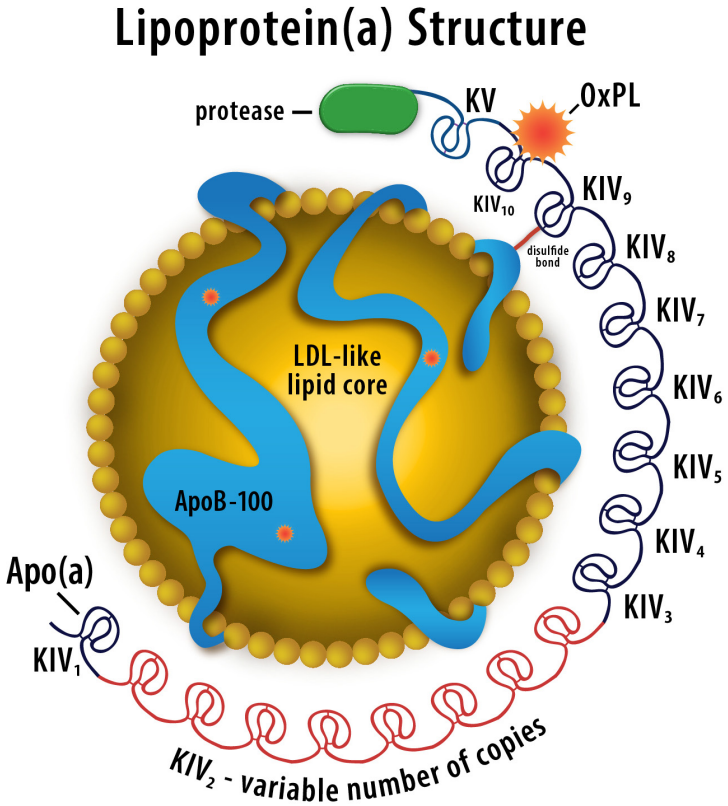
FAMILY HEART FOUNDATION
Lipoprotein(a) & Familial Hypercholesterolemia

Background for Conversations with Family Members

It can come as a surprise to learn that you have elevated Lipoprotein(a), also known as Lp(a) – pronounced "LP little a". While most people understand that having high LDL-Cholesterol (also known as the "bad" cholesterol) can increase the risk for heart disease, chances are that very few of your family members have even heard of Lp(a), and Lp(a) may be new to you as well. Nonetheless, it is important that you share your diagnosis with your family because the truth is, your diagnosis in not just "your diagnosis." It is, simply put, your entire family's diagnosis.

Start with the basics. Although Lp(a) is a very complex lipoprotein that appears to increase the risk for both heart disease and stroke we can understand this lipoprotein by looking at its component parts. Lp(a) looks like LDL cholesterol with some key differences. LDL has a single apolipoprotein B attached on its surface. Lp(a), however, has an apolipoprotein B attached on its surface, and that apolipoprotein B has an apolipoprotein (a) attached to it. Lp(a) also contains oxidized phospholipids (OxPL).

Each of these components come together and results in a lipoprotein than can cause artery blockages (LDL portion), artery clogging (the apolipoprotein (a) portion), and inflammation (the oxidized phospholipid portion). It might help to share the picture below with your family members.



The challenging thing about elevated Lp(a) is it can be invisible. A person can have a totally normal cholesterol panel and still harbor a very high Lp(a). The only way to know if someone is at risk due to a high level, is to measure it. The blood test for Lp(a) is simple and should cost about $30, but it is important to find out if your insurance will cover the test and important to know that some labs charge far more than $30.

Elevated Lp(a) is genetic and caused by a dominant gene. If you have inherited the gene for elevated Lp(a), each of your siblings and children has a 50% chance of having inherited an elevated Lp(a) too. You have elevated Lp(a) because one of your parents has an elevated Lp(a) and passed that gene on to you. Having high Lp(a) leads to an increased risk of early heart disease including heart attacks and strokes.

Whether your family is tight-knit or rarely in touch with each other, you are bound together by your genetics. Communicating with your family about having elevated Lp(a) isn't easy no matter what your relationship is. For some people, sitting down face to face for a discussion is the best approach. For others sending a text, email or letter is better. The most important thing is to provide your family members with accurate information about Lp(a) so they can make decisions about their own health. It is crucial for your relatives to understand that while there is currently only one approved treatment for elevated Lp(a) called lipoprotein apheresis, other therapies are in clinical trials right now. If someone has elevated Lp(a), it is crucial to make sure they manage any other cardiac risk factor such as high blood pressure, high LDL cholesterol and diabetes. It is important that they eat a heart healthy diet, get regular exercise, avoid excess weight and smoking.

What follows is practical information about Lp(a) and some potential ways to communicate with your family.

The Family Heart Foundation has created these resources that you can edit and use to communicate with your family member.

People often ask:

What should my family members do?

Each family member related to you by blood can be screened for FH by making an appointment with their doctor or with an FH specialist. They will need to get a simple cholesterol test often referred to as a lipid panel (including Total Cholesterol, HDL, LDL, and Triglycerides). They should also plan on bringing information about your FH diagnosis and other family history of heart disease and/or high cholesterol to their appointment. The doctor will use this information to make a diagnosis of FH or rule out FH.

How is FH diagnosed?

FH can be diagnosed based on results of a cholesterol test and a family history. This is called a "clinical diagnosis." Genetic testing may be helpful, but it is not required for diagnosis.

FH is suspected:

- in children with a low-density lipoprotein cholesterol (LDL-C) level > 160 mg/dL
- in an adult with an LDL-C > 190 mg/dL
- and with a family history of similar elevations of their LDL cholesterol and/or early heart disease.

We can also diagnose FH with a genetic test, called a "genetic diagnosis." FH is caused by a gene change in three known genes: the LDL receptor (LDLR) gene, the Apolipoprotein B (APOB) gene and the proprotein convertase subtulisin/kexin type 9 (PCSK9) gene.

It is not important for you to remember the exact genes or the specific gene change, called a mutation. It is important to understand that a person's specific genetic mutation in one of the FH genes might not yet have been identified. Since we are still finding new mutations in these genes, sometimes people with a clinical diagnosis of FH have a negative or inconclusive genetic diagnosis. In this case, the clinical diagnosis is still accurate.

The Family Heart Foundation's genetic testing toolkit is available if you are interested in learning more: www.FamilyHeart.org/genetic-testing-and-fh

If I have been diagnosed with FH, what should I tell my family about getting screened for FH?

As noted above, FH is caused by a dominant gene. If you have FH, approximately half of your first-degree relatives will too. Let your family know that the recommendations from many groups including the American Academy of Pediatrics, the American College of Cardiology, and the American Heart Association are:

- all children in families with FH or early heart disease should have a full "lipid profile"(Total Cholesterol, LDL-C, HDL cholesterol and Triglycerides) at the age of 2. Importantly, the initial test can be done with a finger stick and the child doesn't have to be fasting.
- all children, regardless of family history, should have a lipid profile between the ages of 9-11.
- in addition to cholesterol screening between 9-11, young adults should have a lipid profile again between the ages of 17-21.
- adults should have their cholesterol checked every 5 years, and more frequently if they are on a cholesterol lowering medication.

It is important for you to know that a recent study has shown that, in spite of these recommendations, only about 4% of children ages 9-11 are having their cholesterol checked. **You and your family members need to be informed and speak up.** You might need to ask for this lab test.

As noted above, FH can also be diagnosed through a genetic test. If you have been diagnosed genetically, providing your relatives with your exact gene mutation in your genetic test results can make their genetic test easier (and cheaper) because they only need to be tested for the gene mutation that you have inherited.

How is FH treated?

The great news is that FH is very treatable and multiple safe and effective therapies are available today. In addition to a healthy lifestyle, it is important for most individuals with FH to take cholesterol-lowering medications. Medical guidelines recommend getting LDL-C levels below 100 mg/dL for people who do not yet have cardiovascular disease, and below 70 mg/dL for people who already have cardiovascular disease. Adults should begin treatment as soon as they learn they have FH and work with their healthcare team to achieve and maintain their LDL-C goals.

Children with FH who have high LDL-C can begin statin therapy between the ages of 9-11. While this can be a difficult decision for parents, research shows the benefits of early treatment. Recent data from the Netherlands, where there is a very comprehensive program for diagnosis and treatment of individuals with FH, compared outcomes in a group of children who began treatment with statins in childhood with their parents for whom statins were not available when they were young. By young adulthood only 1% of those beginning statins in childhood vs 26% of their parents had experienced a cardiac event such as a heart attack or cardiac surgery. Perhaps even more important by age 39 no one who began statins in childhood had died from cardiovascular disease but 7% of their parents had.

The bottom line is that, for most people, FH is very treatable and starting early improves outcomes. You can help prevent heart disease in your family by sharing your FH diagnosis and helping your family members get screened for FH. Early diagnosis and treatment of FH saves lives.

Where can I go for support?

The Family Heart Foundation has developed tools and resources to help individuals and family members impacted by FH, including diagnosis and treatment guides, as well as community support tools. The Family Heart Foundation can also refer you to a specialist that is an expert in diagnosing and managing FH. You can find more information at www.FamilyHeart.org, through email at info@thefhfoundation.org or by calling 626-583-4674.