KIT FOR THE GENOTYPING OF THE GENETIC POLYMORPHISM S447X OF THE LPL GENE

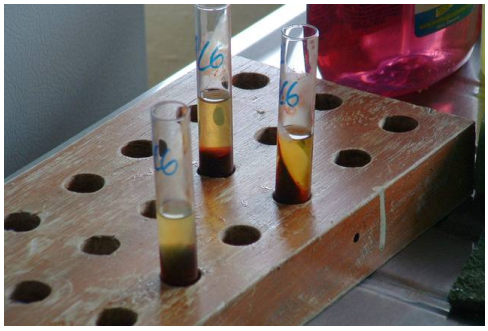
Description:

The enzyme LPL (lipoprotein lipase) plays a key role in lipid metabolism as it hydrolyzes the bulk of triglycerides present in the heart from chylomicrons and very low density lipoproteins (VLDL) that circulate in plasma. Different studies have shown the implication of the gene in question in the development of arteriosclerosis and the appearance of different dyslipidemia of a genetic basis. Of the variants described in the gene, the S447X polymorphism has been associated with lower levels of triglycerides and higher concentrations of HDL cholesterol and therefore with a protective effect against arteriosclerosis and metabolic syndrome. However, the known genotyping methods present a series of drawbacks that are partially or completely solved by the present invention, referring to a set of primers, probes, procedure and kit for the genotyping of the S447X genetic polymorphism of the LPL gene. The basic foundation of the present invention consists of a single PCR reaction in which the 5 'exonuclease activity of the Taq polymerase enzyme is used. Four oligonucleotides are present in the PCR reaction: two specific primers that weaken the polymorphism of interest and two fluorogenic linear probes, specific for each allele. These probes are labeled at the 5 'end with a reference fluorochrome, different for each probe, and a quenching molecule at the 3'. When the probes are intact, the signal emitted by the excitation of the reference fluorochrome is captured by the quenching molecule, due to the physical proximity between them, and therefore is not detected. The fluorescent signal, different for each allele, is detected when the probe hybridizes with the totally complementary allele and the reference fluorochrome is released, by the 5 '? 3' exonuclease activity of the polymerase, during the cycles of the PCR reaction.

Keywords:
Pcr Techniques, Genotyping Kit, Arteriosclerosis, Genotyping, Lpl Gene, S447x Genetic Polymorphism, Metabolic Syndrome

Sectors:
Biotechnology, Health

Areas:
Health Sciences, Diagnosis, Biotechnology, Genetics



Advantages:

The main advantages of the present invention are: -High speed that allows large-scale genotyping since the PCR reaction and the detection of the fluorescent signal are simultaneous. -Automated genotype assignment and immediately obtainable at the end of the reaction. -High sensitivity that allows samples to be genotyped using very low concentrations of DNA. -Lower risk of contamination as it is a homogeneous test.

Uses and Applications:

This technology is useful for the study of genetic variability that affects most common diseases, being of great interest to the biomedical field.

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