ABSTRACT

Background: Primary Open-Angle Glaucoma (POAG) is a chronic neurodegenerative disease characterized by progressive optic nerve damage, leading to irreversible visual impairment. The pathogenesis of POAG is multifactorial, involving complex interactions between genetic and environmental factors. Genetic studies have identified *CDKN2B*/*CDKN2B-AS1* at the 9p21 locus as important contributors to POAG susceptibility through their roles in cell cycle regulation and retinal ganglion cell survival. The rs1063192 polymorphism located in the 3′ untranslated region (UTR) is thought to influence gene expression without directly altering protein structure, thereby potentially contributing to POAG risk. Further understanding of the role of this genetic variation may support the development of precision medicine approaches in glaucoma management.

Objective: To investigate the association of the CDKN2B/CDKN2B-AS1 gene mutation in Primary Open-Angle Glaucoma patients at Dr. Sardjito General Hospital, Dr. S. Hardjolukito Air Force Hospital, and Dr. Yap Eye Hospital, Yogyakarta.

Methods: This study utilized a descriptive method and employed secondary data obtained from medical records.

Results: A total of 10 participants were included (5 POAG patients and 5 non-POAG controls). The rs1063192 variant in the *CDKN2B/CDKN2B-AS1* gene was detected in 100% (5/5) of POAG patients and 80% (4/5) of non-POAG individuals. This variant is classified as a 3′UTR variant that does not directly alter protein structure but may influence gene expression involved in POAG pathogenesis.

Conclusion: The rs1063192 variant in the *CDKN2B/CDKN2B-AS1* gene was frequently detected in POAG patients but was also present in most controls. These findings suggest that rs1063192 is not a specific single genetic marker for POAG but may act as a genetic predisposition factor requiring further investigation in combination with other genetic variants and risk factors in the Indonesian population.

Keywords: POAG, CDKN2B, CDKN2B-AS1, Mutation, Personalized medicine, Genetic susceptibility