What families need to know about inherited high cholesterol in children

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Many parents think of high cholesterol as a health concern that only affects adults. But did you know that around one in 250 children have elevated cholesterol levels that aren't related to diet, exercise or lifestyle?

These kids have a genetic condition called familial hypercholesterolemia, or FH. Even if they eat healthfully and live an active life, they are at risk for developing early heart disease.

Cholesterol is a waxy, fat-like substance found normally in every cell of our bodies. The liver produces cholesterol to keep our cells working smoothly. Cholesterol also supports the production of hormones and vitamins essential for continued good health.

When we eat animal products like dairy, eggs or meat, we take in extra cholesterol. In most cases, our bodies can remove what we don't need. But for people with familial hypercholesterolemia, a mutation in the genetic code gets in the way of this process. As a result, LDL cholesterol (often called "bad" cholesterol) builds up in the arteries from birth. This buildup, called plaque, leads to deposits in the coronary arteries, which causes heart attacks and stroke early in life.

Millions of people worldwide live with familial hypercholesterolemia, but only 30% know they have it.

There are two forms of familial hypercholesterolemia, and they're different based on the way the gene is passed from parent to child. The most common form of FH is known as heterozygous FH (HeFH). A child with HeFH has inherited one abnormal copy of the FG gene from one birth parent. An estimated one in 250 people have HeFH.

When a child inherits an abnormal copy of the FH gene from both parents, they have a condition known as homozygous FH (HofH). HoFH is a rare condition that needs immediate treatment.

Identifying and treating familial hypercholesterolemia early can reduce risks for coronary artery disease by as much as 80%. The process begins with knowing the family health history.

Be sure to tell your pediatrician if a parent, grandparent, aunt or uncle of your child suffered early heart attacks (before age 55 for males and age 65 for females), say doctors. High cholesterol or coronary artery disease in close relatives should be noted, too.

Based on this background, a child's doctor can decide how early to test a child's cholesterol levels. When no family risks are present, the American Academy of Pediatrics (AAP) recommends cholesterol screening starting between age 9 and 11. But when there's inherited heart trouble, screening starts as early as age 2.

If blood tests show a child's LDL cholesterol level is above 160 mg/DL, then familial hypercholesterolemia may be present. Children diagnosed with HoFH often have cholesterol levels that are 400 mg/DL, or more than four times the desired cholesterol level between ages 2 and 19. Extremely high LDL levels of 1,000 mg/dL have been seen in some children with HoFH.

HoFH, the more severe form of inherited high cholesterol, affecting about one in 300,000 people worldwide, but health care professionals are working to widen awareness of the condition. Dangerous buildup of cholesterol caused by HoFH can cause heart attacks in the teen years or younger. In fact, people with untreated HoFH generally succumb to heart disease before age 30.

Having the familial hypercholesterolemia gene does not reflect on diet, fitness level or any other risk factor that can be controlled.

Thanks to continuous research and development, there are many ways to reduce LDL cholesterol levels in kids and adults. The FDA has approved several medications for children, and more are in development. So, the child's medical team will have multiple tools to work with in addressing the child's HoFH.

If a child is diagnosed with HoFH, their pediatrician will recommend a medical specialist focused on heart health and lipids. The child's care plan might include statins, or drugs that reduce the amount of cholesterol made by the liver while helping the liver remove cholesterol already in the bloodstream.

Non-statin medications that work with the liver, small intestine and specific chemical pathways to lower LDL cholesterol might be recommended, or lipoprotein apheresis, a procedure that removes LDL cholesterol from the blood each week.

Along with medication, eating a heart-healthy diet, staying active and avoiding tobacco will be important throughout the child's life. A dietitian may join the child's care team to offer guidance and suggestions for nutritious meals and snacks.