INTISARI

LINKAGE DISEQUILIBRIUM GEN *COMPLEMENT FACTOR H* (CFH) PADA PASIEN *AGE-RELATED MACULAR DEGENERATION* (AMD) DI YOGYAKARTA, INDONESIA

Latar Belakang: *Age-Related Macular Degeneration* (AMD) merupakan penyakit degeneratif progresif yang dapat menyebabkan gangguan penglihatan nonreversibel. AMD menjadi salah satu penyebab utama gangguan penglihatan di seluruh dunia, dengan insidensi yang diprediksi akan mencapai jumlah 288 juta pada tahun 2040. Hingga saat ini, prevalensi AMD dilaporkan mencapai 6% pada populasi Asia berusia 40 hingga 79 tahun. Studi telah menunjukkan adanya keterlibatan gen *complement factor H* (CFH) di kromosom 1q32 pada patogenesis AMD. Diperlukan pemetaan secara lebih mendalam pada mutasi gen CFH untuk memperoleh estimasi lokasi spesifik gen yang berperan dalam patogenesis AMD, di mana interpretasi nilai *linkage disequilibrium* (LD) dapat berperan memetakan secara detail keterlibatan komponen faktor genetik tersebut dalam patogenesis penyakit genetik kompleks. Data LD dapat menunjukkan estimasi lokasi kausatif dan derajat keterhubungan alel antarlokus pada penyakit tertentu pada suatu populasi. Hingga saat ini, data LD gen CFH pada penyakit AMD masih belum banyak diperoleh dan memerlukan penelitian lebih lanjut.

Tujuan: Penelitian ini bertujuan untuk mengetahui nilai dan interpretasi *linkage disequilibrium* gen CFH pada populasi pasien AMD di Yogyakarta, Indonesia.

Metode: Desain penelitian berupa studi deskriptif. Subjek penelitian adalah 15 pasien dengan diagnosis AMD dari poliklinik mata RSUP Dr. Sardjito, RS Mata Dr. YAP, dan RSPAU Hardjolukito di Yogyakarta, Indonesia. Metode pengambilan sampel subjek penelitian adalah *consecutive sampling*. Identifikasi polimorfisme DNA dilakukan menggunakan metode PCR-RFLP pada penanda gen *single nucleotide polymorphisms* (SNP) yang berasosiasi dengan gen CFH, yakni rs3753394, rs1410996, dan rs1065489. Analisis frekuensi genotipe dan *linkage disequilibrium* dilakukan sesuai dengan data frekuensi alel yang diperoleh.

Hasil: Penelitian menunjukkan nilai D’ sebagai ukuran *linkage disequilibrium* untuk rs3753394 terhadap rs1065489 dan rs1410996 terhadap rs1065489 bernilai 1,000.

Kesimpulan: Terdapat tingginya kemungkinan rs3753394 dan rs1065489, serta rs1410996 dan rs1065489 untuk diwariskan secara bersamaan dan tidak dipisahkan oleh rekombinasi.

Kata kunci: *Age-related macular degeneration*, *linkage disequilibrium*, CFH rs3753394, CFH rs1410996, CFH rs1065489

ABSTRACT

LINKAGE DISEQUILIBRIUM OF COMPLEMENT FACTOR H (CFH) GENE IN PATIENTS WITH AGE-RELATED MACULAR DEGENERATION (AMD) IN YOGYAKARTA, INDONESIA

***Background**: Age-Related Macular Degeneration (AMD) is a progressive degenerative disease that cause irreversible visual loss. AMD is one of the main causes of visual impairment worldwide, with an incidence predicted to reach 288 million by 2040. To date, the prevalence of AMD has been reported to reach 6% in the Asian population aged 40 to 79 years. Research have shown the significance of complement factor H (CFH) gene in 1q32 chromosome as one of the pathogenesis pathway in AMD. Further mapping around the CFH gene polymorphisms can be helpful to obtain the estimates of specific gene locations that show the significance in the pathogenesis of AMD, where interpretation of linkage disequilibrium (LD) can be used to map the involvement of these genetic factor components on the pathogenesis of such complex genetic diseases. LD data can be useful to indicate the causative location and the degree of connectedness between loci in certain diseases within a population. Thus far, there has not been much research done regarding values for linkage disequilibrium in the CFH gene associated with AMD therefore demanding further research in this matter.*

***Objective**: This study aims to determine the linkage disequilibrium value and interpretation for the CFH gene within a population of AMD patients in Yogyakarta, Indonesia.*

***Method**: The study design is a descriptive study. The study includes 15 subjects diagnosed with AMD in RSUP Dr. Sardjito, RS Mata Dr. YAP, and RSPAU Hardjolukito in Yogyakarta, Indonesia. Consecutive sampling was done as the sampling method for the subjects. This study identified DNA polymorphisms using PCR-RFLP method to analyze single nucleotide polymorphisms (SNP) associated with the CFH gene, involving rs3753394, rs1410996, and rs1065489. Genotype frequencies as well as LD values were analyzed using the alelle frequency data obtained.*

***Result**: The study shows D' value as the measure of linkage disequilibrium as high as 1,000 for rs3753394 to rs1065489 and also rs1410996 to rs1065489.*

***Conclusion**: The result shows a high possibility for rs3753394 and rs1065489, as well as rs1410996 and rs1065489 to be co-inherited and not be separated by recombination.*

***Keywords** : Age-related macular degeneration, linkage disequilibrium, CFH rs3753394, CFH rs1410996, CFH rs1065489*