

## Frequently Asked Questions about GENECARD Study Participation

### **Now that there are initial results, what happens next?**

The next step in identifying the genes that are associated with EOCAD is to expand the analyses that we have begun in the first genome scan. We will genotype more families and more individuals within the participating families. We will narrow our focus to the chromosome regions where we found a relationship, or "linkage," in the first genome scans. Looking for a specific gene in the human genome is like trying to find a location on a VERY large map. We may not have the exact "address" of the genes as of yet, but, we believe the first genome scan has helped to find "the right road."

We will also look closely at "candidate genes" in the chromosome regions we have identified. These "candidate genes" are genes that we suspect might have a relationship to heart disease based on their biological function and results from other research. We have a tremendous start on this line of study from the work of Dr. Pascal Goldschmidt-Clermont, Chairman of the Department of Medicine at Duke University Medical Center. Dr. Goldschmidt-Clermont has identified many genes that are present and active in greater numbers (referred to as expressed) in diseased heart and blood vessel tissue than in normal, healthy tissues. These genes are major targets for early study.

### **Do you need participation from more individuals and more families?**

This is truly a case where there is strength (and knowledge) in numbers. The study could benefit greatly from the participation of additional families, and even more importantly, the participation of more individuals in the families already participating in the study. The study would especially benefit from the participation of the children (offspring) of the individuals that have participated earlier in the study.

### **How do you select families for the study?**

We try to identify families where there are at least two brothers or sister with early onset cardiovascular disease. The condition is rare enough before the 50s that there is almost certainly an inherited factor in such families.

**What members of these families are asked to participate?**

All adults (over age 18) members of the family are eligible to participate. It is important for us to study other brothers and sisters, parents, offspring, and other willing family members to see how the genes are distributed within the family.

**Why is it important to include brothers and sisters that don't have heart disease?**

We are placing great emphasis on including brothers and sisters without heart disease. Including brothers and sisters is very important in studying whether there is an association between a gene and a disease. For example, it raises our suspicion of a certain gene if we see that the siblings possessing a certain gene have coronary disease and siblings without the gene do not.

**Why are you asking spouses to participate?**

To add to the strength of any study, you need to include 'controls' or a comparison group that you compare to the individuals that meet the study guidelines. Spouses make a very good comparison group because while they are genetically different from the study family, they share a similar environment as their spouse. Many studies have shown that spouses have a tendency to share common lifestyle behaviors such as diet, smoking and exercise. Participation by spouses will help us to sort out causes of heart disease that may be environmental rather than inherited.

**Why are you including (adult) children within these families?**

As we continue to understand more about the genetic factors in heart disease, it will be important to know what is happening with the heart health of the next generation within the study families. We aim to continue the study and analyze longitudinal (over time) data within the families until we identify the actual genes and can target them for treatment and prevention.

**Why are you asking to do follow-up?**

People may participate in the study even if they do not wish to be contacted again for follow-up. However, it is very helpful if you are willing to return a self-addressed, stamped questionnaire to the study staff that you will receive about once a year. The questionnaire will ask for an update on your heart health and related conditions like diabetes and high cholesterol. Because health changes over time, we need to know if we have people identified correctly as having heart disease or not having heart disease.



### **What is involved in participation?**

Individuals are asked to contribute 37 cc (7 and a half teaspoons) of blood **once** and answer questions about their medical and family history. The questionnaire takes about 15 to 30 minutes to complete. You will also receive a follow-up questionnaire about once a year unless you ask that we not send the questionnaire to you.

### **Where do I get my blood drawn?**

When you talk with the study staff, they will help you determine the best way for you to have your blood sample drawn. You will need to sign the "Consent for Research" which explains the study in detail before you have blood drawn and sent to the research laboratory. The study staff will discuss which works best for you from the following choices:

If you live near Duke University Medical Center or one of the participating study sites, an appointment can be made for you to have your blood drawn at the research office.

Study staff can send you a kit containing empty tubes so that you can have your blood drawn at your doctor's office or a lab near your home. Many patients have blood drawn regularly for cholesterol or other tests. This is a good time to have the person drawing your blood collect the extra tubes for the study. The study will reimburse your lab if there is a charge for the blood draw. You will be given a Federal Express pre-paid mailer to use in shipping the blood to our laboratory.

Staff with the research studies may visit you in your home or at another convenient location to interview you and draw your blood. Study personnel travel throughout the country to visit with families for many studies and try to visit with as many families as possible when they travel to distant locations.

### **Is the study confidential?**

Data and samples going into the database are assigned a unique identification number (a barcode label) and do not contain any information from which an individual or family might be identified. The Duke clinical teams involved with collecting samples and data have are the only individual with access to the identity of participants.



**Is there a charge for participating in the study?**

Duke covers all expenses of the study. Participants are not charged to participate and are not paid to participate.

**What are the benefits of the study?**

There are no benefits directly to participants, however, knowledge gained from the study may be of benefit to people at risk of heart disease in the future.

**How do I learn more about the study?**

Please return the questionnaire included with this newsletter or send a note to: attn: Elaine Dowdy, Box 3022, DUMC, Durham, NC 27710.

You may also contact us toll free by phone at 866-643-2785, or email at [dowdy002@duke.edu](mailto:dowdy002@duke.edu)

The study staff members are happy to talk with you about any questions you may have now or in the future.