Abstract

Hypercholesterolemia is a risk factor for cardiovascular disease. Alterations in the genes encoding low density lipoprotein receptor (LDLR) and its ligand, apolipoprotein B-100 (ApoB-100) are associated with hypercholesterolemic phenotype. The aim of this study was to investigate mutations in exon 26 of ApoB-100 gene to contribute to the diagnosis of familial defective apolipoprotein B (FDB). The mutation was investigated by PCR-SSCP and sequencing of a region of 345 bp of exon 26 of ApoB-100 gene in 322 hypercholesterolemic patients. A Arg3480Pro heterozygous mutation was detected in a patient, however this variant was associated with moderate hypercholesterolemia, whereas a silent mutation in codon 3517 of three individuals was identified. Hypercholesterolemic phenotype could be modulated by interaction with alleles versions of the APOE gene identified. The molecular diagnosis of FDB can contribute to the development of individualized therapies and new therapies to reduce cardiovascular risk in affected individuals.

Keywords

Hypercholesterolemia, ApoB-100, familial defective apolipoprotein B, Arg3480Pro mutation