Familial hypercholesterolemia, a very common cause of early cardiovascular death

Familial Hypercholesterolemia (FH) is a group of genetic disorders that result in severe elevations of blood cholesterol levels. Despite being of one the most common serious genetic disorders, few people know about FH unless their life was affected in some way. The prevalence of FH is 1 in 300 to 500 in many populations, and there are over 620,000 FH patients currently living in the United States. Many of these people are unaware that their risk of premature coronary heart disease is elevated about 20-fold in patients that have not been treated.

There are two different types of FH patients. The most serious type is called the homozygous form. This means that the genetic defect was inherited from both parents. Homozygous FH type occurs in 1 out of every 1,000,000 individuals and is associated with the rapid and premature development of coronary heart disease in childhood or adolescence. Many of these patients never live to see their second or third decade of life. The heterozygous form of FH means that that the genetic defect was inherited from one parent and is much more common. In certain populations such as French Canadians, the prevalence may be as high as 1 in 100.

Total cholesterol levels in the heterozygous form of FH are usually in the range of 350-550 mg/dl and from 650-100 mg/dl in the homozygous form. In addition, a LDL cholesterol level greater than or equal to 250 mg/dl in a person 30 years or more, greater than or equal to 220 mg/dl in a person 20 to 29, and greater than or equal to 190 mg/dl in a person under age 20 should prompt a doctor to strongly consider the diagnosis of FH and obtain a detailed family history. Cholesterol screening should be considered beginning at age 2 for children with a family history of premature cardiovascular disease or elevated cholesterol and current guidelines recommend that all individuals be screened by age 20.

FH is a treatable disease. In addition to diet and lifestyle modification, medications are an essential tool in management. Some patients may require LDL apheresis, which is a method of removing the bad cholesterol from the blood. Some patients with the homozygote form have required liver transplantation to treat the disease.

FH is under diagnosed and undertreated and the consequences of this failure can be devastating. For more information, you can go to www.lipidcenter.com or ask your physician for a simple blood test to measure your cholesterol.