

FDC



BEAT

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

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The Familial Dilated Cardiomyopathy Research Project 2005

Hello from the Familial Dilated Cardiomyopathy (FDC) Research Group at Oregon Health and Science University. As 2005 comes to a close, this issue of our newsletter will review for you our activities this past year. It has been a busy year here at the FDC study, with several major projects undertaken. While our time and effort towards this year's projects delayed our newsletter publishings, we hope these improvements will be beneficial to the study and contribute to making participation a positive and useful experience for individuals and their families. We hope to be back on schedule with two to three newsletters per year in 2006. Until then, enjoy this issue of the newsletter, and Happy Holidays and best wishes in the New Year for all.

FDC LAB BECOMES CLIA CERTIFIED (and new equipment, too!)

This past summer the FDC lab completed the certification process for the Clinical Laboratory Improvement Amendment, or CLIA. CLIA was an act passed by Congress in 1988 to establish quality standards for laboratory testing. By becoming CLIA-certified, a lab ensures that the testing they provide is accurate and reliable.



The CLIA-certification process included an application, fees, and inspection of our lab, which we are happy to say we passed in July!

Continued visits from CLIA inspectors every two years will ensure that we continue to comply with proper lab standards.

Many research labs do not have CLIA certification, and therefore are unable to provide research participants with individual test results. So now that we are CLIA certified, what does this mean for study participants? It means that if we contact you about finding the gene(s) we think causes FDC in your family, you will now have the option of getting your individual genetic test results. By submitting a fresh blood sample, CLIA testing would be performed and you would receive a written copy of your test results.

While receiving genetic test results may become an option for some families, CLIA certification does

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not guarantee if or when we might identify the FDC gene(s) in your family. If we contact you about an FDC gene in your family, our genetic counselor, **Jessica Kushner**, will further discuss with you the option of receiving genetic test results, and what this might mean for you and other family members.

The CLIA process took a great deal of time and effort, especially for our lab director, **Sharie Parks, PhD**. Thank you Sharie, for making this happen!



In addition to CLIA-certification, in the past year our lab has had some major upgrades to both the lab itself and to major lab equipment. The lab's layout was redesigned and reconstructed to allow for better organization and more workspace. This was a huge undertaking for all involved. We also upgraded two crucial pieces of equipment - a genetic analyzer and a TCGE instrument. These machines process blood samples to analyze DNA and screen genes for changes (mutations) that may be responsible for FDC. These new machines are able to process more samples at one time and in a more efficient way than the ones previously used. Computer programs that help interpret the information from these instruments have also been upgraded.

All these laboratory improvements will directly contribute towards our ultimate goal of identifying the genetic factors of dilated cardiomyopathy.

FDC DATABASE DEVELOPMENT

There are over 300 families and almost 1700 individuals that are currently participating in the FDC Study. As you can imagine, with this many participants there is an enormous amount of information to keep track of – contact information, blood and medical record data, family history information, consent tracking, etc. Back in 2002, we purchased a powerful computer database program called Progeny. Progeny is designed for family-based genetic studies, and links family

trees (pedigrees) with individual family member data. It has built-in security and is password protected, only accessible to authorized FDC study personnel. It draws pedigrees with the click of a few buttons, and helps us organize and track all of our data.

Understanding what FDC “looks” like is a crucial part of studying this disease and the genes that cause it. With so many people in the study we needed a computerized way to analyze all the medical data from our participants, to compare such things as age of onset of disease, echo and EKG findings, need for transplants, pacemakers, defibrillators, etc. With medical records of over 1000 people, it took us over 2 years to review and enter all records data received since the start of the project back in 1993.

We completed this project this past September, thanks to a group effort from FDC clinical and bio-informatics personnel (see FDC personnel below). Initial analysis of data has been performed, and will be especially important to apply to any FDC gene findings.



We continue to enter medical data into Progeny as updates are received and new participants are enrolled. So please, as always, **keep us updated on any changes in your medical history and that of your family** so we can request any new medical records for our database to be as accurate as possible.

FDC WEBSITE UPDATE

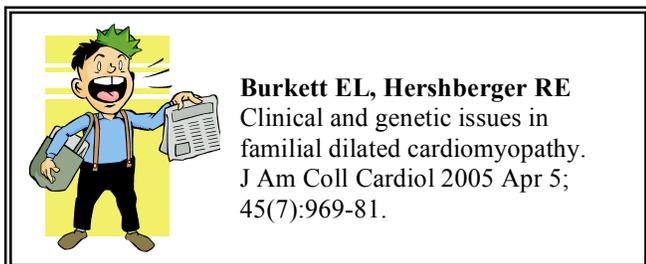
One of the main benefits of a website is the ease with which the most current up-to-date information can reach numerous people anywhere in the world (provided access to the internet, of course). With FDC knowledge always changing and advancing, another project we completed this year was an extensive review of our website, **www.fdc.to**. Each website section was edited and updated, such as the list of FDC-associated genes (now numbering 23), and information regarding availability of CLIA-certified clinical genetic testing. In addition, current and



back-issues of our newsletter, FDC Beat, are now available on the website. Thanks to **Susan Ludwigsen**, Sr. Research Associate, who manages our website, and spent a great deal of time on this update.

FDC GROUP MANUSCRIPTS AND PUBLICATIONS

Publication in a major scientific/medical journal is no small feat. It is an excellent way to share research information with colleagues and contribute to knowledge in the field; but with many research groups studying many different diseases and all wanting to publish, it is very competitive. In April 2005, our group had a manuscript published in the Journal of the American College of Cardiology. This article reviewed what is known about FDC and its genetics. The full reference is below. If you would like to obtain a copy of this publication but are not able to, please contact us and we will assist you.



There are a number of other manuscripts we have been working on this year and submitting for publication. We will keep you updated in future newsletters on any additional publications.

FDC PROJECT APPLIES FOR REFUNDING FOR 2007

In November 2005, we submitted a grant renewal proposal to the National Institutes of Health (NIH), requesting that they continue to fund the FDC project. The NIH originally funded the FDC Project in 1998 for 3 years, and refunded us in 2002 for five more years. Applying for refunding is a long process (hence our submission in 2005 for 2007 money) and is extremely competitive, particularly in current times with minimal increases in the NIH's budget for research funds.

All FDC Project staff (particularly our principal

investigator and director, **Dr. Hershberger**) worked very hard on putting together a good renewal proposal (almost 70 pages long!), so that we can continue our work to improve the understanding of the genetics of FDC, and find better treatment and prevention options.



FDC PERSONNEL UPDATE

The FDC staff is a committed group, with some folks working at OHSU for over 20 years! There have, however, been some changes in our personnel recently. With most of our resources put towards the laboratory's work in identifying FDC genes, FDC Research Assistant positions were not refilled (previous assistants Kelly Smith and Warren Toy left Portland earlier this year). However, our research nurses, **Deirdre Nauman** and **Donna Burgess**, have been extremely helpful in enrolling local participants and screening family members. We also have student assistants who help with our blood kits, filing, and medical record requests. **Jessica Kushner**, our genetic counselor and research associate, is still the main contact for study participants. We do want to hear from you with questions and updates on your health. We appreciate your patience as we address your phone calls and emails as quickly as we can.

THE FDC PROJECT TEAM CLINICAL GROUP:

Ray Hershberger, M.D.
Jessica Kushner, M.S., C.G.C.
Deirdre Nauman, B.S.N., R.N.
Donna Burgess, B.S.N., R.N.

BIO-INFORMATICS: Susan Ludwigsen, B.A., M.A.

LABORATORY GROUP:

Michael Litt, PhD.
Petra Jakobs, PhD.
Duanxiang Li, M.D., M.S.
Sharie Parks, PhD.
Amanda Peterson, B.S.
Julie Partain, B.S.



AND...**YOU!!** Thank you for your important contributions to this research!

FDC BULLETIN BOARD

Notes, Announcements, and Reminders

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 heart tests performed, regardless of results, we would be interested in receiving copies of these results. Please contact us and we will send you a medical record release form.

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



REMINDER

If we have sent you blood tubes and/or medical record release forms, please send back your blood and paperwork as soon as possible. Please call us if you have any difficulty finding someone to draw your blood.

UPDATES



If you have moved and/or have an email address we can contact you at, please call or email us so we can update your information in our database.

FDC BEAT Newsletter

FDC BEAT is a triannual publication of the Familial Dilated Cardiomyopathy Project in the Division of Cardiology at Oregon Health and Science University in Portland, Oregon. The newsletter is not copyrighted and readers are welcome to photocopy its content to share with family members and health care professionals. We welcome your feedback. **Authors and Newsletter Layout/Design:** Jessica Kushner, MS, CGC; Ray Hershberger, MD; Susan Ludwigen, MA.

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