Will my insurance company be able to access this information?

Yes, any information in your medical record can be seen by your insurance company. But, if your test was performed as part of a research study, those research results will not be in your medical record and will not be seen by your insurance company. Your genetic counselor can answer any questions you have regarding insurance companies and genetic information.

The Genetic Information Nondiscrimination Act of 2008 (GINA) is a federal law that protects people from genetic discrimination in health insurance and employment. It says you cannot be denied a job or denied health insurance because of your genetic condition. Your genetic counselor will talk to you about exceptions to this law.

Helpful Websites

Iowa Institute of Human Genetics (IIHG)

Information for clinicians, researchers, patients, and students, including research, clinical and educational initiatives, and resources supported by the IIHG

>> www.medicine.uiowa.edu/humangenetics

National Human Genome Research Institute

Information on the Human Genome project, including the latest research, educational materials (for students, teachers, and patients), and an explanation of the Genetic Information Nondiscrimination Act (GINA) and your rights concerning genetic discrimination

>> www.genome.gov

Genetics Home Reference

Information about how genes affect your health: what genes are, how they work, and how they cause disease

>> www.ghr.nlm.nih.gov

Contact

Iowa Institute of Human Genetics, University of Iowa
285 Newton Road, 5296 CBRB, Iowa City, IA 52242
Phone: (319) 335-3688 | Email: iihg@uiowa.edu

University of Iowa Health Care
Iowa Institute of Human Genetics

Updated August 2013

Exome Sequencing:

Information for Families

This booklet is for you and your family to read before meeting with a genetic counselor. Your genetic counselor will explain this information to you and answer questions you may have.

University of Iowa Health Care
Iowa Institute of Human Genetics

What is your exome?

The exome is the part of the genome (all your DNA) that includes all of your exons. Exons are the parts of genes that contain code to build the proteins found in your body. Exons make up about 1 percent of your DNA. Most known disease-causing variants, or DNA changes, are found in the exome.

What is exome sequencing?

In the past it was only possible to screen one gene at a time to find the cause of a disease. This method is still used for many diseases, but it can be costly and time consuming. It is now possible to screen a person's entire exome at once.

Why is my doctor suggesting this test?

Genes are passed down to us from our parents before we are born. Genes have changes (DNA variants) that determine things like our height or hair color. They can also cause disease or increase our risk for disease. By sequencing your DNA, doctors may be able to find a genetic cause for your disease or health condition.

Finding a genetic cause of a disease **may or may not**:

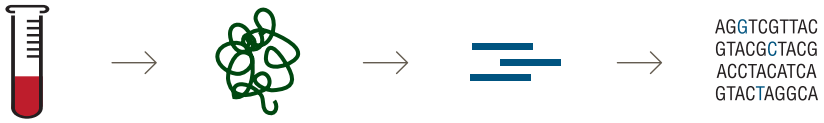
- Tell you the cause for your disease or symptoms
- Help you and your health care provider choose the best treatment for you
- Determine recurrence risks, or the chances that you or your family members will develop a disease

A genetic counselor will talk about the test, possible results, and the meaning of the results with you during the informed consent process.

What will happen to my sample?

We will need a blood sample so we can get your DNA to sequence, or screen.

You may be asked to give another sample of blood if the first sample does not provide enough DNA for the test.



Genetic Testing Possible Results

Types of Findings

1. **Primary Findings:** Changes in your DNA that cause the condition or predispose you to the condition for which you were tested (putting you at higher risk than the general population)
2. **Secondary Findings:** Changes in your DNA that are unrelated to the reason you received this test. These secondary findings could influence your future medical care. To understand more about secondary findings, please talk to your health care provider or genetic counselor.

Types of Results

1. **Positive:** A change in your DNA that is known to cause disease has been found.
2. **Negative:** No changes in the genes that were screened
3. **Variant of uncertain (clinical) significance (VUS):** A change in a gene known to cause disease was found, but the effect of this change is unknown. It could also mean a change in a gene that is not yet known to cause disease has been found. Therefore, the significance of the change is unknown.

How will the results be given to me?

A doctor and/or a genetic counselor will contact you with your results to explain them and answer questions you may have. Your doctor will get recommendations that may or may not include more genetic testing based on the results of your test.

Test Limitations

Your genetic counselor will explain the limitations of exome sequencing to you during your counseling appointment. Sometimes more testing is needed.