



used to say FH affected 1 in 500 individuals. But we now know through very large modern genetic studies and other studies, that FH is much more common than we once thought, ~~about~~ in 220. It affects all races and ethnic groups. It's present in all countries that have been surveyed so far. That means that there's at least 1,300,000 individuals in the United States with FH. Unfortunately, many individuals with FH do not know they have it. They do not know that they're walking around with a risk of early onset heart disease. Maybe as few as 10% actually have been diagnosed properly. There are many reasons for that, but the biggest reason is that people are not looking for it.

- Cat Davis Ahmed: 03:36 I'm so glad we have this opportunity to talk about FH, and hopefully there may be people who are listening who can recognize themselves and their families in this broadcast, quite frankly, and then look into it. If it sounds familiar, ~~how~~ would somebody go about finding out if they have FH? How is FH diagnosed?
- Dr. Knowles: 03:59 That's really the critical question, and unlike many conditions for which individuals might have symptoms, high cholesterol doesn't necessary cause any symptoms ~~til~~ a bad event like a heart attack occurs. But we should really be looking for FH because it can be diagnosed relatively straight forwardly. The first thing is looking at the cholesterol levels. In an adult if the LDL cholesterol is over 190, or if a child, if the LDL cholesterol's over 160, we should have a high degree of suspicion for FH. If there's a family history of early onset heart disease, if your mother or your father, or your sister or your brother had a heart attack before age 60 for men, or ~~50~~ women, they should really strongly consider FH as being involved. Really, a few years ago the American Heart Association endorsed those kind of criteria as when we should really be suspicious for FH.
- Cat Davis Ahmed: 05:04 How about genetic testing? I know there's a lot of information in the news about genetic testing. FH is a genetic disorder. What's the role of genetic testing for FH?
- Dr. Knowles: 05:15 That is also something that's really emerging as being extremely important. In many countries ~~around~~ the world, FH genetic testing has been standard of care for many years, and it's been shown to be highly efficacious in increasing the ability to find individuals with FH, making sure they are motivated to take the right therapies, and then especially ~~by~~ screening other potentially affected family members. In the US, until recently and even currently, FH genetic testing is not as commonly done as it maybe should be. There's many reasons for that. It has been relatively expensive and people haven't known ~~who~~ order

the tests. But just a few weeks ago the FH Foundation helped

the arteries, especially those that feed the heart. A little understanding of how that process works might be useful here. Atherosclerosis is a condition that affects all of us. It progresses in the same way, but at different rates. It can be viewed as a response to injury. Things like diabetes, or high blood pressure, or smoking can all damage the blood vessel wall and trigger this atherosclerotic response. But one of the most potent stimuli for atherosclerosis is high LDL cholesterol. LDL cholesterol can get inside the blood vessel wall, and trigger an inflammatory response. That can lead to plaques in the arteries that get larger and larger over time, and sometimes can rupture and cause heart attacks.

One of the key messages for individuals with FH is, they've been born with a predisposition to extremely high LDL levels. Those LDL levels can trigger that atherosclerotic response early in life. They can progress more rapidly than in other situations. FH individuals are often affected 20 or 30 years prior to their other individuals that don't have FH, and can be very severely affected in their 40s and 50s with heart disease.

Cat Davis Ahmed: 10:19

That's certainly something that I worry about, and inspires me

medical records, and look at their cholesterol and look at how severely they're affected. What we learned from that is that many individuals with FH are affected by heart disease at a very early age, that are not diagnosed until very late, often after they've had a heart attack or stroke. We know from other data, that the good news about FH is that you identify it early enough and treat it aggressively enough, that you can prevent almost all the downstream consequences. One of the major goals of the FH Foundation has is to try to prevent every individual with FH from becoming a patient with FH. The way we do that is by lowering the LDL cholesterol, maybe even more aggressively than we would do for other people. In most individuals, treatment with a single drug like a statin might be enough, but in a substantial proportion of FH patients, more than one cholesterol lowering medication is needed. In our registry, about 40% or 50% of FH individuals required a second medication to lower cholesterol. About 20 required three medications. It's har(u)5.3 Td [(c)-10.8 (o42.83 (c)8)5.3 (s)1.5.3 (a)2.5.3 (

be found online. It's surprising that even many experienced healthcare providers and cardiologists are not as up to date on FH care as they should be, find a lipid specialist, or a cardiologist, or an endocrinologist or a healthcare provider that is really willing to go the extra mile.

Of course the other thing that you need to do is take care of yourself. That means, if you're into a very healthy diet and lifestyle, exercising every day, 45 minutes or an hour, not smoking, making sure you control your blood pressure and your weight and your other risk factors. And then most importantly, control your cholesterol. One of the key messages for FH is that diet and exercise, while important are never enough by themselves. It's only with the use of medications that we can lower cholesterol effectively in these individuals with a genetic predisposition. Taking your medications regularly, getting your LDL cholesterol down, usually down certainly to a LDL less than 100 mg/dL. Many of us treat it to a LDL less than 70, is really important.

Then the final thing that I always say is that again, we never find an individual with FH. We only find family members with FH. A diagnosis of FH in the family should prompt you to get connected to your other relatives. All first degree relatives need to be screened, mother, father, brothers, sisters, children all need to be tested for FH, and that can usually done with a lipid panel or genetic testing is available. That can also help.

That's sort of a long answer, but I think an important answer.

Cat Davis Ahmed: 17:06

Perfect. I think it's a great answer. When I get the chance to talk to people with FH, especially when they're newly diagnosed and worrying about themselves and their children, I often say, you can only do what you know to do. A diagnosis actually can really give you the tools that you need to address this risk factor that most people with FH, we know that heart disease runs in the family. Once we've got the FH diagnosis, we know what to do about it, and to try to prevent heart disease in ourselves and in our children and our grandchildren, and generations to come. I think it's a really hopeful message, as hard as it can be to have FH. Along those lines, what would you say Josh [crosstalk 00:17:55] Yeah, sorry.

Dr. Knowles: 17:56

No. I was just going to say, on that note, I think that word hopeful is a really important one. I remember cutting a video. We had a little illustrated video about being hopeful when you have FH. I think that's really a key message, that even if one individual in a family is affected profoundly by FH, maybe

