DCM beat

Newsletter of the Dilated Cardiomyopathy Project at The Ohio State University
dcmproject.com
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The DCM Research Study Announces Major Expansion

Greetings! Our last issue of DCM Beat was published in Spring 2014. If you have been wondering if the study is still active, the answer is: Yes! After a great deal of effort we have been extremely fortunate and have received new, major federal funding from the National Institutes of Health. With that we have undertaken a major reorganization, not only of our research, but also our branding. We have redesigned DCM Beat with a new look and feel. We will continue using it as a vehicle to stay in contact with our participants while providing study updates and breakthroughs in the genetics of DCM.

So what has been happening with our research study? For those of you who may be reading DCM Beat for the first time, we should explain how we have evolved over time. In 1993 we began studying DCM genetics in The Familial Dilated Cardiomyopathy (FDC) Research Project. In 2013, to better communicate our intention of enrolling participants with and without familial DCM, the study was renamed The Dilated Cardiomyopathy (DCM) Research Project. Please see our website if you are interested in more history of the Project (www.DCMProject.com).

A New Study: DCM Precision Medicine

In 2015 the DCM Research Project received funding for a new study called the DCM Precision Medicine Study. The aim of this study is to prove that DCM, regardless of whether it is familial or non-familial, has a genetic basis.

The term "Precision Medicine," was coined first in 2011. We created and started to seek funding for the DCM Precision Medicine study in 2013. In 2015 the precision medicine term gained prominence when President Obama announced the Precision Medicine Initiative. Sponsored by the National Institutes of Health, the Precision Medicine Initiative aims to use genetic information from a large number of individuals to advance biomedical discovery and the practice of medicine.

The DCM Precision Medicine Study is now underway to enroll 1300 new DCM families. To support this goal, we created the DCM Consortium as a new initiative of the DCM Research Project. The DCM Consortium is an investigator group from different sites around the United States to conduct genetic research on DCM.

How Does the DCM Precision Medicine Study Work?

Participating in the DCM Precision Medicine Study requires the first member of a family to enroll to have DCM, and this individual's participation requires in-person enrollment at any of the DCM Consortium sites. Enrollment also requires a blood draw, family history, release of cardiovascular records, enrollment of family members, and completing yearly surveys. Genetic testing results are provided. The DCM Precision Medicine Study requires that family members enroll in the study. Family members may enroll by mail. While in the past we did not make family member participation a requirement, the success of the DCM Precision Medicine Study depends on family members' participation. This study also involves the evaluation of a family communication tool in new DCM families. For this reason, individuals previously recruited in our study, now called the DCM Discovery Study, are not eligible to participate in the Precision Medicine Study.

The following individuals are also not eligible to participate in the Precision Medicine Study:

- Individual with DCM, and is unable to enroll in person
- Individual with DCM is adopted and does not know his/her blood relatives
- Individual with DCM is not in contact with his/her family members
- Family members of individual with DCM are not interested or unable to participate

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Individuals not meeting criteria for the Precision Medicine Study, as well as those enrolled in the DCM Research Project until June 2016, are now part of the DCM Discovery Study. Participation involves a blood draw, family history, and release of cardiovascular records. DNA is collected for future research. Genetic testing results may, or may not, be available. Participation can be done completely via telephone and mail.

Our Study Since 1993 Is Now Named The DCM Discovery Study

As mentioned above, all families enrolled since 1993 in the FDC Project, renamed the DCM Project in 2013, and now the DCM Discovery Study, remain a fully maintained and extremely valuable component of our research program.

You have all contributed greatly to our efforts for discovery and to understand DCM genetics, and we are forever grateful for the wonderful ongoing participation of hundreds of families prior to 2016.

A progress report of the DCM Discovery Study in early 2016:

- All of our publications prior to 2015 have been from the DCM Discovery Study.
- By 2015 we had identified plausible genetic cause for approximately one-third of the more than 300 families who had undergone sequencing for 16 genes in 2005 and 2008.
- By mid-2015, exome sequences have been produced for approximately 450 individuals in the Discovery Study.
- Because of funding limitations, not all participants have had their DNAs sequenced, but we remain committed to obtain funding to sequence all DNAs from all of our families. DCM gene discovery has been, and remains an essential and central focus of our work.

New Website Redesign

As with our newsletter, we have redesigned our website. The new URL is www.dcmproject.com. The website includes more information about the DCM Precision Medicine and DCM Discovery Studies. The goal of our website is to provide relevant information about the genetics of DCM in families for all study participants. We also aim to inform health care professionals with our recommendations to manage genetic DCM, and provide an update on DCM research.

Introducing New Personnel

While Ana Morales remains the main study contact for Discovery Study participants, we are delighted to welcome our new study personnel, who will work on the Precision Medicine Study. They will be happy to assist and answer any questions:

Esther Barlow, Study Manager

Esther Barlow, BS, brings with her 10+ years of experience in clinical research. Esther earned her bachelor's degree in biology, is ACRP (Association of Clinical Research Professionals) certified as a study coordinator and currently serves on the ACRP Greater Columbus Chapter committee board as vice president.

"To come into the DCM Research Project at a pivotal time when genetics play an integral part in precision medicine is fascinating. Dr. Hershberger's passion in DCM research is clearly evident in his commitment, and dedication. He truly cares for how research can help the population and the very people (participants) we encounter. My role as a clinical research manager for the DCM Precision Medicine Study is to work alongside our study team, and ensure that the study is successful in its entirety. We thank all the participants for their time and contribution; they are the fabric of this very important study."

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Caty Escobar, Study Coordinator

Caty Palma Escobar joins the study with a BS in Psychology and minor in Latino Studies. Caty, who is bilingual, has previous experience in cancer clinical trials, diabetes and special education research, particulary in working with families of Hispanic ethnicity.

"The DCM study allows significant involvement in an exciting area of research and promotes a collaborative environment among all of our consortium sites. Understanding the role of genetics in the development of DCM has become much more important as researchers have been able to link genetics with the onset of disease. Nonetheless, this work must continue! With the DCM study, we have the potential to reach a large number of individuals from ethnic backgrounds that are affected by this disease. The inclusion of families

from different racial and ethnic backgrounds will help our researchers achieve a better understanding of the disease development and progression."
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Caty.Palma-Escobar@osumc.edu

Catherine Roth, Study Coordinator

Catherine Roth, BS, MPH has a background in Public Health Epidemiology. She plans to bring her knowledge of surveillance and analyses of populations to help with the precision medicine initiative.

"I am honored to be working on the DCM Research Project and excited to be part of such a large family-based research study focused on precision medicine. This is such an important and exciting time in research and I can't wait to see what kind of results this study will lead to. A big thanks to all of the participants and family members, we couldn't achieve this without all your dedication and your contributions!"
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Somayya Mohammed, Study Coordinator

Somayya J Mohammad, BS brings her diverse experience in clinical trial data management along with her knowledge of public health, public relations and clinical pharmacology research to the team.

"I am quite priveleged to be involved with the DCM research project and research team here at The Ohio State University. Genetic research is the new frontier of medical research, aiding in the attainment of precision medicine initiative, however, using genetic research for prevention is a novel and exhilarating approach especially on this scale. This project is especially important because it is proposing a unique approach of identifying genetic markers and using regular cardiovascular screenings to identify as well as manage DCM in the asymptomatic phase. So, eventually knowing your family's history with DCM would translate into regular preventive cardiology visits, covered under preventive services by all insurance plans for all 1st degree family members, resulting in intervention before reaching heart failure. I look forward to working on this project with everyone."
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DCM Precision Medicine Study: Key Points to Remember

1. **OSU is the coordinating site for the DCM Precision Medicine Study.** For any study related questions, you may contact our study personnel at OSU or at the site where you were consented.

2. **We need family members to participate.** Family members will allow us to best determine how much of DCM runs in families. Participation of family members is also important as we evaluate if a gene change is responsible for DCM in the family.

3. **Every individual consented will have a follow up phone call on an annual basis for the duration of the Precision Medicine study.** This phone call is to go over a survey with the purpose of learning more about how you communicate with family members about the genetics of DCM.

4. **One year after enrollment, we will inform participants of their genetic testing result.** If results were negative, we will also inform participating family members.

5. **Cardiovascular screening of first degree family members of individuals with DCM is recommended.** This screening consists of an echocardiogram and an ECG. This can be accomplished via personal or private physicians or, at no cost, from within the Precision Medicine study.

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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 a medical record release form(s), please send us the completed form(s) as soon as possible.

CONTACT INFORMATION UPDATES

If you have moved 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.dcmproject.com. 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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