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The Effect of A3243G Mutation of Mitochondrial DNA to the Clinical Features of Type-2 Diabetes Mellitus and Cataract

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Abstract

Type-2 diabetes mellitus (type-2 DM) and cataract are some of the clinical features associated with mitochondrial diseases, the most common mutation is the A3243G in the mitochondrial $tRNA^{Leu(UUR)}$ gene (mtDNA). However, the role of this mutation to type-2 DM and cataract have remained unclear because it was found in these phenotypes plus one or more features of mitochondrial disease such as deafness, myopathy, and cerebellar ataxia. Therefore, the aim of this research are to study the role of A3243G mutation in type-2 DM and cataract without additional features. mtDNA was obtained by lysis from lens and urine epithelial cells. The A3243G mutation was examined using PCR-amplification of specific allele (PASA) method. Here, we showed that A3243G mutations were found in 20 of 57 patients, i.e. 11 of 19 cataract and type-2 DM (Group I), 5 of 16 type-2 DM (Group II), and 4 of 22 pure cataract (Group III patients). Based on statistical analysis using chi-square, the result of this research showed that the A3243G mutation associated with type-2 DM and cataract ($p<0,001$). The study is expected to provide an understanding of the relationship between mutations in mtDNA with cataract disease and type-2 diabetes and this understanding could be used as a basic for further scientific research. On the basic of the mutations found in Indonesia can be developed to the level of molecular methods of diagnosis and selection of appropriate treatment.

Keywords: A3243G Mutation, mtDNA, Clinical Features, Type-2 Diabetes Mellitus, Cataract