LPL Gene

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Lipoprotein lipase

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1. Normal Function

The *LPL* gene provides instructions for making an enzyme called lipoprotein lipase. This enzyme is found primarily on the surface of cells that line tiny blood vessels (capillaries) within muscles and in fatty (adipose) tissue. Lipoprotein lipase plays a critical role in breaking down fat in the form of triglycerides, which are carried from various organs to the blood by molecules called lipoproteins.

Lipoprotein lipase breaks down triglycerides carried by two different types of lipoproteins, which bring fat to the bloodstream from different organs. Fat from the intestine, which is taken in from the diet, is transported to the bloodstream by lipoproteins called chylomicrons. Another type of lipoprotein called very low density lipoprotein (VLDL) carries triglycerides from the liver to the bloodstream. When lipoprotein lipase breaks down triglycerides, the fat molecules are used by the body as energy or stored in fatty tissue for later use.

2. Health Conditions Related to Genetic Changes

2.1. Familial lipoprotein lipase deficiency

More than 220 mutations in the *LPL* gene have been found to cause familial lipoprotein lipase deficiency. This condition disrupts the normal breakdown of triglycerides in the body, resulting in an increase of these fats. The most common mutation in people of European ancestry replaces the protein building block (amino acid) glycine with the amino acid glutamic acid at position 188 in the enzyme (written as Gly188Glu or G188E). Mutations that cause familial lipoprotein lipase deficiency reduce or eliminate lipoprotein lipase activity, which prevents the enzyme from effectively breaking down triglycerides in the bloodstream. As a result, triglycerides attached to lipoproteins accumulate in the blood and tissues, leading to inflammation of the pancreas (pancreatitis), enlarged liver and spleen (hepatosplenomegaly), fatty deposits in the skin (eruptive xanthomas), and the other signs and symptoms of familial lipoprotein lipase deficiency.

2.2. Other disorders

Certain variations in the *LPL* gene have been shown to influence the levels of fats in the bloodstream. The *LPL* gene variants likely result in the production of lipoprotein lipase enzymes with altered abilities to break down triglycerides. In some cases, the enzyme is overactive, resulting in low fat levels. In other cases, the enzyme is impaired, resulting in increased fat levels, a condition called hyperlipidemia. Individuals with hyperlipidemia are at greater than normal risk of developing atherosclerosis, a condition in which fatty deposits accumulate on artery walls. This fatty material hardens over time, eventually blocking the arteries and increasing the chance of having a heart attack or stroke. It is unclear how much of a role *LPL* gene variants play in the development of atherosclerosis, as a large number of genetic and environmental factors determine the risk of developing this complex condition.

3. Other Names for This Gene

- clearing factor lipase
- diacylglycerol lipase
- LIPD
- postheparin lipase

- triacylglycerol protein acylhydrolase

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