Children's Heart Center of Central Oregon

Familial Hypercholesterolemia

What is it?	**A genetic/inherited mutation that affects the way the body breaks down cholesterol. Most commonly, the genetic mutation affects the body's LDL receptor which makes it harder for the body to remove cholesterol circulating in the blood. Mutations in other genes can also cause familial hypercholesterolemia.**
Whom does it affect?	**1 in 200 adults have this genetic mutation and can pass this along to their children.**
How is it diagnosed?	***The American Academy of Pediatrics recommends that all children have their cholesterol tested between ages 9-11 and again between 17-21.*** **Testing can be performed in children less than 9 years old if there are certain risk factors including a family history of hyperlipidemia or early heart attack. Familial hypercholesterolemia is suspected with significantly elevated LDL and can then be confirmed with genetic testing, which will identify a genetic mutation in cholesterol metabolism.** **Familial hypercholesterolemia is different from a family history of hypercholesterolemia, in which other members of the family can have high cholesterol, but do not have an identified genetic mutation contributing to the high cholesterol.** **If one member of the family has a confirmed genetic mutation for hypercholesterolemia, all first degree or immediate relatives (parents, siblings, children) should also be tested for this genetic mutation.**
How is it treated?	**Typically this type of hypercholesterolemia persists even with dietary and other lifestyle changes. This type of hypercholesterolemia usually requires treatment with a statin and/or other cholesterol-lowering medications. Sometimes multiple medications are required.** **It is important to continue with healthy lifestyle habits including a diet high in fiber and limited in processed foods, as well as participating in at least 1 hour of aerobic exercise daily, even if medications are started.**