

FH JOURNEYS FROM AROUND THE GLOBE

Reflections from individuals, families and clinicians affected by familial hypercholesterolemia (FH)












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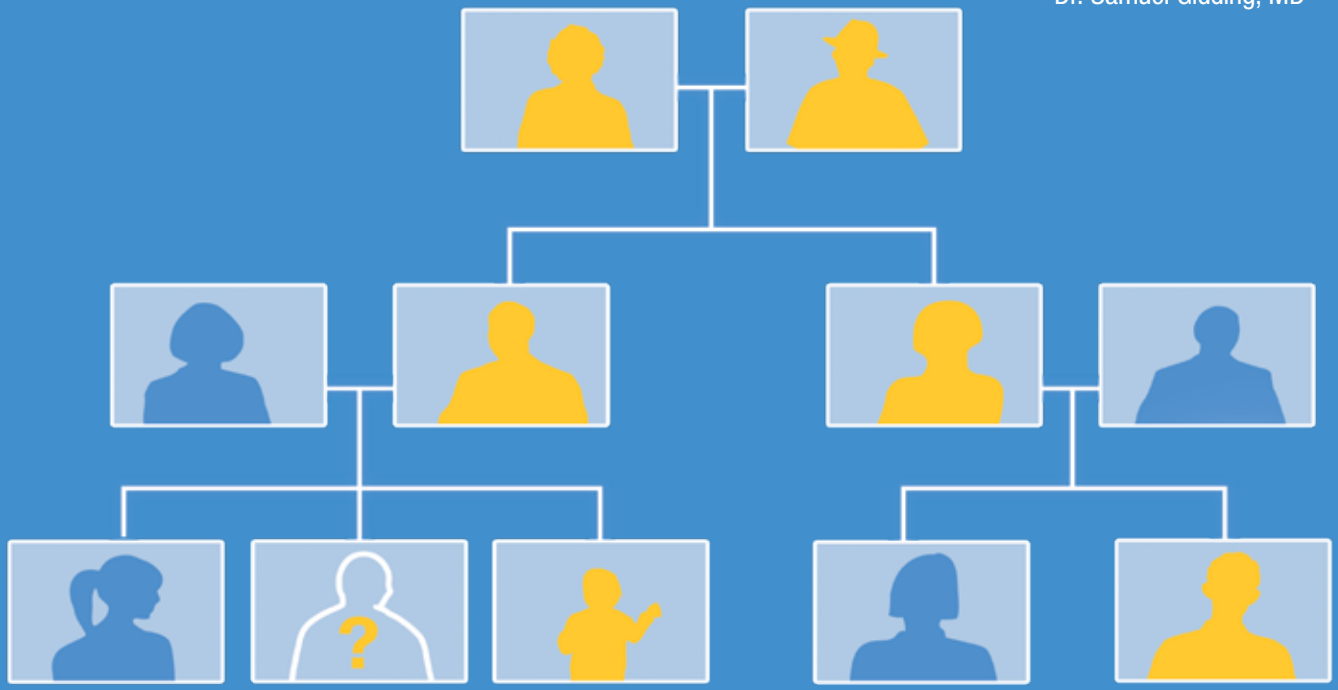
Familial hypercholesterolemia (FH) is different because the patients have serious health problems but often times no symptoms. The family has to believe in the value of prevention, understand the hereditary nature of the problem, and understand the disease. For parents of children with FH, it is often hard to accept the need for lifelong treatment to prevent heart disease in someone who seems otherwise healthy.

Dr. Samuel Gidding, MD



FH is an autosomal dominant disease, meaning all 1st degree relatives have a 50% chance of also being affected by FH. This family pedigree depicts how FH might affect a typical family. This is only a depiction and does not represent an actual family. For an actual family pedigree and story, turn to page 11 and read Joyce's story and see her family pedigree.

FOREWORD

A familial hypercholesterolemia (FH) diagnosis is a lifelong journey for patients and the people that love them. It is often a path of uncertainty. Despite the fact that FH is a relatively common genetic disorder, it affects approximately 1 in 500 people; there isn't a lot of awareness around FH. It is estimated that only about 20% of FH patients are currently diagnosed. Of the nearly 80% that are undiagnosed, many will have a cardiac event or stroke before the FH diagnosis is made. Because of this lack of awareness, newly diagnosed patients often feel that they are alone on their FH journey. I'm here to tell you that you are not.

In this book, we want to introduce you to patients around the world who have faced fear and are learning to cope with the everyday challenges of dealing with FH. Living with FH isn't easy, but as these intimate profiles will reveal, you can do it. You will also meet some groundbreaking healthcare providers and specialists who often hold our hearts and hands and guide us to a healthier life.

It is my hope that after reading these very personal and poignant stories, you will feel connected in this global community. There are people out there who are in the same boat as you. It may not be an easy voyage but let's set sail together. In the words of Louisa May Alcott, "I'm not afraid of storms, for I'm learning how to sail my ship."

Wenter Blair

FAMILIAL HYPERCHOLESTEROLEMIA

Familial hypercholesterolemia (FH) is a common genetic disorder that can lead to heart disease early in life. FH is characterized by significantly elevated cholesterol levels and a family history of premature cardiovascular disease (defined as occurring in men before age 55 and women before age 65, but may occur in adolescence or early adulthood). People who have FH have inherited specific mutations that limit their body's ability to clear cholesterol, increasing the amount of cholesterol in the bloodstream.

FH affects 1 out of every 500 people and is found in all populations and ethnic groups. However, in some groups, FH can be 2 to 5 times more common than in the general population.

(Examples of these groups include French Canadians, Christian Lebanese, South African Afrikaners and Ashkenazi Jews.)[1]

FH is a dominant genetic disorder, which means all of the first-degree relatives (children, siblings, parents) of those affected have a 1 in 2 chance of having FH as well. Other genetic disorders like Cystic Fibrosis are recessive, so you can be a carrier of the disorder without having it. FH is different: if you inherit the gene, you will have it. A healthcare provider can diagnose FH using cholesterol tests and family history.

Early detection of FH and screening of all first-degree relatives is essential to prevent or delay the early onset of atherosclerosis and heart disease. If FH is found early, serious problems of the heart and blood vessels may be prevented or delayed by making changes such as not smoking, exercising regularly, eating a healthy diet and taking your medications.

It is important to find FH and take action at any age, because when treated, the risk of heart disease can be reduced to levels similar to those of the general population.[2]

1. Austin MA, Hutter CM, Zimmern RL, Humphries SE. Genetic causes of monogenic heterozygous familial hypercholesterolemia: a HuGE prevalence review. Am J Epidemiol. 2004;160(5):407-420.

2. Goldberg A, et al. J of Clinical Lipidology. 2011.04.003.

Patients

SEAN AND ANGELINA

Our two-year-old daughter Kennedi led us to this FH community. She started developing blisters on her feet. At first, we thought they were from the shoes she was wearing. She stopped wearing the shoes but the blisters didn't go away. In fact, they spread. Her pediatrician couldn't figure the blisters out and referred us to a dermatologist. The dermatologist thought they looked like xanthomas, which are lipid deposits on the skin but told us that was pretty rare for a child her age. The biopsy revealed that they were in fact xanthomas and lab work was ordered.

We got the call from the pediatrician; Kennedi was very sick. Her cholesterol was through the roof. Her original numbers came back somewhere around 840. We were referred to a pediatric cardiologist who hadn't come across too many patients that young with such high cholesterol levels. He referred us to Dr. Gidding who is a pediatric cardiologist and lipid specialist at DuPont Children's Hospital.

We met with Dr. Gidding for almost four hours. He went over everything. We conducted a complete family history and additional testing. Kennedi was diagnosed with homozygous familial hypercholesterolemia (HoFH), which means as parents we were probably both carriers of the FH gene and each passed on a FH gene to Kennedi. Because Kennedi received a gene from both of us, she had inherited a rare and severe form of FH. Essentially, her arteries have been bathed in excessively high levels of cholesterol since birth. Further testing confirmed that we both had a more common form of FH known as heterozygous familial hypercholesterolemia (HeFH) as did our 10-year old son. It was clear that our family had to immediately change the way we lived our day-to-day life.

We are on an emotional roller coaster. Some days we get good news and other days are uncertain. We don't attend support groups in person but we have an online community that is helpful to us. I go online and talk to other patients from around the world. Everyone throws out questions and discusses treatment options. It has been nice to network that way. We are trying to develop an even stronger FH support group and online community. As a matter of fact, we think the online communities are good sources for getting information and finding the right specialists. Sometimes patients need to stand up and advocate for themselves with their providers. Sometimes doctors aren't 100% right. These communities are great tools.

Right now one of our biggest challenges is trying to figure out what the best treatment options are for Kennedi. What's the right decision for her future? It is a lot to process. There are days where we are worried. Often at night, we have her sleep with us all night because we're afraid something is going to happen. Just talking about it gets me emotional, but you have to stay strong for your child. They are smart. They know when you're worried.

"It is such a bright spot that Kennedi's homozygous FH (HoFH) was diagnosed when she was so young. Because of her age, time is on our side to move forward and find the best treatments."

Patient

KATHY

My story started when I was 24 and pregnant with my second child. Through a routine blood test, I learned that I had "high cholesterol" and was told we would worry about that after I gave birth. Over the next 30 years, I tried every new medication, exercised and ate well but my cholesterol never came down. My physicians didn't seem particularly concerned. I was 46 years old when I was told I needed a quadruple bypass. Even that didn't change the way physicians treated me. My lipid levels were still not at safe levels and cardiologists told me there was nothing more they could do for me. During this time, I experienced several episodes of severe chest pains and was repeatedly rushed to the Emergency Room. All the while, no one ever explained to me what was going on. No one took the time to explain to me what was happening to my body.

It wasn't until I was 54 that I found my way to a lipid specialist who gave me a diagnosis of familial hypercholesterolemia (FH) and explained to me what it meant. Because FH is a genetic disorder, I passed it on to my son. And because we were never educated about the possible ways FH can impact a family, my son had a heart attack at the age of 32. Perhaps looking back at my family history, there were signs along the way.

My journey to a FH diagnosis took 30 years, but I am happy to share that I'm finally doing better because of the additional therapies offered to me by the lipid specialist. Because the lipidologist clearly explained how FH was impacting my life, I finally understood what is really wrong and now I have a solution.

"I wish I knew I had familial hypercholesterolemia (FH) a lot earlier. I would have been much more proactive and I would have kept better tabs on my heart and my base lines. I would have been more active in my care. I would have been a better advocate for my children's care."

Caregiver

JOYCE

My name is Joyce and I am a nurse practitioner in a cardiologist's office. I'm also a person who has been touched by familial hypercholesterolemia (FH). Many years ago, almost 50 as a matter of fact, I happened to fall in love with a man who has FH.

The story begins with my husband Jerry and his parents. The day after my husband graduated from high school, his father died of a heart attack. He was an active marine. We really didn't know much about cholesterol in those days. When my husband was a senior in college, his mother died from a heart attack. And so, within three and a half years, both of his parents were gone. When my husband was 43, he was diagnosed with FH and had heart disease.

We have five children. We had to initiate having their cholesterol levels tested. We were never encouraged by their healthcare providers to do any cholesterol screening despite knowing our family history. Two of my five children have FH. Now the grandchildren need to be tested and I want them tested young. Even if their parents don't have FH, their grandfather does. Every generation is at risk and it can skip generations.

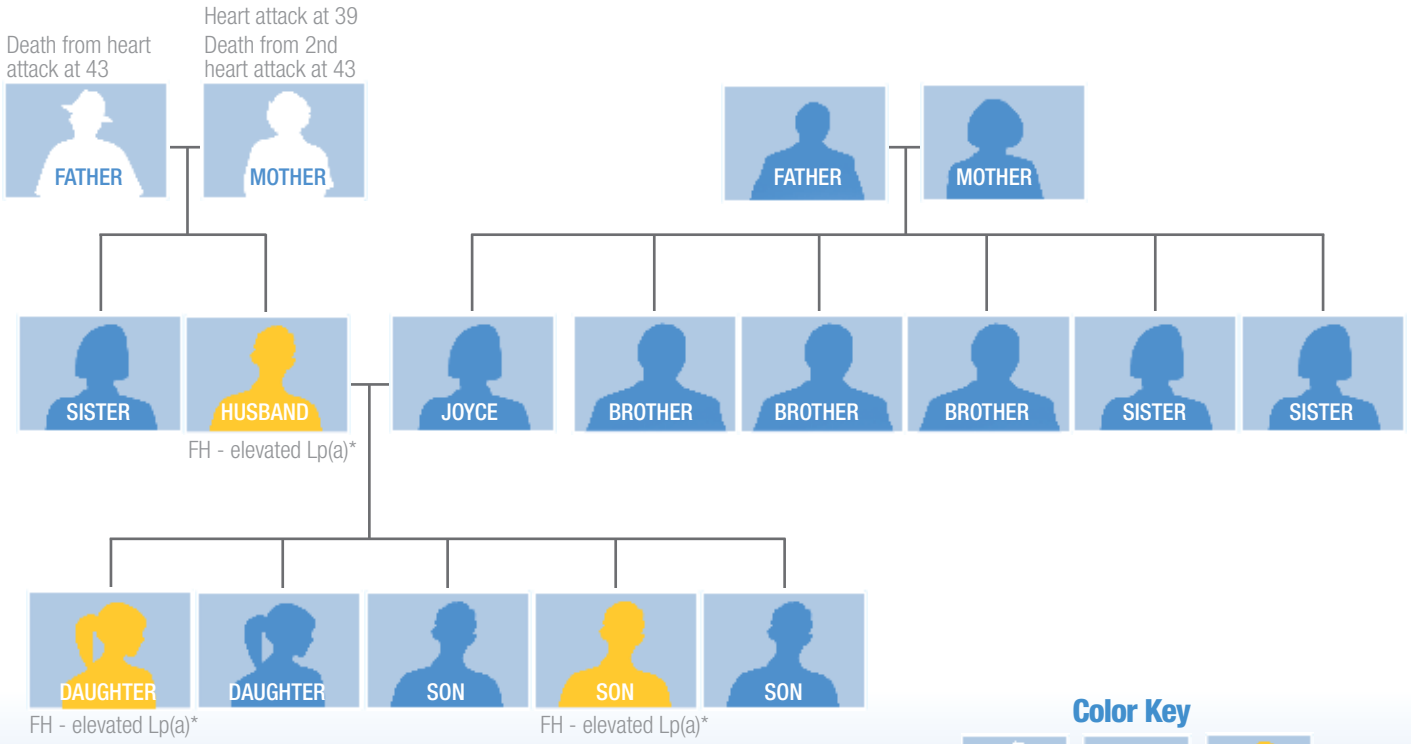
With FH, you can't feel it, touch it, or taste it. If untreated the damage is being done. Get tested. Get your family tested. If you identify a person with familial hypercholesterolemia early and start treating early, you can prevent the deposit of cholesterol in their arteries and hopefully prevent cardiac diseases. Not knowing that you have high cholesterol is not going to help you. This is one genetic problem that, if diagnosed, you can actually manage and hopefully have a normal life.

Although I am a nurse, I'm still a mom. I'm still a wife. Sometimes, I tend to be overly aggressive about wanting them to take care of themselves. Sometimes my husband will laugh at me and say, "Yes, Doctor." But I get frustrated when they are not doing the right things for their health. When my kids were growing up, I was adamant they practiced good lifestyle management. We are a very active family and maintain a balanced diet.

Personal experience, along with my professional experience, is a beautiful blend. I believe that we're planted where we're supposed to grow in this world, and I really think this is where I was supposed to be planted.

"My husband Jerry had heart disease at 43. Both of his parents passed away at 43, but Jerry is still alive—he's 71! He's very much here and very much with us. The reason I believe he's here is because we know how to manage familial hypercholesterolemia (FH) better today. We have treatments and we understand the disease better."

Joyce's Family Pedigree



Color Key

- Represents where there is reason to suspect FH but was never diagnosed
- FH not present
- Confirmed FH diagnosis

*Lp(a) is a type of LDL particle that, when elevated, is recognized to increase risk of events like heart attacks. It is genetically-determined and more common in FH than in the general population

Patient

JANNIE

In my family, familial hypercholesterolemia (FH) is just a way of life. I was diagnosed when I was 11. I was too young to consciously experience any emotional or physical problems. Even before I was officially diagnosed, I knew I had FH. For me, it was normal.

My father had his first cardiac event when he was 28 years old. He had a few more cardiac events and died when he was just 36. Three of his brothers developed heart problems at an early age as well. It was my aunt, a nurse, who suspected the presence of FH in our family. As it turns out, they all had FH. My sister has FH. Many of my cousins have FH. Brothers, sisters, cousins, we've all been tested. It is part of our family's routine care.

Because cholesterol levels are difficult to manage in FH patients, my internist was having a hard time managing mine and referred me to a lipidologist. I'm happy to say I have a good relationship with my healthcare providers. I can tell them anything and they really listen to me. They discuss options for treatment with me and take into account which ones work best for me. They do a good job keeping me informed with respect to the latest developments and medication. I frequently participate in the trials of new medication. Overall, I feel good about my care.

I do wish that healthcare providers would appreciate the fact that it is hard for FH patients to observe the dietary and lifestyle rules. Sometimes I think providers advise dietary and lifestyle changes that are difficult to maintain, which actually has an adverse effect on patients. When you are told you have FH, there are so many changes in your life. Even though high cholesterol and FH have been part of my family's day-to-day life and seemed almost routine to me, it hasn't been an easy road. Since my diagnosis, I've had balloon angioplasty twice, a brain hemorrhage and I've developed heart problems.

I find comfort through my volunteer work with a FH support group and have found that discussing the everyday challenges of the FH journey is helpful.

"Familial hypercholesterolemia (FH) was simply part of us as a family, a fact of life we rarely talked about it."

Caregiver

ANNALIESE

When I first heard about familial hypercholesterolemia (FH) I never, not even in my wildest dreams, thought it would one day become "the theme of my life." I fell in love with a young man who told me he had a heart attack at the age of 33. You don't have heart attacks at the age of 33, I thought, at least not if you are slim, very athletic, health-minded, with a positive attitude. He told me that his family was suffering from some kind of condition called FH. He had been diagnosed at the age of 19, after his mother had a heart attack and bypass surgery at the age of 40. He had been on and off medication and had stopped taking the medication altogether at 33 because nobody had told him he had to continue taking it. He thought he was fit enough to do without it. He thought when he worked out even more at the gym he was safe. And then, one day, he had terrible pain in his chest and went to the hospital. They sent him home with some painkillers for "muscle inflammation." The pain grew unbearable and after 3 days, he was taken to the hospital again where he was finally diagnosed with a heart attack. No one, not even physicians, wanted to believe that at 33-year-old man who looked so healthy and fit could have a heart attack.

I have been married to that young man for 14 years. Because he recovered from his heart attack and was taking his medication, I did not think too much about FH again until in 2000 when our daughter was born. Relatives of my husband who also had FH misinformed me that only mothers, not fathers, pass FH on to their children. I thought my daughter was safe. How wrong! When my daughter was 2 years old, I told our pediatrician about my husband's FH. Thankfully, he was aware of the condition and recommended a blood test for my daughter. My daughter was actually suffering from a very severe form of FH.

In my opinion, this was certainly bad news and it came as a great shock to my husband and me. However, the good news was we were sent to one of the best FH experts in Austria. My daughter's doctor immediately started treatment with medications. Ever since that time, her cholesterol levels, especially the LDL levels, have been almost normal so there is a good chance that she will never develop cardiovascular disease. My daughter is now 11 and lives a totally normal life filled with lots of sports and music and entertainment. She is fit, has a healthy diet and is a good student at secondary school. The other good news, since my husband's heart attack 15 years ago, he's had no other cardiac events.

I feel so strongly that people need to be educated about FH. Knowing about it means you can do something about it. With proper lifestyle changes, diet and medication, hopefully you can prevent early cardiovascular diseases or even prevent early deaths. Getting wise to FH can save lives!

Physician

DR. SANTOS

Diagnosis is a gift! The best advice I can give to a newly diagnosed patient is to bring your whole family in to be tested. I tell patients that the disease is genetic and may affect one in every two family members. It's very important to identify them since they might be at risk of cardiovascular disease in the near future. Most patients can live a normal and healthy life as long as they are diagnosed early and treated.

As a lipidologist, I develop a very close relationship with my familial hypercholesterolemia (FH) patients since we treat many members of the same family. I treat people from different generations and the relationship I've developed with them over time is stronger. Together we share many of life's happy and sad moments.

For example, I was treating a 36-year-old man with coronary disease and very high LDL levels. We screened his family and found that one adult sister and his 13-year-old son also have FH. We started treatment for them. As his son got older, he informed me that he no longer wanted to follow a restrictive diet. I advised him he needed to continue his medications. He assured us he would and would follow the treatment recommendations at an outpatient clinic. At 18 years old, he started to smoke. Three years later the father called me, I assumed to update me on his own health status, but unfortunately he was calling to tell me that his son had sadly experienced a myocardial infarction and passed away at the age of 21. This broke my heart. I didn't know what to say. The father and I cried together on the phone. That was the saddest story of my clinical life. That boy was not supposed to die.

On a happier note, I am treating a 21-year-old woman with homozygous FH (HoFH). She presented regression of a carotid plaque and her coronary computed tomography was totally normal. With medical advances, these patients are living longer. It is my hope that we can do more for them in the future. I'm convinced she'll get a chance for a long and happy life.

"Screening is the most important thing about familial hypercholesterolemia (FH). By having your family screened, you're not preventing the disease in one person but in many people at the same time."

Patient

GERALDINE

I was seven years old when my father had his first major heart attack when he was only 37. At an early age, I had heard of high cholesterol but never familial hypercholesterolemia (FH). That is, until recently.

For me, my FH journey started three years ago while living in South Africa. I was diagnosed with high blood pressure after a couple bouts of bronchitis. At that time my cholesterol was also tested and it was slightly elevated, but the doctor wasn't overly concerned because I was in my early 30s and I was a woman. He was only focused on treating the hypertension.

Because of the hypertension, I have my blood tested annually. Earlier this year, my cholesterol levels came back high again. So, my general practitioner in England referred me to a professor at Oxford's endocrinology and diabetes unit. It was the professor there who conducted further testing and made the diagnosis. As I mentioned, it was the first time I had ever heard of FH.

Once I had time to process the FH diagnosis there was a sense of relief that it was genetic and not caused by something I had done. I've always tried to maintain a healthy lifestyle. Because FH is genetic, there are implications for my family. They may also have FH. My brother is going to be screened, but at this point, my sister is too scared to be tested.

I recently spoke to a physician who referred to an early diagnosis as a blessing. This physician said, "Your diagnosis allows you to make changes and decisions that hopefully will prevent you from ever having a heart attack or some other type of cardiovascular event." I think that this is a lovely way to look at it. I didn't have to reach the edge of the abyss before I realized something was wrong. I thank my lucky stars that I have a wonderful GP and specialists caring for me.

My friends have been supportive. Some have lost family members because of high cholesterol. Although they may not understand the full impact of FH, they are aware of the possible problems that could arise from it. They watch me like a hawk when I eat now and remind me when I've had one too many pieces of cheese!

Because this is a new diagnosis for me, I'm still learning so much about what it all means. But if by sharing my story, I can help one person understand a little more about FH and spare them a heart attack, my work on this planet is done.

"When my cholesterol levels came back high, I should have been screened further, but I wasn't because I am a young woman. I'm just as much at risk of a heart attack as a man."

Professor WATTS

From a physician's point of view, I think familial hypercholesterolemia (FH) is a very interesting and complex disorder that is inadequately understood, and hence under-appreciated, under-diagnosed and under-treated. There is an important mandate that we try and reverse all these shortcomings. FH is ostensibly a simple disorder to treat, but, in fact, when you get into it, it's a complex disorder whose best management requires multiple skills when dealing with affected individuals and families. Nursing staff, genetic counselors, dietitians and psychologists all play a key role in the care plans for people with FH. Many FH patients have other cardiovascular risk factors, such as obesity, hypertension and metabolic syndrome that need treatment in own their right. Screening for subclinical atherosclerosis can play a pivotal role in risk assessment and treatment. Those with symptomatic coronary artery disease need review by a cardiologist and some proceed to bypass surgery. Side effects of medication, e.g. statin myopathy, need monitoring and management, and patients with refractory FH will need LDL-apheresis. So, you see, what initially appears as a simple cholesterol problem has many layers of complexity. So it is critical, at a teaching and training level, to educate medical undergraduates, residents, advanced trainees, postgraduates, primary care physicians, and specialists about FH.

Universally, there is a gap in FH care. By that I mean a gap in detection, a gap in treatment, and a gap in reaching targets. Every audit that has been done in most countries confirms this major gap in prevention. The question is: why is there a gap? Well, there is a gap because, FH falls into preventive medicine and there are limited resources at present being devoted to this important branch of medicine. If you haven't had a cardiac event, you fall into the gap. With FH it's not just about a general population strategy, it's about very high-risk prevention. We've got to identify and treat these high-risk individuals to prevent an event. Identifying index cases in high-risk situations (e.g. those in acute coronary care or coronary rehabilitation units with premature ACs or MI), followed by family tracing or cascade screening for FH, has to be embedded into routine clinical practice. Adequate resources are needed to do this.

Additionally, I don't think the population has been sufficiently educated about hypercholesterolemia. This is important because FH is a silent condition until you develop symptomatic coronary artery disease, or your first heart attack. It's not like severe hypertension where you can get double vision, headaches and feel unwell, or diabetes where you can lose weight, feel thirsty or develop an infection. So it's a latent condition. Again, that's related to the nature of the early phase of the condition. People should insist on having their blood cholesterol checked routinely, especially if there is a family history of elevated cholesterol and early heart disease. They should inform their siblings and those affected should have their children checked, usually after age of 10 years. Empowerment of the community with this duty of care for itself is paramount. More education of primary care physicians and cardiologists about FH is essential. This is very important because there is still a very large gap in awareness and understanding about FH.

My own personal view in the gap in FH care is that a change is required in mindset about coronary prevention. We need to focus on chronic care models that involve preventive cardiology. Health systems haven't been geared towards coronary prevention. That is something we are going to have to change very soon, but it needs to be adequately resourced.

Patient

ERIK

In some respects, I feel very lucky. I learned about my familial hypercholesterolemia (FH) by accident. Thankfully, it wasn't after a heart attack. When I was in college, I tried to take part in a pharmaceutical study. To qualify, there was a complete physical and they discovered my cholesterol was very high and suggested I see a specialist. At 20, I was diagnosed with FH. I had never heard of FH, but I was painfully aware of heart disease - heart attacks ran in the family. My mother died of a heart attack when I was 10 years old. She was 42. That's how old I am now. I think about that everyday.

I'm also lucky that my career path took me to the healthcare industry. I started doing research on cholesterol-lowering medicines and speaking with cardiologists as part of the work I was doing. Through that process, I learned more about myself and worried that my lipids weren't being managed aggressively enough. Until this point, I never really thought my condition was serious because my primary care doctors didn't treat it as though it was an issue. I had to fight with my primary care doctor to do more. I felt I needed more tests, new therapies, medications. I had to start fighting for my life.

By the time my children were born, I was a seasoned advocate for our care. It was critical for my wife and I to have both boys tested to see if they had inherited FH from me. Initially the pediatrician thought we were just overly protective parents who spent too much time on the Internet. We continued to press for testing and by my son's second birthday he saw a cardiologist and was diagnosed with FH. Because of his diagnosis, it was easier to have my younger son tested. He too has FH.

My diagnosis allowed me to be more aggressive and, therefore, gave me some sense of control, but I have to admit, I worry. I think about it every night before I go to bed. I don't like knowing that I'm at significantly high risk for heart attacks and I'm not going to be able to avoid it. It feels like a train is barreling down the tracks and I'm trying to outrun it, but eventually the train will be faster than me. I wish I had been diagnosed earlier, but I'm working hard to stay healthy for my family. I'm happy that my sons were diagnosed at a young age and their chances of a long and healthy life are better because of it.

"Before I was diagnosed, I guess there was always the hope that the heart attacks in my family were just a fluke."

Caregiver

MARTHA

My journey started when I met my husband, Erik (Erik's story featured on page 18). When he was 20, he discovered that he had familial hypercholesterolemia (FH). Erik takes his FH very seriously and is very good about managing it. When I was pregnant with our first child, we discussed that there would be a 50/50 chance the baby would be born with FH. Life takes on a whole new meaning; the responsibilities of taking care of a child, raising a child not to be frightened and teaching them to be in charge of their body.

When the pediatrician called to give us my oldest son's test results, I completely crumbled after I hung up the phone. His body is working against him. As a parent, the diagnosis is devastating. However, I'm now thankful… thankful that I know. What would have happened to him if I didn't know? I now know that both of my sons and my husband have FH. This is a journey we're all on together.

My boys are active and play soccer and other sports just like their friends. Here are my perfect, healthy looking sons, whose bodies have a secret. You can't help but look at other kids and just think, "You are so lucky. You never have to worry about this, but my kids do." They will have to carry this around their whole lives. They will always have to be mindful starting at their early age.

The most important message I would like to share with other FH families is to develop a good relationship with food. After the diagnosis, I cleaned out the cupboards of certain foods. We try to eat healthier. It's not perfect and it's not everyday, a cupcake on special occasions is okay. We try to be kind to our bodies. We take this seriously. If you teach your children how to eat properly, it may not solve the problem but it will keep it from getting much worse. Let's face it, we're guiding them in every aspect of their lives and if we guide them now, then that becomes their foundation.

"It's very hard to look your kids in the face and say you have this problem, and do it in a way that you are kind of smiling at the same time, saying, but isn't it great that we know?"

Patient

WENTER

I've known since college that I have high LDL cholesterol, but it wasn't until I was 40 and had a heart attack that I was diagnosed with familial hypercholesterolemia (FH) by a geneticist. For twenty plus years I had "high cholesterol" but not one physician uttered the words FH until it was almost too late.

I had just spent the afternoon setting up for a children's fall festival that I coordinate every year. I had helped unload two hundred hay bales and thousands of pounds of pumpkins but this year I felt different. I was incredibly tired. Later that night, while visiting with neighbors, I started sweating like never before. A trusted friend, who is a physician, asked me to get checked by a cardiologist. The EKG showed I'd had a heart attack the night before but the cardiologist wanted to chalk it up to a "false positive". Again my friend stepped in and demanded I ask for a nuclear stress echo, which we did. In the middle of the exam, the doctor leapt out of his chair, put his stethoscope over my neck and chest and asked, "Are you okay?" He said, "On paper you are having a heart attack but you're standing here smiling at me!" Again, doctors wrote it off as a "false positive". "See you in a year," he said. Then a miracle happened. A nurse who had been watching the exam pulled me aside and said, "Wenter, if you were my sister or my mother, I would put you in car and drive you to the hospital right now. If you don't have a heart catheterization, you will be dead in a week." She was right. I had four arteries blocked at 90%. Thankfully the nurse in the cardiologist's office spoke up. She saved my life.

Because FH is a genetic disorder, my whole family was screened shortly after my FH diagnosis and as a result, my son was also diagnosed with familial hypercholesterolemia (FH). A FH diagnosis early in life is a blessing. Physicians believe that because my son's FH is being managed early, he may not have the same serious health complications I am facing. As I learned more about FH, I realized that it was the contributing factor to my heart disease. I wish my FH had been diagnosed when I was 19, when I was first told about my "high LDL cholesterol". My FH diagnosis has helped me understand the seriousness of my elevated LDL cholesterol and has motivated me to advocate for far more aggressive management of my disease.

Two years and six stents later, I am still working with my physicians to effectively manage my FH. Just six months ago, while my son was having friends sleep over, I was rushed by ambulance to the hospital because I had another heart attack. Shortly after that frightening experience, we sat our son down to fully explain the medical implications of his lipid disorder. Having that talk was difficult because we weren't aware of a family-friendly book, a website or even a support group to turn to. In fact, I feared there was certainly no one else like us.

Today, we fight this battle as a family and we try to maintain a routine, joy filled life. I recently ran a 10K with my husband. I ride horses. I garden. I'm a hockey mom. I'm blessed! I have a compassionate husband and beautiful children with whom I stand stalwart. FH is not an individual diagnosis; it's a family's diagnosis. I am also learning about the larger FH community that is coming together. I am discovering and overjoyed with the fact that my family is not alone in this journey. There are many other families like mine around the world and together we will learn, love and live life to the fullest!

"The kicker with this diagnosis? There is someone in your immediate circle who has it. A parent, a child, an aunt, someone you love, someone else in your family that gets to fight this battle with you."

Physician

DR. KHERA

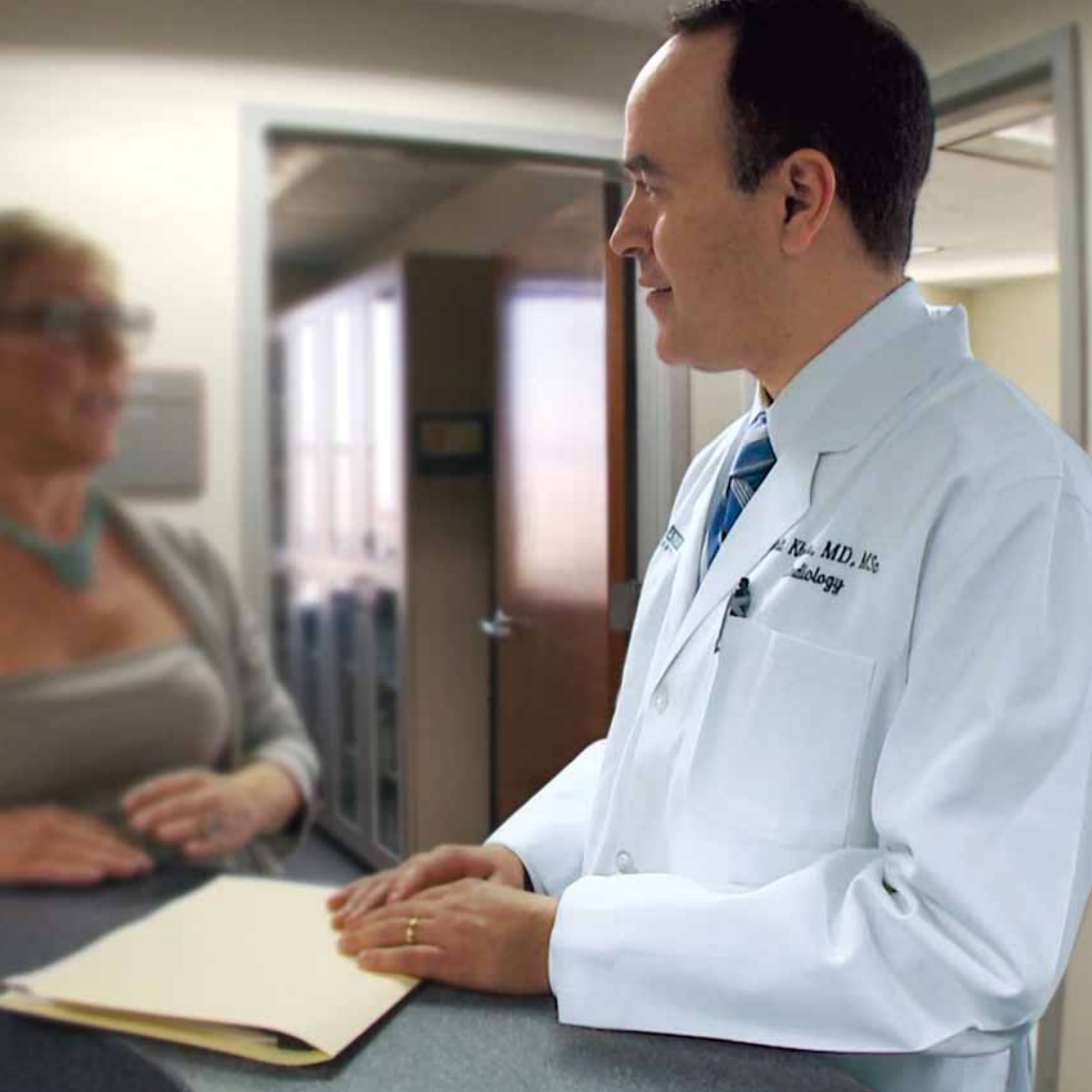
Wenter is my patient. She is also my partner in managing her care. But what impresses me most about her is that Wenter is her own advocate.

Wenter not only has a genetic disorder, but also coronary disease. Because of that, she has more problems that come up along the way. I have to be honest, I probably think about her at least once a week if not more. I am always thinking about what I can do to manage her care differently.

Unlike other patients with high cholesterol, people with familial hypercholesterolemia (FH) have different challenges and we must help them cope. These are often times people who are in the prime of their working lives and have young children. For them, this is a lifelong disease. That's where the therapeutic bond comes in. Wenter and I have to work very closely together initiating various therapies. We make sure we are constantly thinking outside the box. What's interesting and what's really fun about working with her, is that as much as I am on the phone, email or talking to people around the country who are finding new therapies, so is she. Many times she'll get the leads for me and say, "Here I talked to so and so, what do you think about this?" I'll work backwards through her connections to help her with her therapy. It's not the usual physician-patient relationship. We're both partners in trying to get her the best therapy. My job is to then look at this in the broad context of her care, scientifically, to say: does this make sense for her? Then, I can be there to help her sort through the information she's receiving.

Our relationship is based on trust. She has to trust me. I have to trust her. Ultimately, my feeling is always this; I want her to have the best care possible. That doesn't necessarily have to be from me if there is something else that is available. If there is another specialist with a better solution, I'm happy to facilitate that connection. It is about getting her the very best care I can.

"When familial hypercholesterolemia (FH) is diagnosed, you have such an opportunity to impact the whole family. It becomes a tragedy and a travesty when FH is left undiagnosed in family members when it so easily could be."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RESOURCES FOR FH FAMILIES AND CAREGIVERS

Learnyourlipids.com - Patient information from the Foundation of the National Lipid Association

Heart UK (HeartUK.org.uk) - Heart UK is the UK Cholesterol Charity and is committed to raising awareness about the risks of high cholesterol.

FHJourneys.com - A comprehensive FH disease website designed to support patients, families, and healthcare providers affected by FH on their journey to understand, manage, and educate others about this surprisingly common, yet under-recognized, genetic disorder.

The FH Foundation: US (thefhfoundation.com) - The mission of the FH Foundation is to raise awareness of FH (familial hypercholesterolemia) through education, advocacy, and research. The goal is to save lives through increasing the rate of early diagnosis and encouraging proactive treatment of this life-threatening disease.

The International FH Foundation (Fh-foundation.org) - The International FH Foundation works to raise awareness of FH in the public domain, medical community, and government.

YouTube.com/FHJourneys - This website features educational scientific animations and videos of people and families with FH describing their journey with this genetic disorder.

NOTES

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The FH Foundation
raising awareness. saving lives.

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The Preventive Cardiovascular Nurses Association