FDC BEAT

The Newsletter of the Familial Dilated Cardiomyopathy Project at Oregon Health Sciences University
Portland. Oregon, USA **Volume I, Issue 2** **June, 2000**

What To Do After You've Been Screened

Emily Hanson, M.S. frequently answers questions that arise after a person has been screened. In this article, she goes over the basic guidelines and recommendations for screening follow-up.

As part of our research, we have performed clinical screenings (echocardiograms, electrocardiograms (ECGs) and physical examinations) on hundreds of people who have or have family members with familial dilated cardiomyopathy (FDC). Deciding how often FDC family members should be "rescreened" is difficult and varies between individuals; however, here we make some general recommendations. If we screened you and our recommendations to you were unclear, or if you are unsure as to whether you should receive follow-up, please contact us. Dr. Crispell recently received a small grant to rescreen a fewof our families. We will be starting with the earliest screened families, probably sometime in the early fall.

My Screening Results Were Normal...Now What?

Many people we screen do not have any evidence of cardiac problems on echocardiogram, ECG or physical examination. While this is good news, it unfortunately does not rule out the possibility that some problems could develop in the future. Because it is beneficial to detect FDC in it's earliest stages, people who have relatives with FDC should be aware of their and their children's health and be alert for signs of cardiac disease. Children, siblings and parents of individuals with FDC should have a repeat echocardiogram and ECG every 3-5 years. We are unsure if more distant relatives need to undergo screening on a regular basis. However, anyone who experiences any cardiovascular symptoms such as palpitations, shortness of breath, edema (fluid accumulation) or chest pain should seek medical attention, inform their doctor of their family history of FDC and undergo rescreening.

My Screening Results Weren't Normal...Now What?

If we detected a cardiac abnormality on your echocardiogram or ECG at the time you were screened, we probably made specific recommendations for your follow-up. Recommendations differ depending on the abnormalities found and your medical history, age and position in the family tree. Individuals who were diagnosed with dilated cardiomyopathy should see a cardiologist regularly and discuss medical therapy. Those who were told that they may have early signs of FDC - for example, an arrhythmia or a slightly enlarged heart - should establish care with a cardiologist. Some medications (e.g. ACE inhibitors or beta-blockers) may be helpful in preventing or slowing the progression of FDC, even for those who feel fine and do not have any symptoms.

We Want to Hear From You!

Our FDC Families are at the heart of all we do. Therefore, it is extremely important to us that we receive feedback from you. What have your interactions with the FDC Group members been like? How has FDC affected your family? What kind of a screening experience did you have? We are especially looking for things to put in our next newsletter. Submissions may be published anonymously at your request. If you have contributions of any kind (i.e. questions, stories, comments or suggestions), please:

1. call us toll-free at 1-877-800-3430
2. visit our website at *http://www.fdc.to* and send an email from the "Contact Us" page
3. email us at *messages@fdc.to*

4. write us at:

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FDC Research Update:

Lamin Mutations Cause FDC in Some Families

In December, researchers from Harvard Medical School reported an important discovery. It was already known that mutations, or gene changes, in a gene called **lamin** could cause a specific form of muscular dystrophy (called Emery-Dreifuss muscular dystrophy). Because people with Emery-Dreifuss muscular dystrophy sometimes have heart problems similar to those seen in people with FDC, the researchers decided to evaluate 11 families with FDC for mutations in the lamin gene. Lamin gene mutations were in fact identified in 5 of these 11 families. Other family members with this gene change had heart failure, arrhythmias or other problems with the heart's electrical system (often requiring a pacemaker) or sudden death. However, some gene carriers had no or few signs of heart problems, indicating that not everyone who has a lamin gene mutation has significant heart disease. Knowing exactly how lamin mutations cause problems in the heart's muscle and electrical system may help us to formulate better screening tools and treatments for both people with FDC and those at risk for developing it.

Fatkin et al. 1999. *New England Journal of Medicine* 341:1715-24.

Part Two of the Screening Process: Echocardiograms and ECGs

In our January issue, Dr. Kathy Crispell explained the importance of blood draws as part of our screening process. Now she tells why Echos and ECGs are vital to our work.

As part of the screening process, an echocardiogram is performed. This is a very important screening test. This is a moving picture of your heart obtained by using sound waves. By obtaining images of your heart, its structure, size and function can be evaluated. The internal dimensions of the main pumping chamber of your heart (the left ventricle) can be obtained if the quality of the images is good. The pumping function of your heart can be evaluated. The direction and velocity of blood flow through your heart can also be assessed. It is very easy to do an echocardiogram but trained technicians are required. There is no pain involved. During the test, you may be able to actually see your own heart beating and hear the blood flowing through your heart. Children are often frightened during the echocardiogram or have a hard time lying still for the 10-15 minutes it takes to administer the test. They can, however, be distracted by watching cartoons or videos if there is a VCR or TV close by. The information obtained during the echocardiogram is stored on a video or a compact disc for review at any time.

"...Both of these tests are very important..."

An electrocardiogram (ECG) is also done as part of the screening. This is another painless, easy test to do. It records the electrical signals produced by your heart. Some types of abnormal electrical signals and heart rhythm abnormalities can be associated with dialted cardiomyopathy. They will be picked up if they are present at the time of your ECG. Persons of any age can get an ECG with little difficulty.

Both of these tests are very important. It is probably important to repeat these tests about every three to five years if you have a parent, sibling or child with dilated cardiomyopathy. This would be the way to monitor for changes in your heart size and function and to monitor for changes on your ECG. The results of these tests will be reported to you after they have been reviewed and interpreted by Dr. Crispell.

Log on to the FDC Project website at *http://www.fdc.to* for access to articles, information and links to other FDC topics and resources.

Funding the FDC Research Project

Ray Hershberger, M. D., the principal investigator of the FDC Project, explains the grant writing process, which is the first step towards getting the money necessary to keep our research going.

This is grant writing time, and one of the key parts of my job is to write the grants for our program. What is a "grant?" A grant is a formal request to a funding agency for money to support our research. For the FDC program, our support comes from the National Institutes of Health (the NIH), which is one of eight agencies of the Public Health Service, a component of the Department of Health and Human Services, a branch of the federal government. Yes, your federal income taxes are supporting our FDC research program! The NIH is divided into 25 institutes, which are organized according to themes such as cancer, aging, child health, etc. Our support is from the National Heart, Lung and Blood Institute (NHLBI). You may find more information about the NIH at *www.nih.gov*.

What are the components of a grant? A grant is a highly structured document that begins with what we call the "front pages:" a summary, a listing of key professional personnel, a line-item budget that requires justification of all items, curriculum vitae (training, awards, honors and publications) of key personnel, and the resources available, such as space, equipment, etc. The next part is the "main body" of the grant, where most of my time is invested when preparing the grant. It has four sections. The first is a brief description of the specific aims of the work proposed - our specific aims are to identify and characterize large families with FDC, and to find the disease genes which cause FDC. Next is a "Background and Significance" section, where the importance of FDC research is outlined for the health of the nation. Then preliminary data - an accounting of all the work we have accomplished. Finally, the "Experimental Plan" - a highly detailed plan for exactly how we propose to do everything that needs to be done!

The deadline is July 1, 2000. Reviews will be returned by late fall and if our scores are good enough, funding will be received to continue our work. It's competitive - only about 18-25% of grants are funded. Our current budget, May 1, 1998 - April 30, 2001, was $1.45 million. We need a 10% increase to keep up. Wish us luck! We'll put in the hard work necessary to get an extremely high quality grant out on time. And THANK YOU to all FDC Project participants - you make it worthwhile!

Keeping in Touch

Please keep us updated on any name, phone number, or address changes. If any of your or your family's contact information has changed, please fill out the card to the right and mail it back to us. If you would like to be taken off our mailing list, please fill out the card and check the box at the bottom. Thank you!

FDC Research Project
I need to update my contact information:

NAME: _____

ADDRESS: _____

_____

TELEPHONE: _____

EMAIL ADDRESS: _____

☐ **Please remove my name from the FDC Beat mailing list.**

FDC BEAT Newsletter

FDC BEAT is a triannual publication of the Familial Dilated Cardiomyopathy Project at Oregon Health Sciences University in Portland, OR. The newsletter is not copyrighted and readers are welcome to photocopy its contents to share with family members and professionals.

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FDC Bulletin Board

Notes, Announcements and Reminders

SMALL FAMILIES

We have begun collecting DNA samples from "small" families - those with only one or a few individuals with FDC. Please contact us if you would be interested in sending us a blood sample! (We will pay for your blood draw and shipping costs.)

REMINDER

To those of you on heart transplant lists who plan to donate some of your heart tissue to us, please put your tissue kits where you won't forget them when called for your transplant. Call us with any questions about this protocol!

REMINDER

If we have sent you blood tubes and you have not yet had your blood drawn, please do so as soon as possible. If you have questions or are having difficulty finding someone who will draw your blood, please call us! Thank you!

UPCOMING TRIPS

We'll be in Mississippi at the end of June and Tennessee in July or August- We look forward to seeing you this summer!

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Address Service Requested

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