FDC BEAT

The Newsletter of the Familial Dilated Cardiomyopathy Project at Oregon Health Sciences University
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Staying in Touch with our Growing FDC Family

Since the beginning of the FDC Research Project in 1993, our research team at Oregon Health Sciences University (OHSU) has had the opportunity to work with an increasing number of families affected by FDC. When we activated our National Institutes of Health (NIH) grant in 1998, the number of families we were able to work with grew... and grew... and grew... from three to sixteen! And it's still growing!

As our project has grown, we've wanted to stay in touch and share information with you about this exciting work! So we got our heads together and decided to launch FDC BEAT, a newsletter that will come out three times a year and keep you updated on happenings and developments with the FDC Research Project at OHSU.

We're excited about this new addition to our research program and the opportunity to stay connected with you. As always, our goal is to provide you with information and support as we work together towards our goal of finding the cause -- and the cure -- of FDC.

The PI's Files: *notes from our principal investigator*

Ray Hershberger, M.D., is the head of the FDC research team. He'll be writing a frequent column about the project, our progress, and other news you can use!

I was trained, in the late 1980s, as a heart failure/heart transplant cardiologist -- an expert for patients with advanced heart failure. I have treated scores of patients with idiopathic dilated cardiomyopathy (IDC), or heart muscle disease of unknown cause. A common question from patients with IDC and their families has been, "Does dilated cardiomyopathy run in families?" The conventional answer in the 1980s and early 1990s was, "Familial disease has been reported in a few cases, perhaps in 1-2% of families. You really don't have anything to worry about!" That was the best that we knew then.

Now we know better! A provocative research report was published in 1992, which evaluated first-degree relatives of patients diagnosed with IDC by echocardiography. Amazing to me then, 20% of patients with IDC were found to have family members with a similar diagnosis! This meant that IDC was really a familial disease (familial dilated cardiomyopathy, or FDC) at least 1 in 5 times!

I was struck by this report -- even though I was a well-trained heart failure/heart transplant cardiologist, I was routinely missing the FDC diagnosis in the patients (and their families) with whom I worked! I needed to change my practice patterns.

In 1992 I decided that we must learn a great deal more about FDC, and slowly over the past few years we have evolved this FDC research program into the active and productive group you have come to know. How common is FDC now according to the latest research? In 1998, we learned that IDC is likely familial in 35%-50% of cases, meaning up to one in two cases of IDC is inherited! Some speculate even higher numbers.

Please stay tuned to this exciting story. We will be updating you with our progress three times a year. And THANK YOU to each and every participant. We sincerely appreciate all of your help to make this research effort possible and productive. -- Ray Hershberger, M.D.

Terms you can use: Dilated Cardiomyopathy

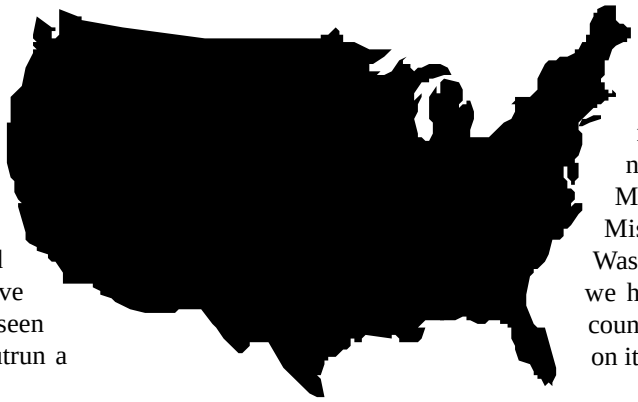
Dilated cardiomyopathy is a type of heart muscle disease in which the heart enlarges, or **dilates**. When the heart dilates, the wall of the heart's main pumping chambers (the **ventricles**) get thinner and weaker. This makes it harder for the heart to do its work.

Moving? New Phone Number? Questions? Comments? Suggestions? Stories? Call us toll-free at 1-877-800-3430, or visit our WEB site at http://www.fdc.to If you've got a great FDC story, we'd love to share it in April's newsletter!

Road Trip!

From Oregon to Maryland, Michigan to Mississippi, the FDC team crisscrosses the country to offer screening to as many of our FDC families as possible

There's almost no place our intrepid research team won't go for a screening trip. So far, we've held 20 different screening trips all over the United States. We've screened people at family reunions, held screenings at local clinics, in a motorhome, in a hotel suite, and visited folks in their homes. We've also had some excellent BBQ, seen lovely parts of the country, outrun a



fast-moving winter storm, spent time in lots of airports, and had the chance to meet many of you, all in the name of research. So far we've made it to Arizona, California, Illinois, Indiana, Iowa, Maryland, Michigan, Minnesota, Mississippi, Missouri, Nebraska, Oklahoma, and Washington State for screening trips. If we haven't gotten to your area of the country yet, don't worry. We're working on it, and we'll see you soon!

Dr. Kathy Tells All!

In this FDC BEAT exclusive, Dr. Kathy Crispell spills the beans on why we draw blood as part of our screenings

There are several parts to our screening process, including a history and physical examination, an electrocardiogram, an echocardiogram and obtaining a blood sample. Obtaining a blood sample is most everyone's least favorite part of the screening process, but it is one of the most important components of screening.

Blood is made up of liquid (serum) and solids (cells). There are two major types of cells in the blood: the red blood cells and the white blood cells. Most of the blood we collect is used to obtain DNA, a person's genetic material, from their white blood cells. Every individuals' DNA is unique. However, since everyone gets half their DNA from each parent, portions of DNA may be shared by members of the same family. Once we have completed screening of an entire family, a group of FDC scientists analyze the DNA patterns of the family and determine which ones are shared. We hope to find a piece of DNA shared among family members who we believe may carry the gene for FDC. If such a pattern is identified, then we will be able to identify what chromosome and what place on the chromosome the disease-related gene is near. This will help us find the gene that causes FDC.

> "...your blood provides very important information ..."

We draw four tubes of blood for our sample, about two tablespoons of blood in all. The first tube we draw has a purple top, and we use the blood from this tube to extract and analyze the DNA patterns in a family.

Two tubes have green tops and we use the blood in those to get DNA for storage. White blood cells are sorted out from the red blood cells. The white blood cells are then frozen. If in the future we need more DNA to run other important tests, the white blood cells can be thawed and grown, thus providing a living source of DNA.

Lastly, the blood in the speckle-topped tube is used to check for an enzyme that is released at high levels from damaged muscle. There are some dilated cardiomyopathies that are associated with increased levels of this enzyme, which is called CPK.

Hopefully, this has helped you see that your blood provides very important information to help us unravel the cause of FDC. While it's not a lot of fun, having your blood drawn is essential to our work — and it helps your family. Thanks!

Don't forget to plug into the new FDC website, where you'll find articles, information, and links to FDC topics and resources. You can send us e-mail through the web site, too! Our address on the world wide web is **http://www.fdc.to** Happy surfing!

Swimmin' in the Gene Pool with Emily

Emily Hanson, M.S., is the genetic counselor on our staff. Emily has her master's degree in genetic counseling and is here to answer your questions about genetics. From what makes DNA twist to exactly what a gene is, Emily's your one-stop gene info shop.

We're always asking FDC study participants for a sample of DNA, but what exactly IS **DNA?** DNA is found in the nucleus, or center, of every cell in your body. It is a long molecule in the shape of a double helix — picture two ladders side-by-side, twisted together in a spiral. The DNA in a cell is packed down extremely tightly to allow a lot of DNA to fit in a very small place. In fact, if the DNA from one cell was taken out and stretched end to end, it would be 6 feet long!

A DNA molecule takes the form of a **double helix**, shown here

There are sections of DNA called genes. Genes are a recipe for creating a living thing (in your case, a human being!) — they direct your growth and development from the time you are just a single cell. Altogether, there are 50 - 100,000 genes in one molecule of human DNA, or **genome**. There are genes that guide the development of your brain, your muscles, and your bones. There are also genes that guide the development of your heart.

It is these heart genes that we think may be altered in people with FDC. With your help, we will be able to identify these genes and find out how they affect the size and function of the heart.

Who discovered DNA's shape?

The discovery of the structure (and shape) of DNA is credited to James Watson and Francis Crick. Watson and Crick used published information from other scientists (particularly Rosaline Franklin, an X-ray crystallographer who took pictures of DNA) as well as their own research in their discovery.

Watson and Crick published their hypothesis in a 1953 issue of the scientific journal *Nature*. As you can imagine, this was big news in the scientific community, and Watson and Crick won the 1962 Nobel Prize for Physiology and Medicine as a result of their discovery.

Help us stay connected!

Staying in touch with our FDC families is important to us! Although we try to call each of you once a year, we can't always keep up with everyone who's moved or changed phone numbers. Help us out by cutting out the card on the right, filling it out and mailing it back to us. That way, you can stay up-to-date on the latest in our research — and we can stay in touch with you!

If you aren't involved in the FDC project yet, but would like more information, send in the card with your address or telephone number and we'll get right back to you.

And don't forget: our WEB SITE is a great way to stay connected, no matter where you are. Visit us at http://www.fdc.to (This is the third time we've mentioned it in this issue. Can you tell we're excited about it, too?)

My family is already involved with the FDC Project and I need to update my contact information.

My name is: _____

My mailing address is: _____

_____

_____

My telephone number is: ( ) _____

My e-mail address is: _____

I'd like to learn more about the FDC Project. Please send information to:

_____

_____

_____

_____

Or call me at: ( ) _____

Meet the Team! (or two of us, anyway)

The FDC Research Project Team is made up of an interdisciplinary group of clinicians, biomedical researchers, and statisticians. Some of you have already met some of us, but we'd like all of you to meet all of us! In this issue, we're profiling Emily and Kendra. Many of you know them as voices on the phone What a relief to find out they actually exist as real people, too!

When you pick up the telephone (or send an e-mail, letter, or FAX) to the FDC Project, chances are good that either Emily or Kendra will be the first people on the FDC research team that you talk to, and they'll be the ones you talk to most frequently throughout your participation in the project. Although Emily and Kendra have different roles in the project, they have one job in common: working with FDC families.

Emily Hanson, M.S., is our staff genetic counselor. Emily has her master's degree in genetic counseling from the Medical College of Virginia. This means that she is trained to talk with people who are dealing with genetic issues, and help them obtain and understand the information they need in order to make good decisions for themselves and their families. Because FDC is inherited (genetic), those of you with questions about how it may be passed on in your family should talk to Emily; she can talk to you about this risk.

Kendra Wise, B.S., is the FDC project coordinator. Working with so many families (and all the new families we're constantly learning about) means that we have a lot of information that needs to be kept organized and easy-to-find. That's Kendra's job. She distributes our information to the people who need it, when they need it.

So Emily is a genetic counselor, and Kendra is the project coordinator. But what work do they have in common? They both work with our FDC families, explaining the FDC project, building family trees (we call them "pedigrees"), obtaining documentation like death certificates and medical records, and contacting family members about the FDC project. They both plan (and go on!) screening trips, making sure that everything goes as smoothly as possible. And they're both here to answer your questions and help you get the information you need. We want to find the gene that causes FDC, and want to help our FDC families while we're doing it. That idea is the basis of everything that Emily and Kendra do.

How do you get in touch with these two? By calling our toll-free number, of course! It's **1-877-800-3430**. Kendra is at extension #1, and Emily is at extension #2.

P.S. Emily is our webmistress, and Kendra is our newsletter editor. If you've got suggestions about either, give them a call and let them know! We'd love to hear from you. Or, (you guessed it!) send us an e-mail using our web page (one more time: **http://www.fdc.to**)

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