ABSTRACT

BACKGROUND: Primary Open-Angle Glaucoma (POAG) is a chronic opticneuropathy influenced by genetic factors. Mutations in the lysyl oxidase-like 1 (LOXL1) gene, specifically the single nucleotide polymorphism (SNP) variant rs1048661, have been linked to glaucoma risk through impairment of extracellular matrix homeostasis in the trabecular meshwork. Objective: To describe the LOXL1 rs1048661 gene mutation profile in Indonesian POAG patients. Method: This was an observational descriptive study. Samples included 5 POAG patients and 5 non-POAG controls at RSUP Dr. Sardjito. Genetic analysis was performed using targeted next-generation sequencing (NGS) on the Oxford Nanopore MinION platform.
Objective: This study aimed to describe the gene mutation profile of the rs1048661 SNP in patients with Primary Open-Angle Glaucoma (POAG) and healthy individuals within an Indonesian population.
Results: The study analyzed 10 subjects, comprising 5 POAG patients and 5 non-POAG controls. The median age of participants was 65 years, and 70% of the cohort was female. No statistically significant differences were observed between the POAG and control groups regarding age ($p = 0.46$), blood pressure (systolic $p = 0.25$; diastolic $p = 0.60$), or BMI ($p = 0.06$). Targeted gene sequencing revealed that the rs1048661 variant occurred in 80% (4 out of 5) of the POAG group compared to 20% (1 out of 5) of the control group. All detected mutations were identified as G>T missense variants located at position Chr15:73927205.
Conclusion: The high frequency of the rs1048661 variant among POAG patients compared to controls suggests a potential association between this genetic marker and the susceptibility to Primary Open-Angle Glaucoma in the Indonesian population. Despite the exploratory nature of the study, these preliminary results highlight the potential role of LOXL1 genetic alterations in disease pathogenesis and underscore the need for larger studies to validate its use as a clinical biomarker for early detection and genetic risk profiling.
Key words: Glaucoma, Primary Open-Angle Glaucoma (POAG), LOXL1, rs1048661, Targeted Sequencing