

## WHAT IS FAMILIAL HYPERCHOLESTEROLEMIA (FH)?

Familial Hypercholesterolemia (FH) is an **inherited cause of high cholesterol and leads to premature coronary heart disease**. FH can be detected in childhood. If treated properly, heart disease caused by FH can be prevented.

**C**oronary heart disease (CHD) is considered premature if present in males before 55 and females before 65 years of age. CHD is considered present in 1st degree relatives with a history of one of more of the following:

- 1) heart attack, 2) angina, 3) angioplasty,
- 4) coronary artery bypass surgery, 5) stroke, or
- 6) sudden death.



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Regardless of who you are or where you or your family members come from, FH is present in 1 out of 250 individuals. It is caused by a genetic variant inherited from one or rarely both parents (You are born with it!). Because this condition is inherited, a parent with FH has a 50% chance of passing on the condition to a child.

Cholesterol is essential for health. It comes from 2 sources: 1) most of it is produced in your body and 2) some comes from the foods and snacks you eat. Individuals with **FH cannot remove cholesterol** efficiently from their bloodstream. As a result, the level of cholesterol becomes very high, causing **cholesterol to accumulate which eventually damages the blood vessels**. Higher levels of cholesterol and longer exposure result in the greatest damage to blood vessels. While children rarely experience symptoms because of their young age, damage to their blood vessels begins as early as 8 years of age!

## HOW DO YOU KNOW IF YOU HAVE FH?

FH can be easily diagnosed with a simple blood test called a “lipid panel”. This test can be performed any time of the day, with or without fasting. Medications are required to lower the cholesterol level because FH is a genetic disorder where your body is unable to clear the cholesterol normally. While helpful and encouraged, genetic testing to confirm the presence of a genetic variant is optional.

Most individuals with FH do not know that they have the condition, and only find out when they, or a loved one experiences a heart attack or undergoes “lipid panel” testing. A “lipid panel” test is recommended for everyone, including children. All first degree (parents, siblings) and extended (grandparents, aunts, uncles) relatives should



**U**niversal Lipid (cholesterol) Screening is recommended by the American Heart Association (AHA), American College of Cardiology (ACC), and American Academy of Pediatrics (AAP):

- at 2 years of age (if family history of premature coronary heart disease)
- at 9-11 years of age (all youth)
- and at 17-21 years of age (all youth)

undergo “lipid panel” testing if a loved one is diagnosed with FH.

Among those with FH, in addition to a heart healthy lifestyle, treatment with a **cholesterol lowering medication** is recommended if the LDL-C or “bad cholesterol” remains 1) 160 mg/dL or higher and there is a family history of premature coronary heart disease or 2) 190 mg/dL or higher in the absence of a known family history of premature coronary heart disease.

## HOW IS FH TREATED?

Unfortunately, **diet and exercise will not treat FH**. Because FH is genetic, your body is unable to clear the LDL-C normally, even if you are eating a healthy diet. Medications are required to lower the LDL-C. Most individuals with FH are born with an abnormally



functioning LDL receptor (LDLR). The LDLR is the structure (protein) responsible for removing cholesterol from the bloodstream. If a diagnosis of FH is suspected or confirmed, meet with your doctor or your child’s doctor to discuss treatment options.

**Early testing and proper treatment of FH saves lives!**

