

## HAS SOMEONE IN YOUR FAMILY EXPERIENCED ANY OF THE FOLLOWING AT A YOUNG (OR PREMATURE) AGE\*:

1. Heart attack?
2. Coronary artery stent placement?
3. Coronary artery bypass grafting?
4. Stroke?
5. Sudden cardiac death?

*\*Before 55 years of age in a male relative or before 65 years of age in a female relative.*

Familial Hypercholesterolemia: Heterozygous and Homozygous Forms

Heart attack (also known as myocardial infarction) that occurs in a “blood related” relative, at a young age, can be the first indication that you and/or other family members have an inherited disorder, **known as familial hypercholesterolemia**. Familial means “family.” Hyper means “high or above normal” levels. Cholesterol is fat in the blood that is largely made by the body that can accumulate to high levels if the body cannot clear (or remove) the cholesterol normally from the blood. Finally, “emia” means blood. Thus, familial hypercholesterolemia is an inherited disorder in which cholesterol cannot be cleared from the blood normally. As a result, these high levels of cholesterol, enter the walls of the arteries and ultimately, contribute to the development of hardened plaque (“atherosclerosis”). When the plaque ruptures, it clogs the arteries and decreases blood flow, resulting in damage to the heart muscle (“heart attack”) or to a portion of the brain (“stroke”).

Fortunately, familial hypercholesterolemia is treatable, but it must be diagnosed in order to be appropriately treated.

## KNOW THE STEPS FOR PROTECTING YOURSELF AND YOUR FAMILY.

### STEP #1



**K**now what familial hypercholesterolemia (FH) is and know the features of FH.

There are 2 forms of FH: homozygous familial hypercholesterolemia and heterozygous familial hypercholesterolemia.

Homozygous FH is the more severe form of FH and is present in 1 in 300,000 to 1 in 1 million persons. “Bad” cholesterol levels (LDL-C) are often 400mg/dL or greater. High LDL-C levels can be detected as early as 2 years of age. Referral to a specialist, such as a cardiologist, endocrinologist, or lipidologist (cholesterol specialist) for treatment is necessary. Heart attack can occur as early as 10- 20 years of age.

Heterozygous familial hypercholesterolemia occurs in 1 in 250 persons. “Bad” cholesterol (LDL-C) levels are often 160mg/dL or above if there is a family history of premature heart disease and 190mg/dL or above if there is no known family history of premature coronary artery disease. Heart attack can occur as early as 35 years of age.

### STEP #2

**K**now that familial hypercholesterolemia (FH) can be diagnosed and treated before disease presents.



Universal Cholesterol Screening is recommended by the American Heart Association (AHA), American College of Cardiology (ACC), and American Academy of Pediatrics (AAP):

- 2 years of age (if family history of premature coronary heart disease)
- 9-11 years of age (all youth)
- 17-21 years of age (all youth)
- Every 3- 5 years in adults > 20 years of age

### STEP #3

**T**reatment of FH can delay or prevent heart disease at a young age.



Healthy diet and regular physical activity are essential for the management of familial hypercholesterolemia.

However, the only effective treatments for FH are with medications or a procedure that temporarily removes the cholesterol from the blood (known as LDL apheresis) but needs to be repeated every 2 weeks. Medications such as statins are required and often are the initial ones used to treat FH. Some individuals may require additional medications to adequately lower the LDL-C (bad cholesterol) such as ezetimibe, PCSK9 inhibitors, inclisiran, bempedoic acid, and evinacumab. Your clinician can determine the best treatment plan for you.