

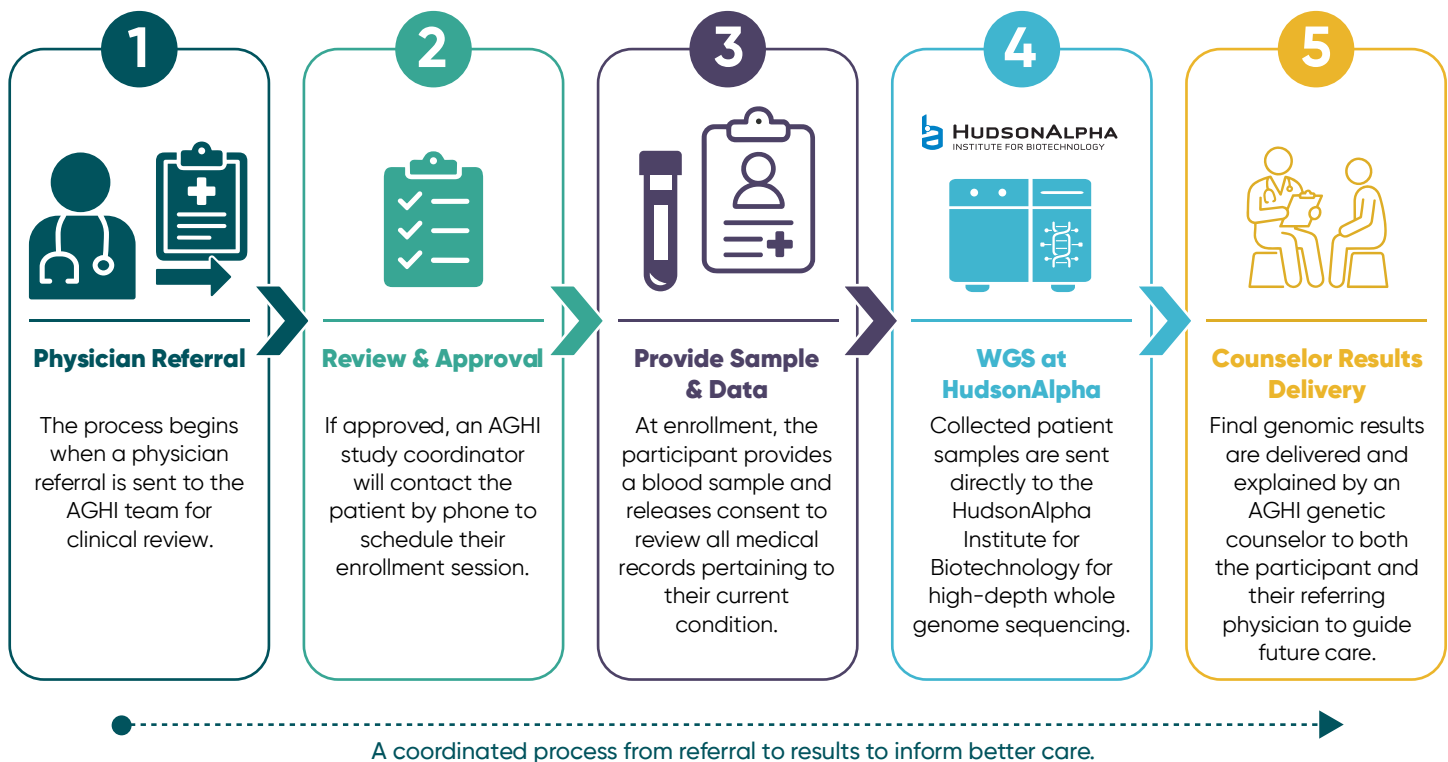


The Alabama Genomic Health Initiative (AGHI) is a statewide, state-funded program that provides free genomic testing, interpretation, and counseling to Alabama residents.

## AGHI Supports Genomic Sequencing (GS) For Patients With Undiagnosed Diseases

**An Undiagnosed Disease** is a medical condition without a known cause or formal medical name, even after extensive testing and consultations with various specialists. These cases can be challenging because symptoms may be atypical, extremely rare, or the disease itself may not have been previously encountered.

**Genome Sequencing** is a laboratory process that determines the complete DNA sequence of a person's genome, including all its genes. This means it identifies the order of all the "letters" (A, T, C, and G) in a person's DNA, providing a comprehensive view of a person's genetic makeup. GS can lead to the identification of genetic cause for a person's condition.



The process begins with a **comprehensive medical record review**. Once doctors believe a condition is genetic, the family attends a specialized **enrollment** visit and **provides consent and a blood sample**. Next the isolated DNA is fragmented into millions of small pieces, tagged with unique barcodes, and loaded into high-throughput Next-Generation Sequencing machines that read the entire 3-billion-letter code, i.e. **the genome sequence**.

Finally, bioinformatics software aligns these fragments to a standard human genome map to identify variants. This is evaluated by **clinical geneticists** against the patient's symptoms, and the **final report** is explained to the patient by a **genetic counselor**.

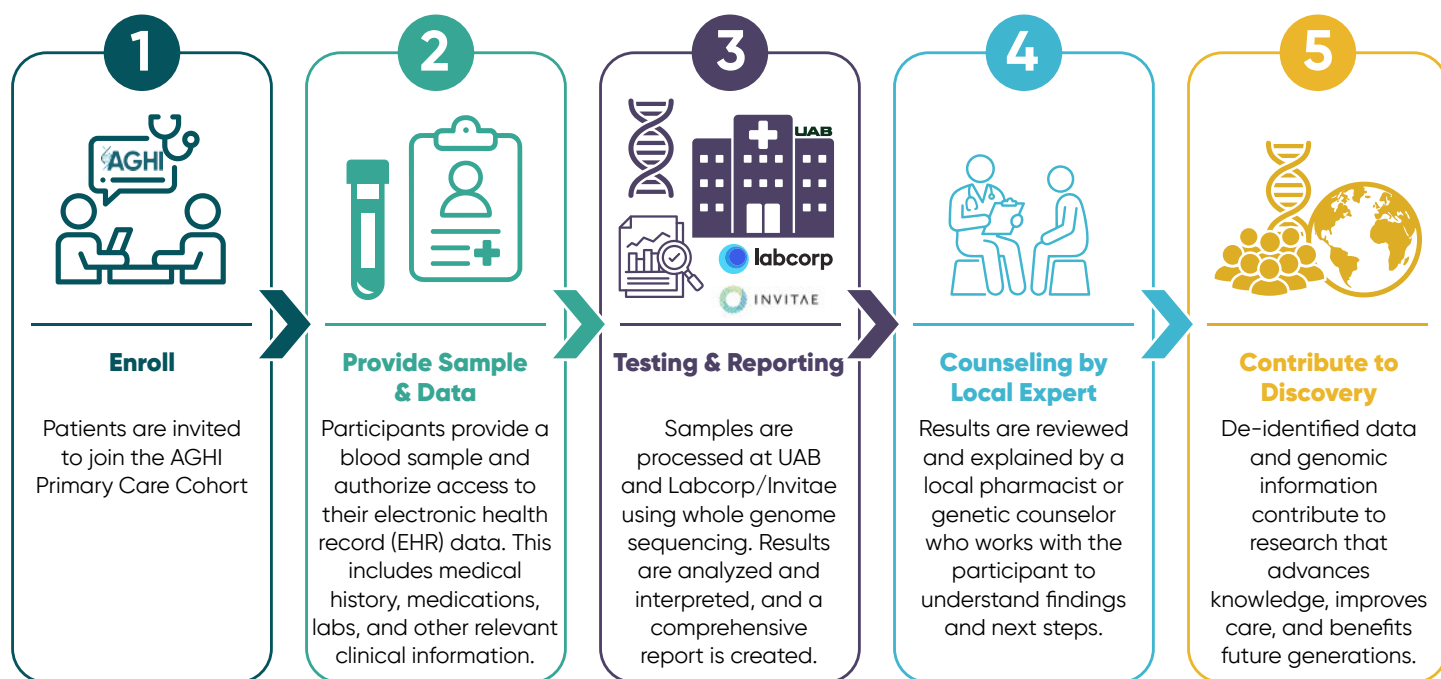


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### AGHI Provides Genomic Screening To Uncover Hidden Risk For Common Chronic Conditions And Pharmacogenomics Testing To Tailor Prescriptions.

Genetic variation contributes to risk of common diseases (like breast cancer) and influences how individuals respond to medications (like statins for lowering cholesterol). Identifying these genetic differences before disease develops or before medications are prescribed can help:

- Detect inherited risks for cancers, cardiovascular disease, and other actionable conditions
- Enable earlier screening and preventive care
- Support personalized medication selection through pharmacogenomics (PGx)
- Reduce adverse drug reactions and improve treatment effectiveness
- Contribute to genomic database to advance research and discoveries



Your participation today helps advance precision medicine for tomorrow.

The process begins with enrollment of participants interested in learning about their health. During the **enrollment visit**, a research coordinator obtains **consent** and collects a **blood sample**.

The DNA from the sample is tested to understand how the individual's unique genetic code affects

- 1. Disease risk** for high-risk genetic conditions such as hereditary breast and ovarian cancer predisposition (BRCA genes) or hidden, dangerous heart conditions like cardiomyopathy.
- 2. Medication response** by assessing how your specific genes affect your response to medications. It focuses heavily on how your liver enzymes break down (metabolize) various drugs.

This is evaluated by **clinical geneticists**, and the **final report** is explained to the patient by a **genetic counselor** (disease risk) or **pharmacist** (medication response).