

Frequently Asked Questions for Patients

How long does it take to get results?

While there are a number of factors that can affect the time it takes to obtain results, we expect most results will be returned in approximately 2-3 months. Because of the way testing is done, samples obtained on the same day are not necessarily processed at the same time. Therefore, please don't be concerned if your results take longer to be returned than you might have expected, and may come back later than for a sample obtained from another person on the same day. The time it takes to obtain a result is not an indication of whether results are indicative of increased risk of a genetic disorder.

What types of information will I get from the test?

You will receive two different types of information from AGHI. You will receive a disease risk result that will tell you whether any gene changes were found that may increase your risk of developing a certain disease. You will also receive a pharmacogenetic result that will tell you whether any gene changes were found that may affect how your body responds to medicines. You will receive two separate reports, one about disease risk and one about pharmacogenetics. You may receive these reports at different times.

What disease risks can be found?

Your DNA sample will be examined for changes in a set of genes associated with actionable diseases. The word actionable means that you could take a certain action about your medical care to lower the chance of getting the disease.

Most of the genes that are looked at are associated with a genetic form of cardiovascular (heart) disease or the risk of cancer. Cardiovascular diseases include problems with the heart muscle, heart rhythm, and heart arteries, or a high cholesterol level. Cancer risks include breast, ovarian, colon, thyroid, and many other types of cancer. Other genes included in the screening are associated with malignant hyperthermia, an adverse reaction to certain anesthesia, as well as several other rare genetic diseases.

There is also a possibility that through the course of testing, the AGHI team will identify and report a gene change believed to be medically actionable that is not on the current list of genes.

If my genetic testing indicates increased risk of a genetic disease, does that mean I'll definitely get that disease?

No – the genetic test indicates increased risk of disease, but not everyone at increased risk goes on to actually develop the condition. If your test is positive, we will recommend follow-up medical testing to determine whether you are currently showing signs of the condition, and a program for surveillance to detect any signs that might occur as early as possible. A person who tests positive still can live a long life and never develop signs or symptoms.

How much does AGHI testing cost?

Participation in AGHI is free. There is no cost associated with enrolling in the study and receiving genomic screening results. Costs associated with your routine medical care after disclosure of results from AGHI participation are not covered by AGHI.

How will I receive my test results?

There are two ways you may learn your AGHI test results. Your doctor will receive a copy of your results as soon as they are ready. Your doctor's office may reach out to you to discuss your results and what they mean for you. AGHI will also send a copy of your results to you in the mail.

Who do I contact if I have questions about my test results?

If you have questions about your AGHI test results, you can contact your primary care provider. You can also reach out to the AGHI study team at aghi@uab.edu.

What is the chance I will have a genetic disease risk identified?

Studies have shown that about 1-2% of people in the general population have a change in one of the disease risk genes included on the AGHI test. We expect that most patients will receive a negative disease risk result, with no disease-causing gene changes found.

If I am not found to have an increased risk of a genetic disease, does this mean that I can't develop any of the conditions tested?

No – the test being done is referred to as a "screening test." This means that it searches for major, well understood changes in the group of genes being tested. It does not pick up all possible types of genetic changes in these genes, and does not test all genes associated with genetic conditions, including those associated with heart disease or cancer.

IF YOU HAVE A KNOWN RISK FOR A GENETIC CONDITION, FOR EXAMPLE DUE TO PERSONAL OR FAMILY HISTORY OF DISEASE, AGHI SHOULD NOT BE USED AS A SUBSTITUTE FOR ROUTINE CLINICAL GENETIC TESTING. CLINICAL GENETIC TESTING IS MORE THOROUGH THAN THE TESTING PROVIDED BY AGHI.

What is the chance I will have a pharmacogenetic variant identified?

Studies have shown that over 95% of people have a genetic change that will affect how they respond to medications. We expect that most patients will have at least one pharmacogenetic change which may affect your current or future medications.

What should I do if I find out I have a pharmacogenetic change that affects a medication I am taking?

Do NOT stop taking or change the dose of any of your medications based on your test result. Many factors (other than your genes) play a role in how you respond to medications. Even if you have a genetic change that affects a medication, in many cases you can still take the medication. Sometimes you may need a higher or lower dose, or your doctor may want to monitor you more closely. That is why it is important not to stop taking any medications based on this report. YOUR DOCTOR CAN WEIGH ALL THESE FACTORS TO TAILOR YOUR MEDICATIONS.

Where will my test results be stored? Will anyone outside my doctors access my private genetic information?

Your genetic disease risk results and pharmacogenomic results will be sent to both you AND your medical provider, so that you and your doctor can discuss the results. These genetic results will be stored in your medical record, but no identifiable genetic information (that is, your genetic results stored under your name, your medical record number, or any piece of information that identifies you) will be disclosed to people outside your healthcare team.

If you choose to participate in the Biobank and future research, after your results are returned to you, all identifiers are stripped off of your data. So, genetic or pharmacogenetic information that originates from you can be used for various research purposes, but your identity will be kept confidential and will not be known to outside researchers without your consent.

Should I say yes to biobank?

If you say yes to biobank, your blood sample will be coded (that is, your name will be removed from your blood sample and the sample will be given a unique code) and then stored in the biobank. Stored blood samples in the biobank will enable researchers to conduct future scientific research on various health conditions that may benefit you as well as many others who have genetic features similar to you. As such, the AGHI research team regards the biobank as a key research tool for future medical breakthroughs.

Respecting your decisions about participation in research is one of the most important ethical principles the AGHI team cherishes. The decision about whether or not to have your blood sample stored in the AGHI Biobank is a personal one, and will not affect your ability to participate in other aspects of this study.

There are some potential factors that you may consider in making this decision such as concerns about the privacy of your genetic information and the opportunity to contribute to medical / genetic research.

To learn more about the AGHI, please contact us:

aghi@uab.edu | aghi.org

Is pharmacogenetic testing useful even if I am not currently taking prescription medications?

Yes. Pharmacogenetic information can help your doctors choose medicines for you in the future. You should keep a copy of your results and share them with any doctors you see, now and in the future.

If my genetic testing is positive, does this mean that other members of my family also might be at risk?

Yes, other family members might possibly also carry the genetic variant, and therefore could be at risk. This might include your parents, siblings, children, and perhaps other more distant relatives. When the result is returned to you, the genetic counselor will speak with you about options to have other family members tested, though this testing would not be done free-of-charge through AGHI.

Are there any laws protecting me from discrimination based on my genetic information?

There are some protections available to prevent genetic discrimination, but they have important limitations and may not prevent discrimination in all settings. The Genetic Information Nondiscrimination Act (GINA) is a federal law that was passed in 2008 to provide protection from discrimination in health insurance coverage or employment based on genetic information.

First, GINA makes it against the law for a health insurance company to use a genetic test result or family health history as a reason to deny you health insurance coverage or to decide how much you are going to pay. Second, GINA makes it against the law for an employer to use a genetic test result or family health history to make choices regarding your employment.

There are limitations to the protections GINA provides, however. GINA's protections for insurance only apply to health insurance. They do not apply to life, long-term care, or disability insurance. Employment protections of GINA do not apply to small businesses/employers with less than 15 employees, or to those in the US Military or employees of the federal government. Signed in to law in 2000, Executive order 13145 protects federal employees from genetic discrimination in employment.

If you have concerns about genetic discrimination, you should discuss these with a genetic counselor or other healthcare provider before undergoing genetic testing, including the genetic testing offered by the AGHI.

What are possible reasons the AGHI research team would re-contact me?

The AGHI research team may recontact you for a number of reasons. We may recontact you to inform you of significant new findings about your health condition or ask you for feedback about your experience enrolling in AGHI. We may also contact you to request fresh blood samples or to inform you of similar research studies that may be of interest to you. You do not have to give us more blood or medical information in the future. Any future blood samples you give to this or any other study will be your decision.