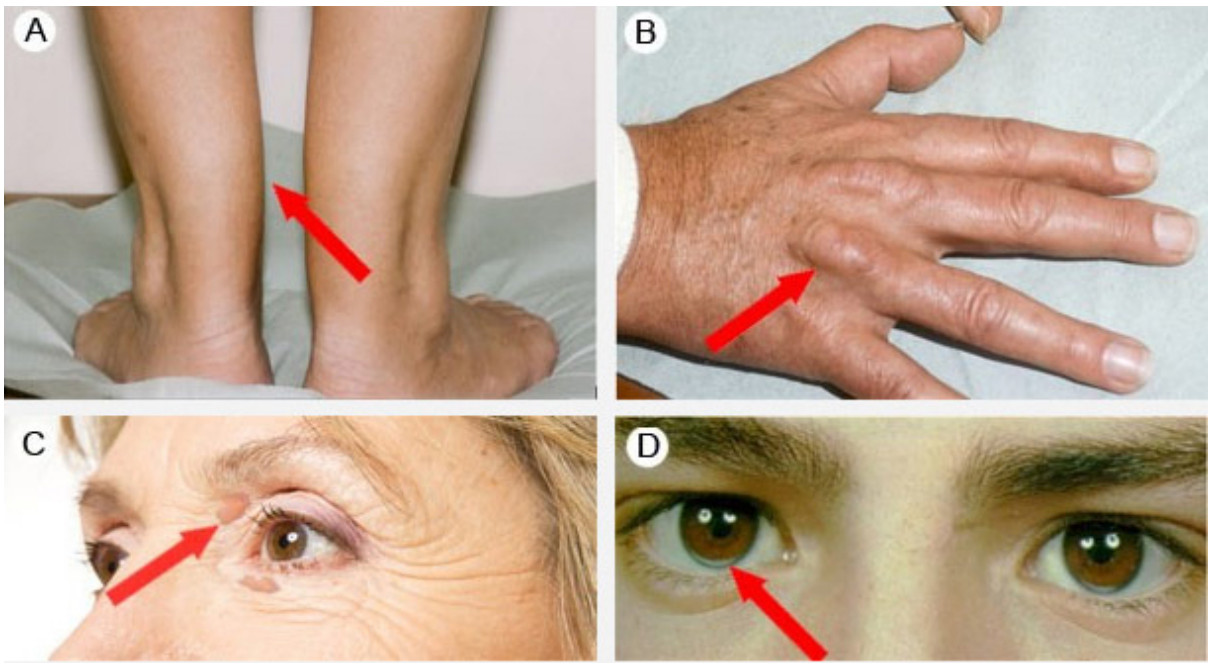
Familial hypercholesterolemia (abbreviated FH) is an inherited disorder that has high cholesterol levels. The levels of low-density lipoprotein (LDL, “bad cholesterol”) in the blood is very high. FH can cause heart attacks and strokes to occur at a younger than usual age.

• Familial means FH runs in families; sometimes it is possible to trace the disease over several generations.
• Hypercholesterolemia means high blood cholesterol.
• The type of cholesterol that is very high in FH is called Low Density Lipoprotein Cholesterol (LDL-C). This can cause an early heart attack.
• Individuals with FH often look perfectly healthy, and may have this disorder without knowing it.

FH patients have mutations (changes) in their genes that limit the body’s ability to remove cholesterol, so that the amount of cholesterol in their bloodstream goes up. Abnormally high levels of cholesterol in the blood can lead to serious and potentially fatal problems with the heart and blood vessels, including heart attack, stroke and even death. FH causes progressive build-up of artery-blocking plaque; patients may not have any symptoms or be aware of the condition. The first symptoms can be a fatal heart attack or stroke.

Diagnosis & Symptoms

FH is usually diagnosed by a blood cholesterol test, called a “lipid panel,” along with personal and family history. Genetic testing may be done, but is usually not necessary. Signs of FH may also be seen during a physical exam. These can include a white ring on the colored part of the eye (d), and cholesterol deposits in the tendons of the ankles (a), hands (b), and elbows. There can also be yellow deposits on the skin around the eyes (c) and ankles.



In families where FH has been diagnosed, it is advised that children are tested for FH at about age 2. This allows parents to make early diet changes to encourage healthy eating. Some children with very high cholesterol levels, or a close family member with a very early heart

attack or stroke, may need treatment with medications to lower their blood cholesterol to a safer level.

Testing close family members enables early detection of the disease.

Treatment

Medicines

A type of medicines, called statins, are the main drugs to lower LDL. In addition, ezetimibe, bile acid sequestrants, and bempedoic acid are other oral medications used to lower LDL cholesterol. A class of medications called PCSK9 Inhibitors are given as injections under the skin every 2 to 4 weeks or every 6months. For homozygous FH, the medications lomitapide and evinacumab can be used.

Diet and Lifestyle

People with FH should follow a heart healthy diet that limits saturated fat, aim to maintain a healthy weight, get 2.5 hours of moderate physical activity or 75 minutes of vigorous physical activity every week, limit alcohol intake, and not smoke or use tobacco products.

LDL Apheresis

LDL apheresis is a medical therapy for patients who are not able to meet their LDL cholesterol goal with medicines and lifestyle changes. This procedure physically removes LDL cholesterol from the blood. Blood is removed from one arm and goes through a special filtering machine that removes LDL cholesterol. The newly filtered blood, with a much lower LDL level, is then returned to the other arm. Medical professionals monitor patients during the procedure, which takes 2 to 3 hours and is generally performed once every other week.

Lipid Specialist

If you have FH and are not able to take a statin medication or to lower your LDL cholesterol adequately with medicines and lifestyle changes, it is important to see a lipid (cholesterol) specialist. In addition, children with FH should also see a lipid specialist. Click here to find a Lipid Specialist in your area.

Resources
https://www.learnyourlipids.com/lipid-disorders/familial-hypercholesterolemia/