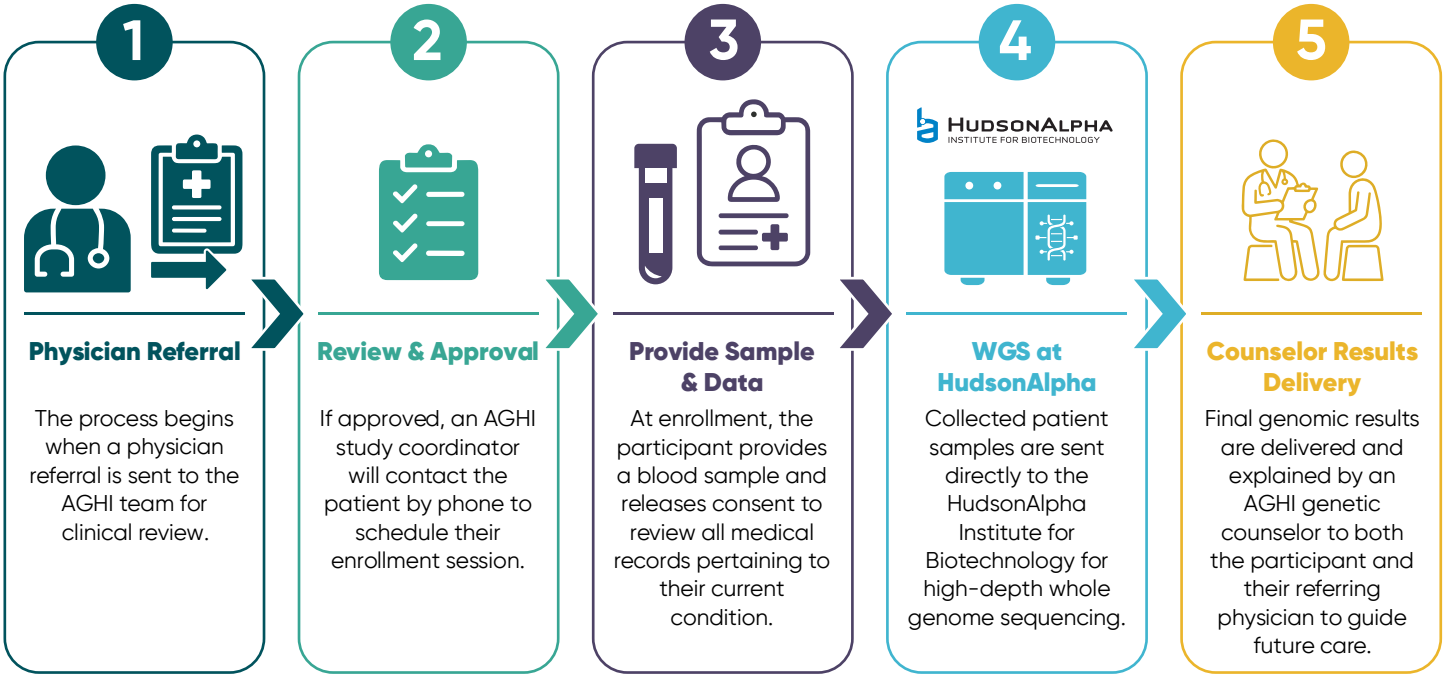
# AGHI Alabama Genomic Health Initiative

**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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