About Research Studies

Research studies try to learn information that may help others in the future. People who join studies may not get any direct benefits. Being in this study is up to you. It will not affect the medical care you get from UNC Hospitals.

It is important that you read and understand the information in this brochure so that you can make your own decision about whether or not to join the study. You will also be given a copy of the consent form for the study and we will discuss it with you at your study visit. If you have any questions about the study that you want to ask before joining, please contact the study office.

Other Information About the NCGENES Study

Who Pays for the Study?

NCGENES receives grant money from the National Institutes of Health (NIH), the University of North Carolina Lineberger Comprehensive Cancer Center (LCC) and the University Cancer Research Fund.

How Long Will the Study Last?

Four years. Most of the things you will be asked to do will happen in your first year.

Will I Be Re-contacted?

We will review genetic information once a year (for up to 4 years) but will only re-contact you if new information is found that would affect your medical care.

Can I Stop Being in the Study?

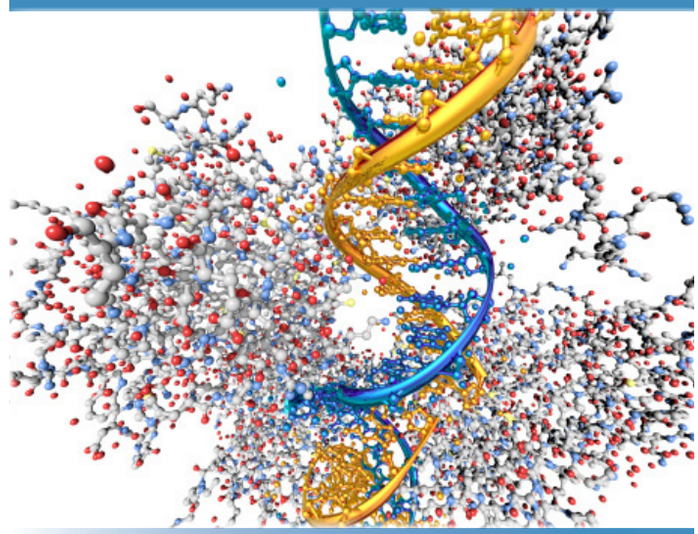
Yes, you can stop being in the study at any time and we will destroy your sample. If you have decided to have your results placed in your UNC Hospitals' Electronic Medical Record, these cannot be removed.

Genetic Medicine Building

UNC Hospitals

University of North Carolina
Room 326 MacNider Hall
Chapel Hill, NC.7599-7240
919-537-3795 ncgenes.org

THE NCGENES STUDY



North Carolina Clinical Genomic Evaluation
by Next-generation Exome Sequencing

NCGENES

North Carolina Clinical Genomic Evaluation by Next-generation Exome Sequencing

What Is NCGENES?

NCGENES is a research project at UNC-Chapel Hill.

We are studying a new kind of genetic test called "Whole Exome Sequencing," or WES.

What Is Whole Exome Sequencing (WES)?

WES is a new genetic test that can find genetic causes of some health problems. The test studies the **exome**, which contains the genetic information used by our cells to make the different proteins in our body. Proteins, along with our environment, affect how our bodies work.

All genes have the same four DNA bases: "A", "C", "T", and "G". The order of these DNA bases in a gene is called the "**sequence**." The DNA sequences tell the cell which protein to make and how to make it.

The DNA sequence of a gene is not exactly the same in everyone. WES reads the sequences of **many genes at the same time** and finds many thousands of differences, called "**variants**".

Some variants that WES finds may give you information about your health, but most will not. Most often we don't know whether or not a variant will have an effect on health. And, WES will not find all the variants that could possibly affect your health

Why Is This Study Being Done?

We want to answer these questions:

How good is WES at finding genetic causes of health problems?
How do people understand and react to the results?

Who Can Join NCGENES?

People whose health concern probably has a genetic cause may be eligible to join.

What Kinds of Health Problems Are Being Studied?

We are studying some types of cancer, some heart problems such as an enlarged heart or an abnormal heart rhythm, some types of vision loss, neurological problems, and birth defects.

How Many Can Join?

Up to 750 people including both adults and children.

If You Join NCGENES, What Will You Be Asked to Do?

1. Come to UNC for 2 study visits; each takes about an hour.

First Visit:
Learn about how WES is used in NCGENES and give a blood sample
Complete an intake form/brief interview; this takes about 15 minutes.

Second Visit: Come back in about 4-6 months to learn what was found.

2. Answer 2-4 telephone surveys; each takes about 45 minutes.

You will be called and asked questions about your NCGENES experiences.

3. Fill out 2 questionnaires; each takes about 20 minutes.

You will answer written questions about your NCGENES experiences.

How Much Blood Is Needed?

4 tubes of blood; ~ 4 teaspoons from adults
and ~2 teaspoons from children

What Will Be Done With the Blood Sample?

Two tubes of blood will be used for WES. The other two tubes will be used by the UNC Hospitals' Molecular Genetics Laboratory (MGL) to confirm the results.

Your sample will be given a unique ID number to help protect your personal health information. All information will be kept in a secure computer database.

How Will You Learn Your WES Results?

We will discuss some types of results with you at your second study visit. You will be given a written report of the results. Some adult participants will be asked to decide if they want us to re-examine their WES to learn other findings. They will be given additional information about making this decision.

Will the Information Learned from WES Be Put Into Your Medical Record?

You will be asked if you want the confirmed results placed in your UNC Hospitals Electronic Medical Record (EMR). If you agree, you will sign a consent form and an official report will be entered. If you do not agree, these results will not be placed in your record.

Does It Cost to Join?

No, there is no charge for the study clinic visits, the blood draw, or the WES.

Will You Be Paid?

Yes, you will receive a $20 check after each survey you complete. You will also get parking vouchers for the clinic visits.

Are There Any Possible Benefits to Joining?

Yes. You will help us understand how to use WES in the future. Some people could learn the genetic cause of their health problem.

Are There Any Possible Risks?

Yes. Having your blood drawn may cause minor bruising or bleeding. Some people might worry if they learn that their health problem has a genetic cause. Some people may also worry about others finding out about their results.

What Will Be Done to Reduce the Risks?

An experienced phlebotomist at UNC Hospitals will draw your blood.

We will help you understand what your results mean for you and your family.

All samples will be coded with a unique ID number. The link between your name and your number will be kept in a secure computer database.

What Happens to the Samples When the Study Ends?

We will destroy the samples if you tell us that you want to stop being in the study. Otherwise, we will continue to study them.

What Happens Next?

Your doctor will let us know whether or not you are eligible to join NCGENES. If you are, someone from the research team will call you. You will have a chance to ask more questions at that time. If you do not wish to be contacted, call the study office to let them know.