FDC BEAT

Newsletter of the Familial Dilated Cardiomyopathy Project at Oregon Health Sciences University
Portland, Oregon, USA **Volume I, Issue 3** **October, 2000**

The Human Genome Project:

Completion of the Draft Sequence and Impact on FDC Research

This summer, leaders of the Human Genome Project (HGP) announced the completion of the draft sequence of the human genome. What impact will this achievement have on our progress in identifying the genetic causes of FDC? While this is a major milestone in a series of steps important to the future of genetic medicine, we should be cautious not to overestimate the direct impact it will have on our project in the short term.

First, what is the HGP and what has it accomplished? The U.S. Human Genome Project is a 15-year endeavor coordinated by the U.S. Department of Energy and the National Institutes of Health to determine the sequence of the human genome. A genome, or all of the DNA in an organism, can be thought of as a long book containing the instructions for an organism's growth and development. The DNA "alphabet" has four letters, or nucleotides, which pair A with T and C with G. Specific regions of DNA, called genes, are responsible for making proteins which perform crucial, specialized functions throughout the body. The goal of the HGP is to eventually determine the order of the 3 billion pairs of letters and to identify the locations of the approximately 80,000 genes within the book.

In June 2000, the completion of the draft sequence of the HGP was announced. The draft sequence comprises about 90% of the human genome. It is not as accurate or as continuous as the finished sequence will be. Over the years, the gaps and errors in the draft sequence will be corrected to achieve the ultimate goal of the HGP: a complete, high-quality human DNA reference sequence by 2003 (every letter of the book, from end to end). The fact that 90% of the sequence has been determined, however, does not imply that the genes have all been located or that their function is known. The next step is to identify which section of DNA are genes and which are "junk" DNA that do not encode protein. Ultimately, the goal is to discover the genes' functions and how changes in genes cause disease.

The completion of the draft sequence will not change the way we do FDC genetic research, but it may help to accelerate the process, from mapping genes in our families to cloning those genes and understanding their functions. The invention of new methods for diagnosing and treating genetic conditions may in turn also be accelerated by the HGP.

For further information on the Human Genome Project, see the official HGP website:

http://www.ornl.gov/hgmis/

Human Genome Project information from the U.S. Department of Energy

FDC BEAT

For patients and families, we hope to educate you about familial dilated cardiomyopathy, inform you of our efforts to understand why and how it develops, counsel you about some of the genetic and family issues of this disease, and seek your support for our ongoing efforts.

Behind the Scenes:

Spotlight on Petra Jakobs, PhD

The FDC Research Project is composed of two main groups: the clinical group and the basic science group. Most of our research participants are familiar with the clinical group, as they are the people participants frequently have direct contact with either on the phone, through the website or in person at various screening events. We feel it is important, however, to introduce you to some of the people in the basic science group who are hard at work behind the scenes and are essential to the success of our research project.

In this issue of the FDC BEAT, we would like to spotlight Petra Jakobs, PhD and the work she does as a member of our basic science group. Dr. Jakobs is a Research Assistant Professor of Medicine in the Division of Cardiology at OHSU with an extensive background in molecular genetics. She received her doctorate at Justus-Liebig University in Giessen, Germany. Dr. Jakobs has been involved in various other medical genetic research projects at OHSU, including studies on Fanconi anemia and cataracts, prior to her joining the FDC Research Group. She began her work with the FDC Research Group in 1998 as a basic scientist who is an expert in conventional methods of pedigree genotyping and gene mapping. She provides full-time support to the FDC Research Project and supervises the day-to-day operations of all laboratory personnel. Recently, Hugh Keegan was recruited as a Research Laboratory Assitant to assist Dr. Jakobs with her work on FDC. Hugh is a recent graduate of Bowdoin College in Brunswick, ME, where he majored in Biochemistry.

After your blood sample is received, the different tubes are distributed to various laboratories on campus. Dr. Jakobs receives the purple top tubes (labelled with an ID number to maintain confidentiality), from which she extracts DNA (genetic material) that can be used for linkage analysis and gene mapping. DNA is isolated from the blood immediately upon receipt of the purple top tube. Once the DNA is isolated, it can be stored for future use.

As you can see, Dr. Jakobs serves a vital function within the FDC Research Group. Her daily work will hopefully bring us closer to our long-term goal of determining the genetic basis of FDC.

Suggestions, Questions, Comments, Ideas???... We Want to Hear From You!

Our FDC Families are at the heart of all we do. Therefore, it is extremely important to us that we receive feedback from you so we can improve our program and address any confusion, questions, ideas or concerns that you might have in an upcoming issue of FDC BEAT. What have your interactions with the FDC Group members been like? How has FDC affected your family? Are there aspects of FDC which are confusing or needing clarification? What are some elements about the disease which you feel should be addressed? What kind of a screening experience did you have? Chances are that if you have suggestions, questions, comments or ideas, so do many others, so please let us know so we can help. Submissions may be published anonymously at your request. You will be contacted prior to publication. If you have any contributions (i.e., questions, stories, comments, or suggestions) or if you wish to no longer receive FDC BEAT, please:

1. call us toll-free at 1-877-800-3430
2. visit out website at *http://www.fdc.to* and send an email from the "Contact Us" page
3. email us at *messages@fdc.to*

4. write us at : FDC Research Project
   Attn: FDC BEAT
   Division of Cardiology, UHN-62
   3181 SW Sam Jackon Park Road
   Portland, OR 97201

Log on to the FDC Project website at *http://www.fdc.to* for access to articles, information and links to other FDC topics and resources.

Grant Received for Rescreening

The FDC Research Project began in 1993, and the first families screened now need to be rescreened. A small grant from the Medical Research Foundation through OHSU has been awarded to Dr. Kathy Crispell for the purpose of rescreening our first few families.

We are frequently asked how often a person in a family with known FDC should be rescreened. FDC is a disease that can present at any age and frequently occurs without symptoms, making it difficult to identify those who carry the genetic alteration. Currently, we are recommending that family members be rescreened every 3-5 years if they remain without symptoms or sooner if they develop symptoms consistent with the disease. However, the answer is not clear. By formally rescreening families who have been previously screened, we may be able to better determine an answer based on scientific evidence. The repeat screening allows us to observe, track and analyze changes that may be occurring in family members who carry the gene change. This information may help us to determine what the first clues are that a person may have the genetic change that causes FDC. The information may also help us to determine when symptoms are likely to occur relative to some of those initial clues. We believe that some of the first clues of the disease include enlargement of the heart, abnormal heart muscle function and certain abnormalities found on the electrocardiogram.

There are other benefits of being rescreened. We may identify people who did not have any symptoms of the disease on their first screen but have developed some signs since then. Through the repeat screenings, we will learn about new family members, update the family tree and screen family members not previously screened. The rescreenings also benefit the family by raising awareness. This is also a good time for us to update family members about the latest information regarding FDC and gives family members an opportunity to ask us questions.

We are currently in the planning stages for rescreening families we studied 5-7 years ago. We will be contacting these families and planning the re-screenings accordingly. You will be notified if and when we decide to rescreen your family. It is very important for everyone to keep his or her contact information updated. You can contact us with any updated information by phone at our toll-free number (1-877-800-3430) or through our website (*http://www.fdc.to*).

Helpful Website Resources

We understand that many of our participants have questions regarding various issues related to FDC. In an attempt to help answer your questions, we have compiled a list of websites related to understanding the genetics behind FDC. We hope that these can help provide you with more information to further your knowledge of FDC and other related health and genetic issues. Of course, there is always our website at *http://www.fdc.to*.

GENETIC EDUCATIONAL WEBSITES

http://www.hhmi.org/GeneticTrail
Howard Hughes Medical Institute, educational information about various topics related to genetics and the impacts of genetic research

http://www.ornl.gov/hgmis/
Human Genome Project Information

http://www.geneletter.org
The Gene Letter, a newsletter dedicated to advances in genetics.

http://www.kumc.edu/gec/
The University of Kansas Medical Center; educational information on human genetics and many links to other genetics sites.

http://www.vector.cshl.org/dnaftb/
DNA from the Beginning, an animated primer on the asics of DNA, genes and heredity.

FDC BEAT Newsletter

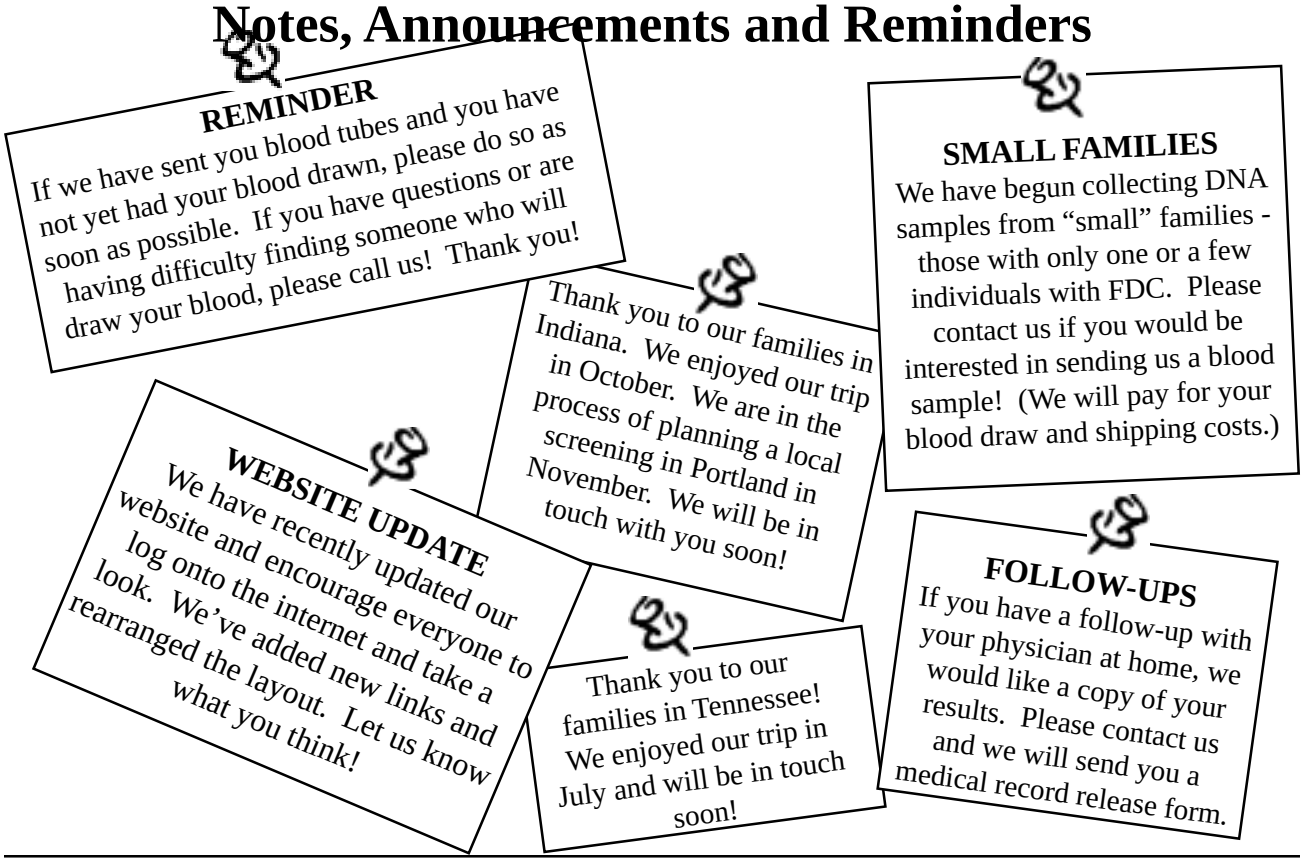
FDC BEAT is a triannual publication of the Familial Dilated Cardiomyopathy Project in the Division of Cardiology at Oregon Health Sciences University in Portland, OR. The newsletter is not copyrighted and readers are welcome to photocopy its contents to share with family members and health professionals.

FDC Project Clinical Group:
Ray Hershberger, M.D.
Kathy Crispell, M.D.
Emily Hanson, M.S.
Warren Toy, B.S.
Kelly Coates, B.S.

Newsletter Layout/Design: Kelly Coates, B.S.
email:*coatesk@ohsu.edu*

Graphic on front from: *http://www.nhgri.nih.gov/DIR/VIP/SI/*

FDC Bulletin Board
Notes, Announcements and Reminders



The FDC Research Project
Division of Cardiology, UHN-62
Oregon Health Sciences University
3181 SW Sam Jackson Park Road
Portland, OR 97201

Address Service Requested

TO: