FDC BEAT

Newsletter of the Familial Dilated Cardiomyopathy Project at Oregon Health & Science University
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Types of Cardiomyopathy:

Dilated (DCM), Hypertrophic (HCM), Arrhythmogenic Right Ventricular (ARVC) and Restrictive (RCM)

At the FDC Research Project, we frequently receive referrals of patients who have types of cardiomyopathy other than dilated cardiomyopathy. In this article, we hope to explore and clarify the ways that the various types of cardiomyopathy affect the heart muscle.

WHAT IS CARDIOMYOPATHY?

Cardiomyopathy is a diagnosis of heart muscle disease (*cardio* = heart, *myo* = muscle, *opathy* = disease). Four types of cardiomyopathy are classified by different morphological characteristics (size, shape and thickness of the heart muscle, as well as examinations of individual cells microscopically). Cardiomyopathy usually causes heart dysfunction, except in early stages of disease. Three types of dysfunction may occur. 1) Difficulty pumping blood (heart failure). The main function of the heart is to pump blood to the body. If the heart is unable to pump enough blood, heart failure is the term used. Heart failure commonly causes swelling (edema), shortness of breath with exertion and fatigue with activity. 2) Heart rhythm disturbances (arrhythmias). The heart normally beats with a regular rhythm. Cardiomyopathy may cause rhythm disturbances (too slow, too fast or irregular), which can cause syncope (sudden loss of consciousness), or worse, sudden cardiac death (SCD). 3) Blood clots forming in the heart and moving to other organs (stroke or embolus). In general, treatment options for the varying types of cardiomyopathy are frequently focused on the symptoms and the treatment of heart failure and the prevention of blood clots and SCD.

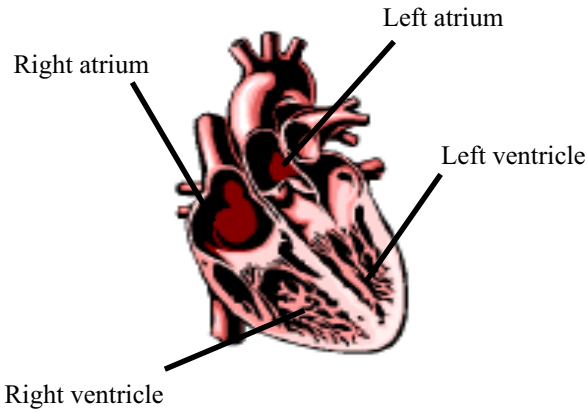

Figure 1. Heart anatomy.

DILATED CARDIOMYOPATHY (DCM)

Dilated cardiomyopathy is characterized by dilatation (enlargement) of the left or both ventricles with relatve thinning of the heart muscle and decreased systolic function (decreased strength of pumping). DCM frequently results in progressive heart failure and SCD from arrhythmias. People with DCM will usually present to their doctor with a range of symptoms, from being completely asymptomatic to having heart failure, stroke, arrhythmias or SCD. Approximately 35-50% cases of DCM are genetic (familial).

HYPERTROPHIC CARDIOMYOPATHY (HCM)

Hypertrophic cardiomyopathy is characterized by thickening of the muscle of the heart wall (ventricular hypertrophy). Clinical presentation of HCM also ranges widely from the asymptomatic to SCD. A diagnosis of HCM is often made on the basis of a heart murmur, abnormal ECG findings or an abnormal echocardiogram. HCM is a genetic disease.

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FDC BEAT Question & Answer

At the FDC Research Project, we frequently have been approached by people in our families who would like to see a Q & A section in the FDC BEAT. You requested it, we listened and here it is...

QUESTION: If I have IDC and there is no apparent history of family members with any other cardiac problems, should my family members be screened?

ANSWER: Yes! We recommend screening for any first degree relatives (brothers, sisters, children and parents) of patients with IDC. Many people can have early findings of this disease with no symptoms. Therefore, even though a family member may not appear to have any problems, many people can have early findings of this disease with no symptoms. If they are identified early in the disease process and started on drug therapy, their eventual quality of life and life expectancy may be improved. If the heart is found to be enlarged and/or its function abnormal after receiving an ECG and echocardiogram, then early disease may be identified and treatment considered. If these tests are normal, it is reassuring, but a normal test does not eliminate the possibility that disease could develop later. Therefore, we recommend repeat screening every 3-5 years in families with people who have IDC.

QUESTION: Does participating in the FDC Research Project affect my insurance?

ANSWER: No, it should not affect your insurance. Only the study personnel will have access to any of your medical records that identify you by name. This information will be kept in locked files or in password secured computer databases. All third parties, including insurance companies, will be refused access to the information, unless you provide written permission, or unless we are required by law to do so. An ID number will be assigned to you and your DNA. Only the people in the clinical research group will be able to link the code number to you. Any investigator, who may receive a sample of your DNA, will only be given a code number and will be unable to identify you or your relatives.

If you have any specific questions about FDC, disease progression, treatment, family implications, etc, please email, write or call and we will include your questions with answers in an upcoming issue. Chances are that if you have a question, there is someone else who is going through the same thing with the same kinds of questions.

Cardiomyopathy

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ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC)

Arrhythmogenic right ventricular cardiomyopathy (formally called arrhythmogenic right ventricular dysplasia (ARVD)) is characterized by progressive fibrofatty tissue replacement of the heart muscle in the right ventricle. Patients with ARVC frequently present with arrhythmias of right ventricular origin. ARVC progression can ultimately affect the left ventricle and cause subsequent heart failure. In contrast with DCM , which affects both ventricles, ARVC usually presents with disproportional right ventricular dilatation and dysfunction. ARVC presents in a genetic form in approximately 30% of patients.

RESTRICTIVE CARDIOMYOPATHY (RCM)

Restrictive cardiomyopathy is characterized when the ventricle walls thicken and become increasingly rigid, due to infiltration of abnormal tissue. As a result, the heart cannot fill adequately with blood and can lose the ability to pump properly. Patients with RCM frequently present with decreased cardiac output, symmetrical thickening of the ventricle walls, and/or elevated diastolic pressure in the heart. Patients with RCM may have complications with lethal arrhythmias and progressive heart failure. RCM is genetically inherited, however the extent to which is not currently known due to the rarity of the disease.

IDC versus FDC

Diagnoses of Idiopathic Dilated Cardiomyopathy (IDC) and Familial Dilated Cardiomyopathy (FDC)

IDC is the second most common type (20-25%) of DCM (ischemic DCM, or cardiomyopathy resulting from a previous myocardial infarction (heart attack) being the most common type). Idiopathic means 'cause is unknown.' Therefore, for a patient to receive a formal diagnosis of IDC, other causes of cardiomyopathy have been ruled out. Of individuals with IDC, about 30-50% seem to have family members with similar disease. IDC that is inherited, or runs in families, is FDC. A diagnosis of FDC can be made when IDC is identified in two or more members of the same family.

Hello and Welcome...

As you may know from our last issue of FDC BEAT, *Emily Hanson, MS moved to Madison, Wisconsin in September... Jessica Kushner, MS, a genetic counselor with extensive clinical genetic counseling experience, has replaced her. Please join us in saying hello and welcome from the FDC Project Group.*

Hello! In October, I had the privilege of joining the FDC Research Project as the in-house Research Associate/Genetic Counselor. Since that time, I have spoken to some of you in person, by phone or email. You have been so open and sharing, and I'm happy to take this opportunity to share with you some information about myself and what I hope I can bring to this project.

As Emily mentioned in her gracious introduction in the previous newsletter, I received my Masters degree in Medical Genetics/Genetic Counseling from the University of Wisconsin-Madison in 1997. As a genetic counselor, I have been trained to assess family histories, provide information about inheritance, occurrence and recurrence of a condition in a family, explain and interpret medical information about a condition and address the complexities and issues surrounding the option (or lack) of genetic testing. I also serve as an advocate for patients and their families and am aware of the emotional impact that a familial/genetic condition can have on family members.

Before coming to the FDC Project, I acquired a diverse background of clinical experience with many conditions in prenatal, pediatric and adult genetics clinics. I have also worked on a number of genetics education projects for both health professionals and the general public. I first worked in genetics research in my home town of Chicago, as clinical study coordinator of trials for patients with the genetic conditions of Fabry and Pompe disease. As I return to live in Portland, I am happy to now bring my skills and expertise to the study of idiopathic and familial dilated cardiomyopathy.

Working with the FDC project has already become an enjoyable and rewarding experience. I am impressed with the hard work and dedication of everyone involved, and the collective efforts that make up the FDC team - the FDC personnel and YOU, the individuals and families which make this research possible.

Much of my time is spent discussing FDC and the research project, enrolling families in the study and maintaining contact with those already involved. Through these and other activities, I hope to promote awareness of this condition, the current screening recommendations and the continued study of IDC/FDC, to both family members and those within the health profession. I also hope that my experience applying genetic knowledge and testing to a person's medical care will offer and important contribution to the underlying goals of this project - to better characterize and identify the genetic basis of FDC.

I look forward to getting to know many more of you as the FDC project continues. Please do not hesitate to call me at my direct line, (503) 494-3959 or through the toll-free number, (877) 800-3430, if you have any questions or concerns, or if you just want to introduce yourself!

FDC BEAT Newsletter

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

Newsletter Layout/Design: Kelly Coates, B.S.
email: *coatesk@ohsu.edu*

FDC Project Clinical Group:
Ray Hershberger, M.D.
Jessica Kushner, M.S.
Warren Toy, B.S.
Emily Hanson, M.S.
Kelly Coates, B.S.

FDC Bulletin Board

Notes, Announcements and Reminders

MEDICAL FOLLOW-UP

If your doctor performs additional heart testing for you or your family, especially if you have new or a change in any heart symptoms, we would like a copy of your results. Please contact us and we will send you a medical record release form.

UPDATES

If you have moved or have new family additions, please contact us so we can update your information in our databases.

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!

Remember....if you are currently followed by a cardiologist, he/she may see other patients who have family histories suggestive of FDC. Please pass our phone number and website along to your cardiologist so we can include more families in our research.

STAY TUNED

Please watch for updated information on our website. As always, please feel free to drop us an email through the "Contact Us" page if you have suggestions or ideas of material you would like to see covered on the web!

The FDC Research Project
Division of Cardiology, UHN-62
Oregon Health & Science University
3181 SW Sam Jackson Park Road
Portland, OR 97201

Address Service Requested

TO: