FDC BEAT

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

Portland, Oregon, USA Volume V, Issue 1 March 2004

Causes of Dilated Cardiomyopathy

Greetings from the Familial Dilated Cardiomyopathy (FDC) project group! We hope 2004 brings you and your families good health and happiness.

In many families we speak to, each family member has been told of a different cause for their cardiomyopathy. For example, one family member may be diagnosed with viral cardiomyopathy, while another is said to have cardiomyopathy due to excessive use of alcohol. In this newsletter issue, we will be reviewing what is known about the causes of dilated cardiomyopathy, and why differing explanations for the cause of cardiomyopathy are sometimes given to family members with FDC. Please see the glossary on page 2 for definitions of some important terms discussed in this article.

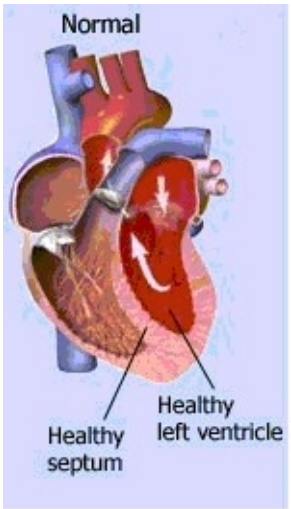
What is dilated cardiomyopathy (DCM)?

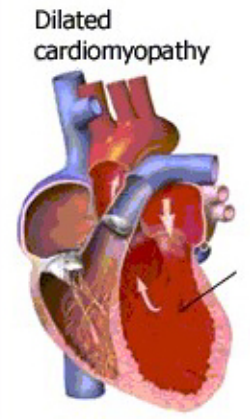
DCM is a disease of the heart muscle. In DCM, the heart muscle weakens, the main pumping chamber (left ventricle) is impaired, and the heart enlarges. DCM frequently results in progressive **heart failure** (HF), where the heart is unable to pump enough blood to the body. HF commonly causes swelling, shortness of breath with exertion, and fatigue with activity. DCM can also be associated with heart rhythm disturbances (arrhythmias) or sudden cardiac death, and blood clots forming in the heart, which can move to the brain and cause stroke. People with DCM usually present to their doctor with a range of symptoms, from being completely asymtomatic to having HF, stroke, arrhythmias, or sudden cardiac death.

What are the causes of dilated cardiomyopathy?

The medical and scientific literature includes a long list of causes for DCM. For some on this list, health professionals and scientists agree on a direct link between the specific cause and DCM development. **Coronary artery disease (CAD)**, which damages the heart muscle by cutting off its oxygen supply (a heart attack), is by far the most common condition that plays a direct role in causing DCM. A number of much less common causes are also known. For example, DCM is known to occur in people with significant **thyroid problems**. DCM can also be caused by an overload of iron in the heart (**hemochromatosis**), or replacement of heart muscle tissue with abnormal growths or substances (**sarcoidosis and amyloidosis**). Additionally, DCM can be caused by over-exposure to the anticancer drug **adriamycin**. DCM may also develop if someone has severe **heart valve abnormalities**. **Genetics** also plays a main role in causing cardiomyopathy, as mutations in certain genes can cause DCM. Genetics can also play a role in the other causative factors mentioned above; for example - CAD, hemochromatosis, and the others commonly run in families.

For other causes, however, questions still remain about the true nature of their association with DCM; some of the more common and perhaps controversial are viruses (**viral cardiomyopathy** or **myocarditis**), alcohol (**alcoholic cardiomyopathy**), high blood pressure





Please see **Cardiomyopathy**, Page 2

FDC BEAT

Causes of DCM

Continued from Page 1

(**hypertensive cardiomyopathy**), and late pregnancy (**peripartum cardiomyopathy**). These diagnoses are usually made when all other causes have been ruled out. When no specific cause can be determined, DCM is called **idiopathic dilated cardiomyopathy (IDC).**

What are the differences between IDC and viral, alcoholic, hypertensive, or peripartum cardiomyopathy?

These questionable causes of DCM are commonly confused with IDC. One reason is that definitive tests to diagnose these said causes do not exist, as it does for CAD in the form of an angiogram or catheterization. So viral, alcoholic, hypertensive, and peripartum cardiomyopathy are often diagnosed based on the medical history, without definitive testing, when all other causes of cardiomyopathy have been ruled out. IDC is diagnosed in the same way. Thus, while two people may have very similar symptoms or history, one may be diagnosed with IDC and the other with, say, viral cardiomyopathy.

Another very important difference between IDC and the questionable causes of DCM are the role that genetics is thought to play. Approximately 35-50% of patients with IDC are thought to have a genetic abnormality that has caused their IDC, which may

Glossary of Terms	
Dilated cardiomyopathy (DCM)	A disease in which the heart muscle weakens, the main pumping chamber is impaired, and the heart enlarges.
Heart failure	Various signs and symptoms including fatigue, shortness of breath, swelling in the legs and feet, difficulty laying flat in bed caused by insufficient blood supply to the body due to a heart problem.
Idiopathic dilated cardiomyopathy (IDC)	The enlargement and dysfunction of the heart where the cause is unknown. Other causes of heart enlargement/dysfunction must be ruled out.
Familial dilated cardiomyopathy (FDC)	FDC can be diagnosed if two or more closely related family members are diagnosed with IDC.
Genetic mutation	An alteration in a gene that disrupts the normal function of that gene. There are mutations in certain genes that are known to cause DCM.
Thyroid problems, coronary artery disease, hemochromatosis, sarcoidosis, amyloidosis, adriamycin, heart valve abnormalities, genetic mutations	Medical conditions or problems known to cause DCM. There are tests available (with the exception of genetic mutations, available in only certain limited situations currently) that can define these problems to link them with DCM development. Genetic mutations are thought to account for 35-50% of IDC.
Viral, alcoholic, hypertensive, and peripartum cardiomyopathy	Cardiomyopathy diagnoses that are commonly made based on medical history rather than definitive testing, raising the question of IDC as a more appropriate diagnosis. Genetic factors may play a significant role in these types of DCM, so IDC/FDC should still be considered when these diagnoses are made.
First-degree relative	Family members who are directly related, such as parents, brothers, sisters, and children.

also cause IDC in other family members. In contrast, genetic factors are usually not thought of as significant to the development of a patient's DCM when it is diagnosed as viral, alcoholic, hypertensive, or peripartum cardiomyopathy. For this reason, when someone is diagnosed with DCM due to one of these said causes, a family history may not even be taken and no thought may be given to recommending family screening.

So if a patient is diagnosed with viral, alcoholic, hypertensive, or peripartum cardiomyopathy, but the diagnosis could just as well be IDC, family members may not realize that they too might be at risk for DCM.

Why are multiple causes of DCM sometimes diagnosed in families with familial dilated cardiomyopathy?

A diagnosis of FDC can be made if there are at least two first-degree relatives (parents, brothers, sisters, children) with a diagnosis of IDC. Given the difficulty in differentiating between IDC and the said causes of DCM, multiple family members may be each diagnosed with one of the questionable causes, when the more appropriate diagnosis may actually be IDC and thus FDC.

Additionally, the relationship between the genetics of FDC and other said causes of DCM is not well understood. It may be that an underlying genetic alteration makes someone more susceptible to DCM when other risk factors are present. Exposure to certain types of viruses, alcohol or drug use, high blood pressure, or pregnancy, may trigger cardiomyopathy to develop in a person that carries a genetic mutation. So even if a test attempts to confirm one of these causes of DCM, this does not rule out a genetic cause and the possibility of FDC. In addition, because we are still learning about these relationships, and the genetics of heart disease is such a new field, many health care professionals, including cardiologists, may not realize the significance genetic factors may have in causing DCM.

What should I do if a family member or I have been diagnosed with DCM?

If you or a family member has been diagnosed with DCM from any suspected cause, you may want to consider the following:

- Ask your health care provider how the diagnosis of DCM was made, what the cause is thought to be, and if IDC is a possible diagnosis.
- Report any family history of heart problems, especially cardiomyopathy of any cause, (including IDC or FDC) to your health care provider. This may prompt them to consider IDC/FDC as a more appropriate diagnosis.
- For people without a definite cause of DCM (such as coronary artery disease), clinical screening of relatives should be considered, especially if more than one family member is diagnosed with any certain type of DCM, such as viral cardiomyopathy.

It is currently recommended that first-degree relatives have an echocardiogram and electrocardiogram (ECG or EKG) to look for early signs of cardiomyopathy. They may benefit from early treatment. This screening should be performed every few years if baseline tests are normal. Any relatives with symptoms such as shortness of breath, swelling in the legs and/or feet, difficulty sleeping flat in bed, palpitations, and/or dizziness/fainting should be evaluated right away.

We hope this information has been helpful to you. If you have any further questions about the causes of DCM, please contact us on our toll-free # (877-800-3430) or email us from our website at: www.fdc.to.

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 Smith, B.S. CHES
email: *smikelly@ohsu.edu*
Newsletter Author: Jessica Kushner, MS, CGC
email: *kushnerj@ohsu.edu*

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 97239

Address Service Requested

TO: