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PERSPECTIVE

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Signs and Symptoms of Familial Hypercholesterolemia

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Description

A genetic condition known as Familial Hypercholesterolemia (FH) is characterized by very high levels of low-density lipoprotein in the blood as well as early cardiovascular disease. The majority of mutations reduce the amount of liver LDL receptors that are functional. Their elevated cholesterol levels are less susceptible to the kinds of cholesterol control strategies that are often more effective in persons without FH because the underlying body biochemistry is slightly different in people with FH. However, treatment is typically successful.

Type 2 familial dyslipidemia is the classification for FH. Familial dyslipidemia can be divided into five different categories based on both the changed lipid profile and the underlying genetic defect. For instance, type 2 is high LDL. Defects in the metabolism of chylomicrons, triglycerides, and other cholesterol-containing particles like VLDL and IDL are among the others. Apolipoprotein B, which is the portion of LDL that connects with the receptor, and the LDLR gene, which encodes the LDL receptor protein, which typically removes LDL from the circulation, are the two most frequently affected genes; mutations in other genes are uncommon. Cardiovascular disease may manifest in people with one defective copy of the LDLR gene between the ages of 30 and 40. Children who have two faulty copies may have severe cardiovascular disease. Statins, bile acid sequestrants, or other lipid-lowering medications that lower cholesterol levels are typically used to treat heterozygous FH. Genetic counselling is typically provided for new patients. When medical therapy for homozygous FH is unsuccessful, further therapies including LDL apheresis and possibly liver transplantation may be needed.

Signs and symptoms

Physical signs: Normal high cholesterol levels do not manifest any symptoms. On the body, yellow deposits of cholesterol-rich fat can be visible in a number of locations, including the tendons in the hands, elbows, knees, and feet, especially the Achilles tendon, as well as the area surrounding the eyelids and the outer edge of the iris.

Cardiovascular disease: Atherosclerosis, the fundamental cause of cardiovascular disease, is brought on by an accelerated buildup of cholesterol in the walls of arteries. The development of coronary artery disease in FH patients most frequently occurs far earlier than would be anticipated in the general population. Angina pectoris or heart attacks could result from this. Less frequently, brain arteries are impacted; this may occasionally cause strokes or transient ischemic episodes. People with FH who smoke are more likely to develop peripheral artery occlusive disease, which can result in decreased blood flow to the feet and symptoms including walking-related calf pain that goes away with rest. Age, smoking, diabetes, high blood pressure, and a family history of cardiovascular disease all raise the risk of atherosclerosis.

Pathophysiology

After 2.5 days of normal circulation, LDL cholesterol is taken up and digested by the liver cells when the apolipoprotein B part of LDL cholesterol attaches to the LDL receptor on those cells. LDL is eliminated from the circulatory system as a result of this procedure. The HMG-CoA reductase pathway inhibits the liver's ability to synthesise cholesterol. In FH, LDL receptor function is diminished or missing, and LDL circulates for an average of 4.5 days. As a result, there is an abnormally high quantity of LDL cholesterol in the blood, whereas other lipoprotein levels are normal. Reduced LDL particle binding to the receptor results in higher levels of LDL cholesterol in ApoB mutations. In PCSK9 and ARH mutations, the exact mechanism by which the LDL receptor becomes dysfunctional is unknown.

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