dengan prevalensi sebesar 8,7% di dunia. Etiologi dari terjadinya AMD bersifat multifaktorial, yaitu melibatkan interaksi antara faktor genetik dan lingkungan. Faktor genetik utama yang terlibat dalam pathogenesis AMD ialah CFH dan ARMS2/HTRA1. ARMS2 dan HTRA1 telah banyak diteliti berhubungan dengan kejadian AMD pada populasi di Asia, yaitu pada ras Jepang dan Cina/Tionghoa. Meskipun polimorfisme pada ARMS2 (rs10490924) dan HTRA1 (rs11200638) terkait AMD efeknya tidak dapat dibedakan dengan menggunakan analisis statistik, tetapi SNP rs10490924 (ARMS2 A69S) sendiri dapat menjelaskan sebagian besar hubungan antara wilayah kromosom 10q26 dan AMD. Akan tetapi, hingga saat ini, penelitian mengenai kontribusi gen ARMS2 pada kejadian AMD di Indonesia belum ada, sehingga perlu dilakukan penjelasan lebih lanjut.

Tujuan: Penelitian ini bertujuan untuk mengetahui profil SNP gen ARMS2 pada pasien AMD.

Metode: Penelitian ini merupakan penelitian deskriptif yang menggunakan data sekunder berupa data demografi dan persebaran *Single Nucleotide Polymorphism* (SNP) pada gen ARMS2 pasien AMD di 3 rumah sakit yaitu, Rumah Sakit Umum Pusat Dr. Sardjito, Rumah Sakit Pusat TNI AU dr. S. Hardjolukito, dan Rumah Sakit Mata Dr. Yap Yogyakarta dalam periode Agustus 2016 hingga Oktober 2017. Sampel diambil menggunakan metode *consecutive sampling*. Sampel darah tepi pasien AMD diuji di Laboratorium Fakultas Kedokteran Universitas Gadjah Mada dengan dilakukan *Polymerase Chain* Reaction (PCR).

Hasil: Profil *single nucleotide polymorphism* (SNP) rs10490924 gen ARMS2 pada 100 pasien yang didiagnosis *age related macular degeneration* (AMD), baik tipe basah maupun kering, didominasi oleh genotip TT (49%) diikuti oleh genotip GT (41%) dan genotip GG (10%). Dari 100 pasien didiagnosis AMD, jumlah pasien yang didiagnosis AMD tipe basah, kering, dan campuran, ialah sebanyak 90, 4, dan 6 pasien.

Kesimpulan: Profil *single nucleotide polymorphism* (SNP) rs10490924 gen ARMS2 pada pasien *age related macular degeneratio* (AMD) di Yogyakarta (n=100), didominasi oleh genotip TT.

Kata Kunci: *Single Nucleotide Polymorphism*, *Age Related Maculopathy Susceptibilty* 2, degenerasi makula terkait umur, SNP, gen ARMS2, AMD

ABSTRACT

DESCRIPTIVE STUDY OF rs10490924 SNP ON ARMS2 GENES FROM PATIENTS DIAGNOSED AMD IN YOGYAKARTA

Background:
Age-related macular degeneration is a degenerative condition that affects the macula in the eye which causes the loss of one's central vision in the elderly, with a prevalence of 8.7% in the world. The etiology of AMD is multifactorial, that involves the interaction between genetic and environmental factors. The main genetic factors involved in the pathogenesis of AMD are CFH and ARMS2 / HTRA1. Association between ARMS2/HTRA1and AMD incidence have been extensively studied in Asian populations, such as in Japanese and Chinese races. Although the polymorphism of ARMS2 (rs10490924) and HTRA1 (rs11200638) significantly associated with AMD, each effect cannot be distinguished using statistical analysis, but the SNP rs10490924 (ARMS2 A69S) alone mostly can explain the relationship between 10q26 chromosomal region and AMD. However, at present, ARMS2 gene contribution to AMD incidence in Indonesia has not been established yet and there is not enough data about SNP ARMS2 gene profile, so it has to be explained further.

Purpose: This study aimed to find out the SNP profile of ARMS2 gene in AMD patients.

Metode: This study is a descriptive that uses secondary data in the form of demographic data and distribution of Single Nucleotide Polymorphism (SNP) on ARMS2 genes of AMD patients in 3 hospitals namely, RSUP Dr. Sardjito, RSPAU dr. S. Hardjolukito, and RSM Dr. Yap Yogyakarta. Samples were taken using the consecutive sampling method in the period August 2016 to October 2017. The peripheral blood sample of AMD patients were tested in the Integrated Laboratory of the Faculty of Medicine, Gadjah Mada University by using Polymerase Chain Reaction (PCR) method.

Result: The single nucleotide polymorphism (SNP) profile rs10490924 on ARMS2 gene of 100 patients diagnosed with age related macular degeneration (AMD), both wet and dry type, were dominated by TT genotypes (49%) followed by GT genotypes (41%) and GG genotypes (10%). Of 100 patients diagnosed with AMD, the number of patients diagnosed with wet, dry, and mixed AMD were 90, 4, and 6 patients.

Conclusion: Based on the results of this study, it can be concluded that the single nucleotide polymorphism (SNP) profile rs10490924 ARMS2 gene in patients (n = 100) diagnosed with age related macular degeneration (AMD) from Yogyakarta, were dominated by TT genotypes.

Key Words: *Single Nucleotide Polymorphism*, *Age Related Maculopathy Susceptibilty* 2, Age-related Macular Degeneration, SNP, ARMS2 gene, AMD