Familial Hypercholesterolemia (FH) in Children

What is Familial Hypercholesterolemia (FH)?
Familial Hypercholesterolemia (also called FH) is a genetic disorder that results in severe high levels of blood cholesterol and an increased risk of early heart disease (defined as starting in men before age 55 and in women before age 65). FH is caused by changes in a gene that lowers the body’s ability to remove low-density lipoprotein (LDL) cholesterol (the “bad” cholesterol) from the blood. This makes the levels of LDL cholesterol very high. FH is inherited. In most types of FH, a parent with FH has a 50% chance of passing the gene that causes FH to their child. If someone is found to have FH, their parents, siblings, and children should also have their cholesterol checked.

Children with FH have high blood LDL cholesterol levels that can cause heart disease at an early age. They also often have other family members that have high cholesterol, early heart disease, or heart attacks. For many people, high cholesterol levels are the result of a lifestyle of eating a diet that is high in saturated fat, being overweight, having diabetes, or not getting enough exercise. Children with FH can be normal weight, have a good diet, exercise enough, and still have a high LDL cholesterol level.

Why is FH a problem for my child?
Familial hypercholesterolemia results in rapid atherosclerosis, often starting before 10 years of age. Atherosclerosis happens when fat, cholesterol, and other substances build up in the walls of arteries and form hard structures called plaques. Over time, these plaques can block the arteries and cause early heart disease, heart attacks or strokes. If your child has FH, plaque can build up and narrow the arteries and make them stiffer. These changes make it harder for blood to flow through them. Clots may form in these narrowed arteries and block blood flow. Pieces of plaque can also break off and move to smaller blood vessels, blocking them. The blockage stops blood and oxygen from reaching parts of the body, which can result in damage or tissue death. This is a common cause of heart attack and stroke. If left untreated, these people have up to a 100-fold increased risk for early cardiovascular disease. In 50% of men and 25% of women these cardiovascular issues start by age 50.

How common is FH?
Worldwide, FH happens in about 1 in 200-300 people. In some areas of the world, FH can occur in 1 in 100 people. In children FH is a "silent" disease and does not cause any symptoms.
How is FH diagnosed?
FH can only be diagnosed with a cholesterol blood test. This is one of the reasons checking cholesterol levels in all children at 9-11 years of age is now recommended. Diagnosing FH at a young age is important because the treatment works best when started early before too much extra cholesterol in the blood vessel walls builds up. Early treatment of FH in adults has been shown to reduce their risk for future heart disease. Treating FH in children has been shown to reduce atherosclerosis.

How is FH treated in children?
Eating a healthy diet low in saturated fat and cholesterol can lower LDL cholesterol. For children with FH, this is often not enough. Most children with FH will need to take medicine to lower their cholesterol. Most children need to make these changes throughout their lives.

The most common medicines that lower cholesterol in FH are called statins. They work by decreasing the amount of cholesterol made in the liver. Statins have been used with success to treat adults with high cholesterol. Statins cannot be taken during pregnancy because they may cause birth defects to the unborn baby. If your child takes a statin, part of the treatment is to check blood cholesterol levels, and do other blood tests on a routine basis.

Some children with FH need more than one type of medicine to lower their LDL cholesterol level.

Familial Hypercholesterolemia Websites:
The FH Foundation: www.Thefhfoundation.org
www.Youtube.com/fhjourneys
Patient information from the National Lipid Association: www.Learnyourlipids.com
Make Early Diagnosis to Prevent Early Death (MEDPED): www.medped.org

Please contact AFCH Pediatric Preventive Cardiology Clinic (PPCC) for more information or questions: 608-263-6420

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