

ABSTRACT

Latar Belakang: *Age-related Macular Degeneration (AMD)* merupakan gangguan pada mata khususnya makula yang bersifat kronis, progresif, degeneratif, meningkat prevalensinya setelah usia 50 tahun, dan ditandai dengan hilangnya lapang pandang sentral. AMD merupakan penyebab kebutaan keempat paling banyak setelah katarak, glaukoma, dan kelainan refraksi yang tidak dikoreksi. Pada tahun 2020, terdapat 1,8 juta kasus AMD global dan 492 ribu kasus pada wilayah Asia Tenggara, Asia Timur, dan Oseania. AMD merupakan penyakit multifaktorial dan berkaitan dengan gaya hidup, status sosioekonomi, usia, genetik, dll. Faktor genetik memiliki pengaruh tinggi terhadap patogenesis AMD. Penelitian sebelumnya menunjukkan adanya hubungan yang kuat antara single nucleotide polymorphisms (SNPs) pada gen CFH dengan kejadian AMD di populasi Asia seperti Jepang, Tiongkok, dan Korea. Namun, hingga saat ini belum ada penelitian yang luas mengenai profil kejadian variasi gen CFH terhadap risiko kejadian AMD pada populasi di Indonesia. Penelitian ini bertujuan untuk menginvestigasi profil variasi gen CFH rs3753394 pada pasien AMD di Indonesia.

Tujuan: Mengetahui profil variasi genetik CFH rs3753394 pada pasien *age-related macular degeneration (AMD)* di populasi Indonesia.

Metode: Penelitian ini merupakan studi deskriptif pada 15 subjek yang direkrut dari pasien AMD poliklinik mata RSUP Dr. Sardjito, RS Mata YAP, dan RS Hardjolukito Yogyakarta. Genotipe varian rs3753394 gen CFH diidentifikasi dengan melakukan *polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP)*.

Hasil: Dari 15 subjek penelitian yang merupakan pasien AMD, terdapat sebanyak 9 subjek penelitian (60%) memiliki polimorfisme pada gen CFH rs3753394 dan berhubungan dengan munculnya penyakit AMD. Analisis multivariat untuk seluruh data karakteristik dasar subjek penelitian dengan munculnya alel risiko pada subjek penelitian menunjukkan nilai-P >0,05.

Kesimpulan: Profil variasi gen CFH rs3753394 pada 15 pasien AMD yang didapatkan menunjukkan sebanyak 40% subjek AMD memiliki alel C/C, 46,7% memiliki alel C/T dan 13,3% subjek memiliki alel T/T.

Kata Kunci: *Age-related macular degeneration*, profil variasi, *complement factor H*, rs3753394, *PCR*

ABSTRACT

Background: Age-related Macular Degeneration (AMD) is a chronic, progressive, degenerative disorder of the eyes, particularly affecting the macula. Its prevalence increases after the age of 50 and is characterized by the loss of central vision. AMD is the fourth leading cause of blindness, following cataracts, glaucoma, and uncorrected refractive errors. In 2020, there were 1.8 million global cases of AMD, with 492 thousand cases in the Southeast Asia, East Asia, and Oceania regions. AMD is a multifactorial disease linked to lifestyle, socioeconomic status, age, genetics, and more. Genetic factors have a significant influence on AMD pathogenesis. Previous research has indicated a strong association between single nucleotide polymorphisms (SNPs) in the CFH gene and AMD occurrence in Asian populations such as Japan, China, and Korea. However, there has been no extensive study on the profile of CFH gene variations and their impact on AMD risk in the Indonesian population. This study aims to investigate the profile of the CFH gene variation rs3753394 in AMD patients in Indonesia.

Objective: To determine the variation profile of CFH gene rs3753394 in AMD patients.

Method: This study is a descriptive study involving 15 subjects recruited from AMD patients at the eye clinics of Dr. Sardjito General Hospital, YAP Eye Hospital, and Hardjolutito Hospital in Yogyakarta. The genotypes of the rs3753394 variant of the CFH gene were identified by performing polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP).

Results: Out of the 15 research subjects who were AMD patients, a total of 9 research subjects (60%) had polymorphism in the CFH gene rs3753394, which was associated with the occurrence of AMD. Multivariate analysis for all basic characteristic data of the research subjects with the emergence of risk alleles in the research subjects showed a P-value >0.05.

Conclusion: The profile of CFH gene variation rs3753394 in 15 AMD patients reveals that 40% of AMD subjects have the C/C allele, 46.7% have the C/T allele, and 13.3% of subjects have the T/T allele.

Keywords: Age-related macular degeneration, variation profile, complement factor H, rs3753394, PCR