

# [Familial hypercholesterolemia inheritance pattern](https://assignbuster.com/familial-hypercholesterolemia-inheritance-pattern/)

### Familial Hypercholesterolemia

Familial hypercholesterolemia (FH) is an autosomal dominant disorder characterised by high cholesterol levels, specifically low-density lipoprotein (LDL) and an increased risk of having a heart attack early in life (MEDPED 2009). Heterozygous FH is a common condition which occurs in 1 in 500 people. FH is an inherited disorder, you are born with it or your not (Citkowitz 2009). FH was the first inherited disorder to be acknowledged as being a cause of heart attacks (King 2009). People with FH are treated through diet and medication and in extreme cases liver transplants (MEDPED 2009).

The gene mutation that causes FH is located on chromosome number 19, which contains the LDR receptor, which is responsible for clearing up LDL from the blood stream (National Human Genome Research 2009). Inherited FH results from mutations in the lipoprotein receptor gene (LDLR), the apolipoprotein B-100 gene (APOB) and the recently found proprotein convertase subtilsin/kexin type 9 gene (PCSK9) (Austin et al. 2004). This condition has an autosomal dominant pattern of inheritance, meaning one copy of an altered gene is needed to cause the disorder. Heterozygous FH occurs when one copy of the altered gene is received from an affected parent and a normal gene is received from the other parent. If a heterozygote affected person had children there is a 50% chance that the mutated gene will be inherited. Homozygous FH, which is quite rare, occurs when both parents pass on an altered gene to a child (Genetics Home Reference 2007). This condition is more severe and usually results in a heart attack or death occurring before the age of 30 (National Human Genome Research 2009). If FH is caused by a mutation in the LDLRAP1 gene, the condition is inherited in an autosomal recessive pattern, meaning both parents must of carried the disorder (Genetics Home Reference 2007).

Cholesterol is found in all cells of the body and is carried in the blood stream in lipoproteins. Cholesterol is a fat-like substance that is found is some foods. There are two main kinds of lipoproteins, LDL and HDL, low and high density lipoproteins. LDL normally bind to the receptor, which regulates cholesterol metabolism in two ways, it reduces the rate of cholesterol synthesis and it increases the rate at which fibroblasts breakdown the protein component of LDL. Cells from those with FH show a deficiency in the number of functional LDL receptors, resulting in the overproduction of cholesterol and a reduction in the ability to breakdown the protein in LDL. The body needs a certain amount of cholesterol to work properly and uses it to produces hormones, vitamin d and other substances involved in digestion. In a normal person, LDL is removed by the liver. People with FH have high levels of LDL cholesterol because they are unable to remove it from the liver. HDL is sometimes referred to as good cholesterol, carrying cholesterol from body parts to the liver. Higher levels of HDL reduce people’s chances of having a heart attack (National Human Genome Research 2009).

The main aim in treatment of FH is to lower the risk of atherosclerotic heart disease by lowering levels of LDL cholesterol in the blood stream (National Human Genome Research 2009). FH is treated through diet modification, exercise and certain medications which can reduce blood-fat levels to a safer level. People can reduce their fat intake by eating less beef, pork and lamb, avoiding coconut oil and choosing low-fat dairy products, Cholesterol intake is reduced by avoiding organ meats, egg yolks and sources of animal-derived saturated fats. Another way to lower cholesterol levels is exercise, specifically to reduce weight. If diet, exercise and weight-loss programs do not help lower cholesterol levels, drugs are implemented. There are various drugs available including cholestryamine, colestipol, fenofibrate, gemfibrozil, nicotinic acid and statin drugs. In homozygous FH some people may even need surgery such as a liver transplant to reduce cholesterol levels to a harmless level. Those people who are diagnosed with FH early and receive appropriate treatment may not suffer heart attacks. Heterozygous FH is less severe in comparison to homozygous FH and has a reduced chance of having a heart attack. Another factor that would influence the risk of a heart attack would be smoking because of the damage that occurs to the lining of the arteries (Health NY Times 2009) .

Familial hypercholesterolemia is a common inherited condition that increases the risks of having a heart attack at a young age. Unlike in a normal person, the liver is unable to remove excess amounts of LDL. If the disorder is treated early and the patient modifies their lifestyles, the individual may not suffer a heart attack.

### References:

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