

Genetic Counseling for DCM: What to Expect

While many of our participants have received genetic counseling, most participants we've spoken to recently are unaware of genetic counseling and how it would be helpful for both themselves and their family. "The Guidelines for the Genetic Evaluation of Cardiomyopathy," published by the Heart Failure Society of America in March 2009 states that genetic counseling should be part of care for someone who is diagnosed with dilated cardiomyopathy. In this issue of DCM Beat, we discuss what a genetic counselor does and what a cardiovascular genetic session would be like. We also provide answers to some of the most frequent questions that we receive as we interact with participants and their families.

What is 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 guide a patient into taking a specific course of action; instead, the process of genetic counseling is designed to help individuals and their families make informed decisions that work best within their specific life situation.

Although the word "counseling" may give the impression that the process involves some form of psychotherapy, the genetic counselor is more like a support person with specialized knowledge in genetics and the ability to communicate complex information using simple, yet accurate terms. Because having or being at risk for a genetic condition can invoke a range of emotions, genetic counselors are trained to provide guidance and assist with navigating these feelings that can arise during the genetic evaluation process. For example, through genetic testing a family member may find out that they do not carry a genetic predisposition which is present in their family. When compared to other family members who carry the genetic predisposition or actually have the family condition, these otherwise good results could bring about feelings of shame or excessive

guilt. This is known as "survivor guilt." Genetic Counselors understand the complexity and sensitivity of genetic information and have completed specialized graduate level training to develop the skills necessary to properly interpret and communicate results such as this to individuals.

Should I see a genetic counselor?

Deciding whether or not to have genetic testing is a personal choice based on the individual's own circumstances. One of the goals of genetic counseling is to assist during the process of genetic testing. For a person with DCM, genetic testing can be a key component of their cardiovascular evaluation. It offers the possibility of identifying one's genetic cause for DCM, which then allows other family members the opportunity to evaluate their personal risk for DCM.

Clinical genetic testing for DCM must be ordered by a health care provider. This is a complex, rapidly developing field, and therefore it is advisable to consult with a health care professional who has expertise in cardiovascular genetic medicine. This consult should include genetic counseling to ensure the benefits, risks and limitations of genetic testing are properly communicated and understood.

What could genetic counseling be like for you?

First, a targeted family history is obtained. A family history is a summary of your family's medical or health related information. By "targeted" what we mean is history focused on cardiovascular disease is gathered. It is helpful before your session with the genetic counselor to gain as much information as possible about your relatives' cardiac health history. Asking family members for their symptoms or medical records provides a more comprehensive cardiac family history, which will allow a genetic

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counselor to be able to determine if DCM runs in your family and interpret where the heart condition in your family may have started. Also, the genetic counselor will be able to provide the probability other family members may have for developing the same condition.

Screening, preventive and management options are discussed. A genetic counselor will recommend who should be clinically screened in your family, and can advise for other preventative measures to be taken by families dealing with a genetic condition.

If indicated, available genetic testing is offered. There are about 40 known genes in which changes (mutations) may cause DCM. The genetic counselor will discuss your personal and family history in order to decide the best genetic test for you. This consult allows the genetic tests to be most cost efficient and prevent any unnecessary testing.

Relevant social and emotional issues are explored. There are many emotions involved with deciding to receive genetic testing, as well as the results that may come along with this decision. The genetic counselor is there every step of the way, providing you with tools to cope with these emotions.

Results are communicated. If testing is ordered and you have received your results, it is expected that you may have questions. A genetic counselor will call or meet with you to discuss your results to ensure you understand what your result means for you. A report summarizing the consult and results is usually sent to the patient and the referring provider.

Cardiovascular Genetics Clinics

With new genetic discoveries and increasing clinical genetic testing options for cardiovascular conditions such as DCM, cardiovascular genetic clinics have been developed to provide expert cardiovascular subspecialty care, genetic counseling and clinical genetic testing. Cardiovascular genetic clinics offer consultations with physicians who specialize in genetic cardiovascular conditions. Genetic counselors are on staff to provide their expertise in hereditary cardiovascular conditions and genetic testing is also coordinated in the clinic. In addition, some clinics help patients work with their insurance company to determine genetic testing coverage. Other services offered by cardiovascular genetic clinics include:

- Screening and diagnosis of cardiac conditions
- Genetic testing results disclosure

- Ongoing cardiac follow up
- Enrollment in research studies

Enrollment in Research Studies

One of the current limitations of clinical genetic testing for DCM is that some test results may not be informative. This means that the result obtained cannot be interpreted; the result is neither positive nor negative, but uninformative. Research provides a venue to further explore this type of result in hopes that more definitive answers can be obtained in the future and cardiovascular genetic clinics can provide an environment for this research. Research such as our very own “DCM Research Project” can be aided with the participation of individuals seen by cardiovascular genetic clinics. Research genetic testing, which is what we at the DCM Research Project do, is an option when clinical genetic testing is unavailable or results come back uninformative.

Frequently Asked Questions about Genetic Counseling and Testing for DCM

We spend a considerable amount of time on the phone and corresponding with our research participants. We truly enjoy speaking with you and helping in any way we can to raise awareness about DCM and available testing options. Below are some of the most frequent questions about genetic counseling that we have received in the last year. Please keep the questions coming; we want to hear from all of you!

What type of training is required to become a genetic counselor?

Genetic counselors must obtain a Master’s degree in Genetic Counseling from an accredited program. Once the training requirements are met, candidates must pass a national certification exam to demonstrate that the standards necessary to provide competent genetic counseling services have been met. Some states, such as Ohio, also require that genetic counselors have a professional license in order to practice as a genetic counselor.

How is a genetic counselor different from a nurse?

While nurses provide physical exams, medication guidance and do some procedures, such as blood draws,

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genetic counseling training does not focus on these skills. The genetic counseling scope of practice includes assessing family history and patterns of disease, providing risk and inheritance counseling (including psychosocial concerns), identifying and interpreting appropriate genetic testing, and effectively communicating their significance to patients and their family.

What do I need to bring to a genetic counseling appointment?

Prior to your cardiovascular genetic counseling appointment, it is helpful to prepare questions that you and your family have about the hereditary nature of DCM or any other heart problems that are running in your family. During your visit, the genetic counselor will review your family history in detail with you. Collecting as much family history information as possible from your relatives before your appointment will help you get the most out of your consult. When obtaining family history information, take special note of any heart attacks, sudden cardiac deaths, heart failure, arrhythmia, fainting, sudden unexplained deaths, any procedures such as internal cardiac defibrillators (ICDs), pacemakers, or any cardiac surgeries.

Will I see a doctor during my genetic counseling appointment?

All clinics are different, but in most cases, yes. You may see a cardiologist and/or a medical geneticist in addition to the genetic counselor. A cardiologist will be responsible for ordering procedures that help to establish if a heart condition is present. Depending on the extent of your condition, the cardiologist will recommend a follow up plan that may include medication or additional proce-

dures. When health conditions outside of the cardiac system are observed or suspected, consultation with a medical geneticist will be sought. The medical geneticist is a physician that specializes in conducting a full body physical exam that may help to establish a more accurate diagnosis and therefore, better risk assessment for family members and appropriate genetic testing recommendations.

What is the benefit of pursuing clinical genetic testing?

If a mutation is detected that causes DCM, then that mutation can be searched for in at-risk family members. If unaffected relatives are positive for the DCM-causing mutation that runs in their family, they can pursue early and frequent screening and beginning a regimen of preventative medication such as ace inhibitors or beta-blockers. For example, if a person who has DCM is found to have a DCM-causing mutation, then other at-risk family members can be tested to see if they carry the same mutation. If a seemingly unaffected family member is tested and found to carry the mutation, they will be at increased risk to develop DCM and can therefore undergo early intervention to try and prevent or lessen the severity of DCM.

Can I get genetic counseling through the study?

Although we do have a licensed genetic counselor, Ana Morales, on our staff willing to answer any of your questions, this is not a substitution for genetic counseling. If you are interested in receiving a genetic consult, you can talk with your doctor or visit the National Society of Genetic Counselors at www.nsgc.org or call 312-321-6834. We are also happy to help you find a genetic counselor near you. Please contact us!

Welcome Jennifer Rinehart!

Our project continues to flourish and new staff follows along. Jennifer Rinehart, BS joined our group in October 2013 as a full time research assistant. Jennifer will be supporting our research by enrolling participants and helping implement special projects. Please join us in welcoming her! She introduces herself in this issue of DCM Beat:

I am honored to be part of the DCM research team here at the OSU Wexner Medical Center! As a Research Assistant on the team, I will be assisting with the enrollment of new participants and acting as an additional point of contact for current study participants. I have a great understanding for families with heritable conditions, as personal life experience with this sparked my initial passion for the field of genetics. I completed my Bachelors of Science in Molecular Genetics at the Ohio State University in 2011 and witnessed firsthand the University's commitment to advances in science, especially in the field of genetics. I am so pleased to be working on a team that puts their heart into the research process. My other passions include exploring the city of Columbus, singing in a band, and meeting new people, which is why I am so looking forward to working with all of you!

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Clinical genetic testing?



While our research continues even after a research result is identified in a family, we recommend that all individuals with DCM consider undergoing clinical genetic testing. This testing was not routinely available when our study began in 1993. Clinical genetic testing is done similarly to any other blood test that is ordered by your doctor and sent out to a laboratory. Your results would be provided to your doctor. According to medical guidelines for the evaluation of cardiomyopathy, clinical genetic testing can be a complex process. Therefore, referral to centers expert in genetic evaluation should be considered. We can help you identify a clinic offering genetic counseling and testing for DCM. *If you have undergone clinical genetic testing and have results, please provide us with a copy of your result for our database.* This information will help us in our approach to identifying the gene or genes that may be causing DCM as well as how these mutations lead to DCM. Please contact us (toll-free) at **877-800-3430** or email Ana Morales, MS, LGC at ana.morales@osumc.edu.

MEDICAL UPDATES



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 or genetic tests performed, 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.

CONTACT INFORMATION UPDATES



If you have moved and/or have a new phone number or email address, please let us know. Call **877-800-3430** or email us through the "Contact Us" page on our website: www.dcm.to. This way we can get in touch with you for any follow-up and continue to send you the newsletter.

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.

DCM BEAT Newsletter

DCM Beat (formerly FDC Beat) is a publication of the Dilated Cardiomyopathy Research Project (formerly Familial Dilated Cardiomyopathy Research Project) in the Division of Human Genetics at The Ohio State University, in Columbus, OH. The newsletter is not copyrighted and readers may photocopy its content to share with family members and health care professionals. We welcome your feedback.

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