FDC BEAT

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Newsletter of the Familial Dilated Cardiomyopathy Project at the University of Miami
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New Medical Guidelines Published for Genetic Cardiomyopathy: What Do They Mean for You?

Greetings from the FDC Research Study! *Since the inception of our research project in 1993, we have recommended clinical cardiovascular screening for first degree relatives of individuals with dilated cardiomyopathy (DCM). As genetic research identified specific DCM genes in families, clinical genetic testing, which can help to predict who might be at increased risk for DCM in a certain family, was added to our recommendations. These recommendations have finally been collected in an official guideline for medical providers, titled, "Genetic Evaluation of Cardiomyopathy: A Heart Failure Society of America Guideline." In this FDC Beat issue, we provide a summary of the DCM portion of the guidelines, while providing you with information that you can use. FDC Research Project participants are encouraged to discuss these guidelines with their doctors, family members, and friends.*

The guidelines are particularly helpful if you:

1. Have DCM of unknown cause , also called idiopathic dilated cardiomyopathy (IDC), with or without a family history of DCM, and would like to have genetic counseling and testing.
2. Are (or know of) a first degree relative (parent, child, sibling) of someone with IDC, regardless of desire for genetic testing or any genetic testing results.

Over the last decade, much progress has been made in discovering the genetic cause of IDC. Studies in familial dilated cardiomyopathy (FDC), where one or more family members are affected with IDC, have shown a genetic cause in approximately 25-30% of cases. Our research has also identified mutations in individuals who report no family history of IDC (apparently sporadic IDC).

Based on these data, the Guidelines for the Genetic Evaluation of Cardiomyopathy, which were published by the Heart Failure Society of America (HFSA) in March 2009, are meant to bring uniformity to the medical management of all individuals with IDC. Authored by expert physicians in the field, with Dr. Hershberger, Principal Investigator of the FDC Research Project as lead author, the document recommends that physicians do the following:

- Obtain a family history
- Offer clinical cardiovascular screening to first degree relatives of individuals with IDC
- Provide genetic counseling
- Consider genetic testing

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Requesting Your Support for Our Work

Our research would not be possible without the continued gifts of time and participation that each person enrolled in this study has given us. Your greatest contribution, without question, is your participation in our research project. Now, for the first time since the research project's inception in 1993, we ask you to consider financial support of our research.

Our research effort is funded primarily through the National Heart, Lung, and Blood Institute (NHLBI) of the National Institutes of Health (NIH) in Bethesda, Maryland. However, NIH grant support can be intermittent and unpredictable. Grants are usually awarded for 3-5 years, but the submission, review, and approval process predictably takes from 10 to 24 months. This can lead to interruptions in funding between grant cycles, which creates an atmosphere of uncertainty that can disrupt a research program.

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Obtain a Family History

A family history (also known as a pedigree or family tree) should be obtained from all individuals with DCM, regardless of whether the cause is known. For example, in our research we have identified families in which the cause of DCM in an individual was attributed to a viral infection but upon taking a family history, more family members were reported as having the same condition. In this case, a genetic cause would be more likely.

A pedigree will not only help to determine if the DCM is familial. By obtaining information from at least three generations, your doctor will be better able to determine relationships to establish who in your family is at risk for DCM.

It may be helpful to your doctor if you bring your family history information to your appointment. Let your doctor know about family members who have been diagnosed with DCM, and also about those with symptoms suggestive of DCM (for example: heart failure, heart attack, palpitations, stroke).

Obtaining records from family members who may have had heart disease and bringing them to your appointment will also help your doctor. A useful website where you can write down and print your family history is available at: **www.hhs.gov/familyhistory**.

Offer Clinical Cardiovascular Screening to Relatives of People with DCM

While most DCM cases present with heart failure, the heart enlargement and pumping dysfunction characteristic of DCM can be present without any symptoms. Therefore, the goal of screening is to detect disease before the onset of heart failure or other life threatening events. Detection of early disease by clinical screening followed by intervention may delay disease progression. Screening for DCM is recommended for:

- **Individuals with no signs and symptoms of DCM, but who are at risk for DCM because of their family history.** All first degree relatives (parents, children, siblings) of individuals with IDC should obtain screening. Children and adults in this group should be screened every three to five years. If abnormal, repeat screening should take place in one year.
- **Individuals who show signs and symptoms of DCM.** Individuals with any signs or symptoms (such as palpitations, shortness of breath, chest pain, stroke) should be evaluated right away. Follow-up, screening should be performed according to physician recommendations.
- **Individuals with positive genetic testing results, regardless of symptoms.** Thanks to clinical genetic testing, some individuals know the gene mutation responsible for DCM in their family. Those who test positive for a known family mutation should be screened regularly, and screening should begin as soon as a mutation is detected. Children in this group should be evaluated every year. Adults in this group should be evaluated every one to three years.

Clinical screening for DCM should consist of a four-tiered approach, including a medical history with special attention to symptoms of heart failure and irregular heart beat (arrhythmia), physical examination, electrocardiogram, and echocardiogram.

Although it is unlikely that children will have any symptoms or detectable abnormalities on cardiovascular screening, parents should always be alert for symptoms and have the child evaluated if any signs occur. As a child's heart is naturally enlarging as part of normal growth, screening tests should be performed by centers that can interpret pediatric studies.

Provide Genetic Counseling

Genetic counseling is the process of assisting individuals to make informed genetic decisions while providing emotional support. The process is not meant to coerce a patient into taking a specific course of action; instead, the process of genetic counseling is designed to help patients and their families make informed decisions that work best within their specific life situation.

Genetic counseling for DCM involves the following activities:

- Obtaining a family history
- Education about the genetics of DCM
- Discussing and offering available genetic testing
- Interpretation of genetic testing results
- Exploring relevant social and emotional issues

Consider a Genetic Evaluation Referral

The HFSA guidelines recommend that physicians caring for individuals with DCM consider making a referral for a genetic evaluation. A complete genetic evaluation can be time consuming. Also, cardiovascular genetics is a rapidly evolving field requiring providers to stay up to date at a fast pace. Therefore, some physicians may want to refer their patients to centers with expertise in genetic DCM. These centers can provide genetic counseling and offer additional research opportunities. Please contact us if you would like assistance in locating a center near you.

Consider Genetic Testing

Genetic testing should be considered for individuals with a positive family history of IDC, called FDC. The guidelines are less clear about testing for individuals without a positive family history. However, those without a family history may also pursue testing, as our research has demonstrated that there may be a genetic cause regardless of the presence of a family history.

Genetic testing for DCM has proven to be helpful in a number of families. If the mutation can be identified in the family, it can predict who is at increased risk for DCM.

Mutations responsible for DCM have been found in approximately 30 genes. Genetic testing is available for many of these genes. Only specialized clinical laboratories offer this testing, which can be expensive, although insurance often covers at least a portion of the testing.

A limitation of genetic testing for DCM is that, currently, this testing cannot identify all known genes. The known 30 genes only account for approximately 25-30% of cases. This means that the responsible gene has not been identified in 70-75% of cases. Therefore, a negative result does not mean that a mutation is absent. Genetic testing may also provide inconclusive results. Research groups, such as ours, play an important role in enhancing the currently available genetic testing by discovering additional genes and understanding how mutations lead to DCM.

Obtaining Copy of the Guidelines

The guidelines can be obtained at the ***Journal of Cardiac Failure Vol. 15 No. 2 March 2009***. Most medical libraries carry the Journal of Cardiac Failure. Let your doctor know about these guidelines. Your doctor can also look on 'The Professionals' section of our website for related recently updated information (**http://fdc.to/professionals.htm**) or email us through the website **(http://fdc.to/contact.htm)**.

Supporting the FDC Research Project

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When such gaps in funding exist, we look to other sources to reduce the impact of cyclical funding, which is essential to maintain the continuity and stability of established research personnel and projects. Further, even with NIH support, our funding is limited. Most grant support is restricted to covering costs of personnel and laboratory supplies. Other important activities vital to the progress of a major research program require additional support. Most importantly is seed money to pilot the latest cutting-edge research in the laboratory. For a family based genetic study such as this, maintaining long term follow-up with all of our research participants is also key, but difficult to support with NIH funds. Other expenses include expanded recruitment of individuals at multiple sites throughout the country and new ways of collecting clinical data on research participants.

We ask you to consider financial support of our research program. We will accept your donations of any amount. We have already received several donations from participants that have played a key role in our research, helping to bridge gaps in funding, or allowing us to try exciting new research techniques that otherwise would not be possible. Such donations help to ensure our continued success.

Please feel free to call or email us to inquire about our current funding needs. We thank you for your support of our research project through participation and possibly your financial support. We look forward to continuing to work with you and your family in the years to come!

Examples of How Philanthropy Supports the FDC Research Project

- ♥ Start-up support for new research avenues
  - Personnel
  - Supplies
  - Equipment
- ♥ Bridging support between NIH grants
- ♥ New multi-center study funding
- ♥ Summer student support

All contributions are tax deductible and all funds will be used to support this project. *Donations should be sent to:*

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Checks should be payable to:
University of Miami Miller School of Medicine

FDC BULLETIN BOARD

CALL US AT OUR TOLL FREE NUMBER: 877-800-3430

MEDICAL FOLLOW-UP

If anyone in your family is newly diagnosed with heart problems, please let us know. Also, if you or anyone in your family has had heart tests performed, either for follow-up or for the first time, regardless of results, we would be interested in receiving copies. Please contact us and we will send you a medical record release form. If we have sent you medical record release form(s), please send us the completed form(s) as soon as possible.

EMAIL US THROUGH THE "Contact Us" PAGE ON OUR WEBSITE: WWW.FDC.TO

ADD ME TO THE MAILING LIST

If you are not currently a participant in our study, but would like to receive our newsletter, please contact us with your name and address, and we will be pleased to add you to our mailing list.

CONTACT INFORMATION UPDATES

If you have moved and/or have an email address we can contact you at, please call or email us so we can get in touch with you for any follow-up and continue to send you our newsletter.

FDC BEAT Newsletter

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