## Abstract

Myocardial infarction (MI), more often known as a "heart attack," occurs when a clot in the luminal coronary artery stops blood flow to a portion of the heart, killing the myocardium in that area. Many environmental variables, genetic factors, and their interactions may be relevant for the pathogenesis of MI, even if the exact etiology of MI has not been established. Age, cigarette use, diabetes, high blood pressure, obesity, high levels of HDL-C and total cholesterol (TC), and low levels of LDL-C in the blood all contribute to the risk of MI. Therefore, the study was conducted to identify the susceptibility of the Apo B gene in MI patients of the Pakistan population. For this purpose, a case-control study was conducted based on 100 patients heaving MI patients and 50 healthy control samples. According to the WHO criteria all the patients were clinically diagnosed with myocardial infarction and assessed for clinical parameters. Genomic DNA was isolated from the blood through manual extraction. Primers were optimized and genotyping was done by PCR which was followed by DNA sequencing and RFLP. The current study looked at two SNPs of the APO B gene at location codon 4154 G/A (rs1801701) and codon 2488 G/A (rs1042031). rs1801701 and rs1042031 SNPs cause the substitution of guanine into adenine in both SNPs. Carriers of minor allele A of SNP rs1801701 ($p=0.000$) and carriers of minor allele A of rs1042031 ($p=0.0006$) had a significantly increased risk of MI. Haplotype analysis also showed that the AA (rs1801701 and rs1042031) haplotype has a significantly increased risk of MI (OR = 3.845). Our findings confirm the association of Apo B gene mutations with the risk of MI in the Pakistani population.

**Key words:** APOB, SNPs, Genetic polymorphisms, Myocardial Infarction, Pakistani Population