Familial Hypercholesterolemia (FH) Fact Sheet

Familial hypercholesterolemia, also known as FH, is a genetic disorder which causes severely elevated cholesterol levels, particularly high levels of low-density lipoprotein (LDL, or "bad cholesterol"). Among people with FH, high cholesterol levels are present in childhood. The condition is caused by a mutation in the LDL receptor and can potentially lead to early heart attack and death, even in otherwise healthy people. While normal cholesterol levels are approximately 200 mg/DL, patients with heterozygous FH (they've inherited the condition from one parent) typically have levels in the range of 350 to 550 mg/dL. Patients who are homozygous (they've inherited FH from both parents) are in the range of 650 to 1000 mg/dL.

FH is a treatable condition. Early heart disease and death caused by FH can be prevented by diagnosing and treating this disorder early in life.

Risk Factors of Familial Hypercholesterolemia

If one parent has FH, there is a 50 percent chance that their son or daughter will also have it.
Certain populations are known to have a higher prevalence of FH, particularly French Canadians and Dutch Afrikaners.

Diagnosis of Familial Hypercholesterolemia

Despite the prevalence of FH, it is underdiagnosed, especially in children.

- Some estimates suggest that only about 20 percent of all FH patients are diagnosed.
- Patients can be screened with a simple blood test called a "lipid panel" for elevated serum cholesterol to determine the presence of FH.
- Universal screening is recommended between the ages of 9 and 11 for all children, or beginning at age two if the child has a family history of high cholesterol or heart disease.
- A genetic test may also be done to identify the abnormal LDL receptor gene.

FH Facts and Figures

- FH is one of the most common inherited metabolic disorders, affecting more than 600,000 Americans, and is more common than type 1 diabetes, cystic fibrosis and Down's Syndrome
- Heterozygous FH occurs in about 1 in every 300-500 people
- Homozygous FH occurs in about 1 in every 1,000,000 people
- The risk of premature coronary heart disease is elevated about 20-fold in untreated FH patients

Treatment of Familial Hypercholesterolemia

FH can be treated, but requires lifelong follow-up, as well as dietary and lifestyle changes to lower LDL cholesterol. It's important that a person is diagnosed at a young age because early changes in diet and eating habits can help reduce the impact of FH later in life and treatment is more effective when started early before cholesterol deposits in blood vessels become too advanced.

- The disease is currently undertreated and estimates indicate that less than half of patients receive appropriate treatment.
- Aggressive lipid lowering is important in order to achieve the target LDL cholesterol reduction of at least 50 percent.
- Patients are typically prescribed statins as an initial treatment to lower cholesterol levels.
- Homozygous FH may require more intense treatment, such as apheresis, a dialysis-like cleaning of the blood.