AGHI Alabama Genomic Health Initiative

Annual Report 2025-2026

11,000+ Alabamians. One Extraordinary Milestone

Delivering genomic insights that improve lives, inform care, and shape the future of health in Alabama.

AGHI Alabama Genomic Health Initiative

We are pleased to present the 2025–2026 Annual Report of the Alabama Genomic Health Initiative (AGHI), a transformative partnership between the University of Alabama at Birmingham and HudsonAlpha Institute for Biotechnology.

Investment in genomics is an investment in the future of healthcare. This report highlights the achievements made possible through your support and outlines the next steps in our shared journey to improve the health of Alabamians.

AGHI has expanded access to genomic medicine and advanced our understanding and responsible use of genomic information in healthcare. Together, we have impacted the lives of more than 11,000 Alabamians by:

- **Providing genomic screening to identify hidden risks for chronic disease**
- **Using pharmacogenomics to help tailor medications to an individual's genetic profile**
- **Delivering genome sequencing to help families find answers to undiagnosed diseases**
- **Generating evidence that informs the integration of genomic medicine into clinical care**
- **Advancing research discoveries while training the next generation of scientists and healthcare professionals**

With your support, AGHI has emerged as a national leader in genomic medicine and precision health. Our investigators play key roles in landmark national initiatives focused on population genomic screening, rare disease diagnosis, implementation science, and precision medicine, including All of Us, Electronic Medical Records and Genomics (eMERGE), Implementing Genomics In Practice (IGNITE), the Undiagnosed Diseases Network, the Population Genomics Screening Network, and the Southeast Genomic Medicine eConsult Service.

What began as a bold vision - to improve access to, understanding of, and use of genomics to address Alabama's healthcare needs - has evolved into a nationally recognized model for integrating genomic medicine into patient care.

The work highlighted in this report demonstrates the power of collaboration among researchers, healthcare providers, policymakers, and the communities we serve. Together, we are advancing genomic medicine, improving health outcomes, reducing the burden of disease, and strengthening Alabama's leadership in precision health.

Thank you for your continued partnership, commitment, and investment in a healthier future for Alabama.

Sincerely,



Nita Limdi, PharmD, PhD, MSPH, FAHA, FACTS

Ray L Watts Heersink Endowed Chair in Neurology
Associate Dean for Genomic Medicine
Director, UAB-HAIB Genomic Medicine Training Program
Director Alabama Genomic Health Initiative



Greg Cooper, PhD

Faculty Investigator,
HudsonAlpha Institute for Biotechnology
Co-director Genomic Medicine training Program
Co-director Alabama Genomic Health Initiative



Matthew Might, PhD

Director, UAB Hugh Kaul Precision Medicine Institute
Co-Director, Alabama Genomic Health Initiative

Table of Contents

Alabama Genomic Health Initiative: Executive Summary	5 - 6
Genomic Screening Uncovers Hidden Risk For Chronic Conditions	8 - 11
Pharmacogenomics: Tailoring Prescriptions For You	12 - 15
Genome Sequencing Brings Answers To Families With Undiagnosed Diseases	16 - 18
Glossary of Terms	19
A Guide to AGHI Participation and Referral	20 - 23
Shaping the Future of Genomic Medicine Together, Generating Insights for Clinical Integration of Genomic Medicine	24 - 26
Fueling the Future: Advancing Genomic Research and Empowering the Next Generation of Scientists	28 - 33
Publications	34 - 35
The Team and Timeline	36 - 41
How Alabama is Leading the Way in Genomic Medicine	42
Skating to Where the Puck Is Going in Genomics: Charting Our Path Forward	43

ALABAMA GENOMIC HEALTH INITIATIVE
Executive Summary

A groundbreaking, state-funded partnership between University of Alabama at Birmingham and the HudsonAlpha Institute for Biotechnology, the Alabama Genomic Health Initiative (AGHI) delivers free, advanced genetic testing across all 67 counties to eliminate healthcare guesswork. By mapping unique DNA profiles, AGHI empowers doctors to uncover hidden risks for catastrophic illnesses like cancer and heart disease before symptoms appear, personalize prescriptions, and solve rare medical mysteries.

This vital state investment anchors a cutting-edge clinical research database and trains Alabama's medical workforce in genomics and precision health. Having surpassed 11,000 participants, the initiative has secured significant matching federal grants - cementing Alabama's emergence as a dominant national leader in genomic medicine and precision population health.

Goals	Achievements	Impact
Access to genomic medicine to Improve Lives	**11,000 + have received genomic insights from all 67 counties**	
	9,299 received genetic risk information for chronic conditions	For **9%**, testing uncovered hidden risks for chronic disease, guided proactive care
	2,768 received pharmacogenomics results	For **23%**, testing personalized current medications. Results can personalize prescriptions throughout life for all
	628 patients and **856** family members received genome sequencing	For **22%**, sequencing provided a diagnosis and a plan for healthcare
Drive Innovation	**Advancing discoveries, Training the next generation** of clinician scientists and researchers	Supporting **research for chronic conditions** including heart failure, gout, multiple sclerosis and Alzheimer's disease Building **polygenic risk scores** for multiple chronic conditions and medication response
Strengthen Alabama	Building a **clinical-genomic database** for future research and clinical trials	**11,000+** participants, **41,000+** biological specimens. **90%** consented to future research. Of these, **99%** also have data from genome-wide testing.
	Recognition as a **national leader in genomic medicine** and precision population health	Leadership in All of Us Research Program, the Electronic Medical Records and Genomics (eMERGE) Network, Implementing Genomics in Practice (IGNITE), Undiagnosed Diseases Network (UDN), Population Genomic Screening Network and the Regional Genomic Medicine eConsult Network

Core Values: Respect - Transparency - Confidentiality - Fairness

AGHI Alabama Genomic Health Initiative

The Alabama Genomic Health Initiative (AGHI) is our statewide precision medicine program led by University of Alabama at Birmingham and HudsonAlpha Institute for Biotechnology. It uses genomic testing to identify genetic risks for certain cancers, cardiovascular conditions, metabolic disorders, rare diseases, and medication-response differences (pharmacogenomics). The program also supports research aimed at improving healthcare for Alabamians.



For the Participant

AGHI helps people learn whether they carry genetic changes that may increase their risk for serious but potentially preventable or treatable conditions.

Knowing this information early can allow:

- Earlier screening for cancer
- Prevention of disease before symptoms develop
- More personalized treatment plans
- Better medication selection and dosing through pharmacogenomics (PGx)
- Answers for families with undiagnosed genetic conditions
- More informed family planning decisions



For the Provider

AGHI brings genomic medicine into routine clinical care and helps providers identify patients at increased risk for actionable genetic conditions before disease develops. It supports accurate diagnoses, preventive medicine, and precision prescribing.

Providers receive reports that may include:

- Pathogenic or likely pathogenic variants associated with actionable disease risk
- Pharmacogenomic findings affecting medication selection or dosing
- Clinical recommendations and referrals when appropriate
- Answers for some families with undiagnosed genetic conditions



For the Researcher

AGHI has created one of the largest statewide genomic resources in the United States. The program combines genomic data, clinical information, and participant consent for future research, enabling studies of disease risk, health outcomes, pharmacogenomics, and precision medicine implementation.

What does AGHI offer researchers?

- Large genomic datasets from participants across Alabama
- Associated clinical and health-history information
- Bio-banked samples (where consented)
- Opportunities for genotype-phenotype research
- Pharmacogenomic and population-health research resources

AGHI is our statewide genomic medicine initiative that provides genetic testing, counseling, and pharmacogenomic guidance to patients, supports precision healthcare for providers, and creates a valuable genomic research resource that advances the understanding and prevention of disease.



Genomic Screening Uncovers Hidden Risk For Chronic Conditions





9,299 Participants Received Genetic Risk Information for Common Conditions. **9%** of those results were actionable

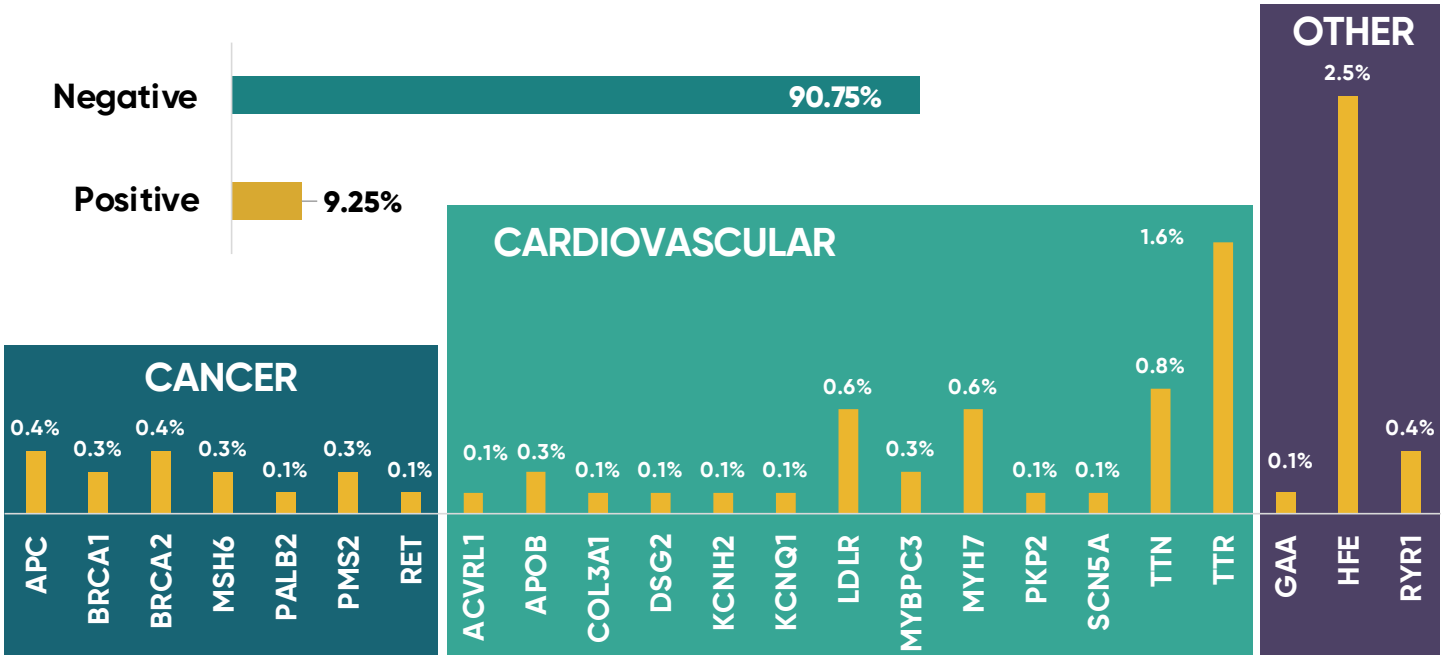
Genomic Screening Uncovers Hidden Risk For Chronic Conditions

In traditional clinical care, genetic testing for conditions such as breast cancer and heart disease is typically prompted by a personal diagnosis or a strong family history of disease.

In contrast, AGHI evaluates unselected individuals, enabling the identification of hereditary risks before symptoms arise. Through population screening, anyone can access potentially life-changing genetic information regardless of their personal or family history. By removing barriers to testing and offering genetic screening first, counseling later, AGHI identifies health risks in people who may never have otherwise qualified for or pursued genetic testing.

Since its inception, AGHI has delivered medically actionable insights by analyzing participants' genomes to identify genetic disease risks, including conditions such as breast cancer, Lynch syndrome, and familial hypercholesterolemia and providing genetic counseling to all participants.

During 2025–2026, 9.25% of AGHI participants were found to have a high-risk genomic variant.



These findings have important clinical implications, providing participants and their healthcare providers with information that can guide preventive and therapeutic decisions. Identification of a pathogenic variant enables evidence-based interventions to reduce disease risk, including enhanced surveillance, risk-reducing surgery for certain cancer syndromes, and preventive cardiovascular measures such as statin therapy and early imaging. Early detection and intervention can substantially improve the health of individuals with genetic risk.

Importantly, it is estimated that more than 90% of individuals who carry pathogenic variants in these highly actionable genes are unaware of their risk because they do not have a clear family history. By offering population-based genomic screening, AGHI helps uncover these otherwise hidden risks, creating opportunities for earlier intervention and improved health outcomes.

Answers in the Genes: Turning Uncertainty Into Clarity

Participants receiving a pathogenic finding meet with a certified genetic counselor who explains the results, answers questions, and shares relevant information with the patient's provider to support ongoing care.
By removing barriers to testing and offering genetic screening first, counseling later, AGHI identifies health risks in people who may never have otherwise qualified for or pursued genetic testing. This approach fundamentally changes the genetic counseling experience. For many participants, the phone call delivering a positive result is their first interaction with a genetic counselor. They enrolled out of curiosity or a desire to contribute to research and do not expect to receive a positive result.

In a single conversation, curiosity becomes reality: The reactions are often immediate and profound: shock, confusion, and concern. A genetic counselors (GC) role is to help participants move through those emotions toward understanding and action. Shock is met with empathy. Confusion is met with information. Concern is met with support and empowerment. What makes this work so meaningful is what happens next. Armed with new knowledge, participants can take steps to protect their health - pursuing preventive care, increasing cancer surveillance, lowering cardiovascular risk, and sharing potentially lifesaving information with relatives who may also be at risk. A single genetic result can influence not only one person's future, but the health decisions of an entire family.

Uncertainty gives way to clarity: For some participants, the impact is even more personal. Rather than revealing a future risk, their result finally explains a condition they have already been living with for years. A young man who has done everything right yet struggles with high cholesterol learns his genetics are responsible. A woman whose muscle weakness forced her to give up activities she loves receives a diagnosis that explains her symptoms and opens the door to treatment. For these individuals, genetic information provides more than medical guidance. It brings validation, relief, and hope. Longstanding questions are answered. Feelings of guilt and self-blame are replaced with understanding.

One story stands out as an example of how AGHI provided answers for symptoms a patient was currently experiencing and informed him about risks for potential symptoms that could cause damage if left untreated. This patient had a long history of chronic pain. Not knowing the cause was psychologically burdensome for the patient and made it difficult for his providers to know how to treat him.

The patient's AGHI results revealed he was positive for two genetic conditions-one that causes increased cholesterol levels and another that can cause chronic pain and organ damage. Both conditions are potentially treatable. Since discovering his results, the patient has attended specialist appointments to learn more about his conditions and has begun treatment for both. Upon investigation, his cholesterol was elevated, but it is now under control with medication. He still struggles with pain, but is receiving treatment, and is relieved by finally knowing the origin of something that has been affecting him for so long. For this patient AGHI provided long-sought-after answers, and answers he never knew he needed.

"Returning results through AGHI has given me the privilege of being the first stop on participants' personalized health journeys. Whether revealing a previously unknown health risk or providing an explanation for years of unexplained symptoms, these results have the power to change lives. They equip participants, families, and healthcare providers with information that can guide decisions, improve outcomes, and shape the future" says Denton.

Every conversation reminds GCs that genetic information is more than data. It is a doorway to understanding, empowerment, and a future shaped by newfound clarity.



Jessica Denton, PhD, MS, CGC (UAB)



Nathaniel Robin, MD (UAB)

AGHI Genes Tested and the Associated Medical Conditions

Medical Condition	Associated Genetic Difference(s)
Cancers	
Familial adenomatous polyposis	APC
Familial medullary thyroid cancer, multiple endocrine neoplasia type 2	RET
Hereditary breast and/or ovarian cancer	BRCA1, BRCA2, PALB2
Hereditary paraganglioma-pheochromocytoma syndrome	MAX, SDHAF2, SDHB, SDHC, SDHD, TMEM127
Juvenile polyposis syndrome	BMPR1A, SMAD4
Li-Fraumeni syndrome	TP53
Lynch syndrome	MLH1, MSH2, MSH6, PMS2
Multiple endocrine neoplasia type 1	MEN1
MUTYH-associated polyposis	MUTYH
Neurofibromatosis type 2	NF2
Peutz-Jeghers syndrome	STK11
PTEN hamartoma tumor syndrome	PTEN
Retinoblastoma	RB1
Tuberous sclerosis complex	TSC1, TSC2
Von Hippel-Lindau syndrome	VHL
WT1-related Wilms tumor	WT1

Medical Condition	Associated Genetic Difference(s)
Cardiovascular Conditions	
Aortopathies (includes Marfan syndrome; Loeys-Dietz syndrome, type 1, 2, and 3; Familial thoracic aneurysms and dissections	ACTA2, FBN1, MYH11, SMAD3, TGFBR1, TGFBR2
Arrhythmogenic right ventricular cardiomyopathy	DSC2, DSG2, DSP, PKP2, TMEM43
Catecholaminergic polymorphic ventricular tachycardia (CPVT)	CASQ2, RYR2, TRDN
Hereditary paraganglioma-pheochromocytoma syndrome	MAX, SDHAF2, SDHB, SDHC, SDHD, TMEM127
Dilated cardiomyopathy	FLNC, LMNA, TTN, TNNT2
Ehlers-Danlos syndrome, vascular type	COL3A1
Lynch syndrome	MLH1, MSH2, MSH6, PMS2
Familial hypercholesterolemia	APOB, LDLR, PCSK9
Romano-Ward long-QT syndrome types 1, 2, 3, 14, 15 and 16, Brugada syndrome	KCNQ1, KCNH2, SCN5A, CALM1, CALM2, CALM3
Hypertrophic cardiomyopathy	ACTC1, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, TNNI3, TPM1, TNNT2, GLA, LMNA, FLNC, TTN, BAG3, DES, RBM20, TNNC1

Medical Condition	Associated Genetic Difference(s)
Metabolism Conditions	
Biotinidase deficiency	BTD
Fabry disease	GLA
Ornithine transcarbamylase deficiency	OTC
Pompe disease	GAA
Hereditary hemochromatosis (HFE p.C282Y homozygotes only)	HFE
Hereditary hemorrhagic telangiectasia	ACVRL1, ENG
Malignant hyperthermia	CACNA1S , RYR1
Maturity-onset diabetes of the young	HNF1A
RPE65-related retinopathy	RPE65
Wilson Disease	ATP7B
Hereditary TTR amyloidosis	TTR

Tailoring Prescriptions Through Pharmacogenomics



2,768 participants received pharmacogenomics results. Over **99%** had at least one genetic variant affecting medication response. **23%** were currently taking a medication impacted by their PGx results. Results can help guide safer and more effective medication use throughout life

Tailoring Prescription Through Pharmacogenomics

Pharmacogenomics is the study of how your unique genetic makeup influences your body's response to medications. **It combines pharmacology and genomics** to ensure you receive the safest and most effective drugs at the correct doses, moving healthcare away from a "one-size-fits-all" approach.

Participants enrolled in the Alabama Genomic Health Initiative (AGHI) receive valuable insights into how their genetic makeup may influence medication response. Through ongoing efforts to expand and integrate pharmacogenomics into clinical care, AGHI Pharmacogenomics Pharmacists have developed comprehensive educational resources, optimized workflows for incorporating pharmacogenomic results into the electronic medical record (EMR), and created clear, user-friendly summary reports for both participants and providers.

Most Common Medications Affected by PGx

Medication Class	Example Medications	Percent of General Population with Actionable PGx Findings
Antidepressants	• Elavil (amitriptyline) • Celexa (citalopram) • Lexapro (escitalopram)	>60%
Cardiovascular (cholesterol lowering)	• Zocor (simvastatin) • Crestor (rosuvastatin) • Lipitor (atorvastatin) • Pravachol (pravastatin)	(11-36%)
Cardiovascular (antiplatelet)	• Plavix (clopidogrel)	(20-50%)
Pain (NSAIDs)	• Advil (ibuprofen) • Celebrex (celecoxib) • Mobic (meloxicam)	~3%

All example medications are Top 300 medications prescribed in the United States***

Impact of Pharmacogenomics (PGx) on Prescription Drug Efficacy

Understanding how an individual responds to medications is essential for delivering personalized and effective healthcare. Pharmacogenomic (PGx) testing identifies genetic variations that influence drug metabolism, efficacy, and safety, enabling clinicians to optimize medication selection and dosing. A substantial proportion of patients carry actionable genetic variants that may affect their response to commonly prescribed medications, particularly antidepressants and cardiovascular therapies, highlighting the significant potential of PGx-guided prescribing to improve patient outcomes.

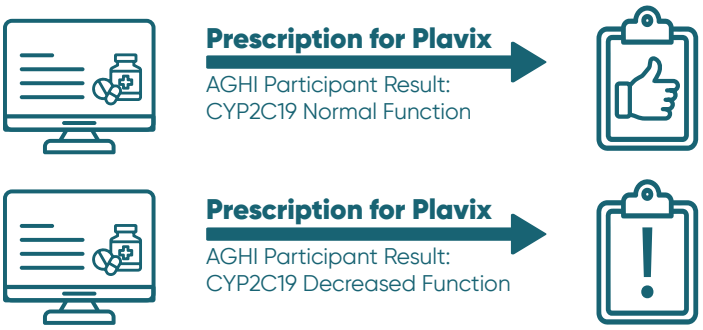
As illustrated in the figure, the prevalence of actionable PGx findings varies by medication class, with more than 60% of individuals having actionable results for certain antidepressants. For other medications, such as clopidogrel (Plavix), the frequency of actionable variants differs considerably across ancestral populations. Approximately 20–30% of individuals of European or African ancestry carry reduced-function variants affecting clopidogrel metabolism, while the prevalence may approach 50% among individuals of Asian ancestry. Translating these genetic insights into clinical practice remains a key strategic priority. We continue to refine our implementation processes by incorporating emerging evidence and best practices from leading scientific organizations and established pharmacogenomics programs. By providing clinicians with actionable genetic information at the point of care, we aim to support more informed prescribing decisions, reduce adverse drug reactions, and increase the likelihood that patients receive the safest and most effective therapy from the start.

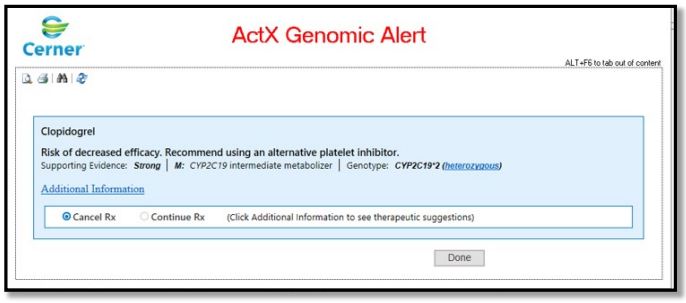
Bringing Pharmacogenomics to the Point of Care

Pharmacogenomic results are returned to both providers and participants to support informed, personalized medication decisions—both now and in the future. As part of this process, AGHI Pharmacogenomics Pharmacists are available for consultation, ensuring that results are well understood and effectively applied in clinical care.



Through a clinical decision support system called ActX, providers will receive targeted alerts when a prescribed medication is impacted by a patient's genetic profile, along with clear recommendations for action. Importantly, alerts are designed to be selective—only triggering when clinically relevant—to minimize alert fatigue. For medications not affected by genetic results, no alert is generated.





In addition to these real-time alerts, ActX provides a proactive review tool that allows clinicians to explore medication options based on a patient's genetic profile. For instance, when considering antidepressant therapy, providers can view a list of medications alongside any gene–drug interactions and associated guidance prior to prescribing.

By transitioning from static PDF reports to integrated, workflow-driven decision support tools, the AGHI Pharmacogenomics team increased the clinical adoption and impact of pharmacogenomics, particularly within family medicine and other frontline care settings. Pharmacogenomics Pharmacists remain available to support providers, ensuring effective interpretation and application of these results in patient care.

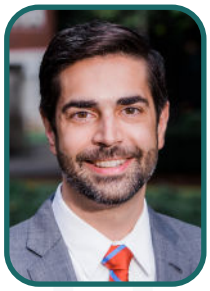
Expanding Participant Engagement Through Direct Pharmacogenomic Consultation

In the coming year, AGHI Pharmacogenomics Pharmacists will enhance participant engagement by introducing proactive, pharmacistled outreach for individuals with actionable pharmacogenomic (PGx) results. Participants whose genetic findings have meaningful implications for medication therapy will be contacted by a pharmacist to review and discuss their results.

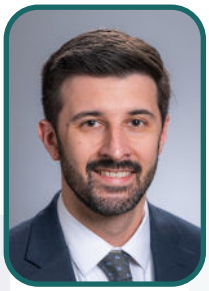


This personalized approach is designed to ensure that participants clearly understand how their genetic profile may impact current or future medication use. During these consultations, pharmacists will provide tailored education, explain potential risks or considerations, and answer questions to support informed decision-making.

By moving beyond written reports to direct communication, AGHI aims to improve comprehension, increase participant confidence, and promote the safe and effective use of medications. This initiative also strengthens the connection between participants and the clinical team.



Blake Goff, PharmD



Josh Cortopassi, PharmD



Brittney Davis, PharmD



Nita Limdi, PharmD, PhD, MSPH, FAHA, FACTS

Pharmacogenomic Testing

Pharmacogenomic testing may be used to help predict:

- Which drug may work best for you
- The proper dosage
- Your risk for negative side effects

Currently, pharmacogenomic testing is done for genes that affect certain types of drugs, such as medications for treating depression, pain, and heart disease. It will be expanded to other genes as researchers learn more about how genomic differences affect the way medications work in the human body.

A person's genes may determine what type of response they have to certain medications.



Gene	Common Drugs Metabolized
CYP2C19	citalopram, escitalopram, pantoprazole, esomeprazole, omeprazole, clopidogrel
CYP2C9	warfarin, ibuprofen, celecoxib, meloxicam
CYP2D6	amitriptyline, nortriptyline, tramadol, codeine, atomoxetine
CYP3A5	tacrolimus
CYP4F2	warfarin
CYP2B6	efavirenz
SLCO1B1	simvastatin, atorvastatin, rosuvatatin
DPYD	capecitabine, fluorouracil
NUDT15	azathioprine, mercaptopurine, thioguanine
TPMT	azathioprine, mercaptopurine, thioguanine
UGT1A1	atazanavir, irinotecan
VKORC1	warfarin

** Not a comprehensive list*



Genome Sequencing Provides Answers for Families with Undiagnosed Diseases





628 patients and **856** family members received genome sequencing; **22%** had a genetic basis identified

Genome Sequencing Provides Answers for Families 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. As one can imagine, this places a profound physical, emotional, and financial toll on patients and their families and healthcare systems.

Genomics has been a valuable tool in shortening the "diagnostic odyssey" for rare diseases. AGHI has supported the development, advancement and deployment of state-of-the-art "genome sequencing" technologies to achieve this goal and combine those technologies with research and discovery projects to constantly push the boundaries of rare disease knowledge.

To look at the entire genome, instead of testing one gene at a time, scientists use Whole-Genome Sequencing (WGS) to scan thousands of genes simultaneously. Short-read sequencing breaks DNA into tiny pieces (50–300 base pairs) to read them with high precision and low cost. Long-read sequencing spans tens of thousands of bases, offering a panoramic view that easily resolves complex genomic structures.

Since 2017, helping patients and families with undiagnosed diseases has been a major goal. We have applied "genome sequencing" to 628 participants affected by rare or undiagnosed disease. We have also tested 856 family members including parents and siblings to accelerate the search for answers.

Over 22% of these participants have a genetic finding that led to a clinical diagnosis ending their "diagnostic odyssey". Receiving a definitive genetic diagnosis ends the psychological toll of uncertainty and provides immediate clinical benefits: It enables treatment access and enrollment in targeted therapies, clinical trials, or disease-specific registries. Importantly it halts unnecessary, often invasive, and costly medical procedures.

One example is the discovery of genetic variation in an AGHI participant affecting the gene ZMYM3. This gene provides instructions for making a protein that acts like a "cellular switch" to "turn off" specific genes and play an important role in repairing broken DNA and shaping cells.

Through collaboration with other testing labs, we identified a total of 27 people with a neurodevelopmental condition that have genetic variation in this gene. For the first time, work led by AGHI investigators showed that ZMYM3 underlies a rare disease. This knowledge can now be used to diagnose people into the future who previously would have been left undiagnosed.

AGHI scientists and investigators have identified 143 other genes for additional research; 10 of these have been shown to cause a rare genetic disease, pushing the boundaries of genetic knowledge.

Another 27% of participants receive reports with uncertain findings or Variant of Uncertain Significance (VUS). It means that a genetic test has found a change in your DNA (a variant or mutation), but there isn't enough scientific evidence right now to determine whether it is harmful and causes disease, or is simply a harmless, natural variation. AGHI scientists and geneticists review these findings and update results and patient reports as knowledge advances. AGHI scientists also lead research projects on VUSs to discover new disease genes to improve diagnostic yield in the future.

For rare disease patients that receive genetic test results, even if there is no immediate cure, obtaining a definitive diagnosis is highly valuable. It allows patients and doctors to better manage symptoms, understand potential disease progression, plan, and connect with other families facing similar challenge.

Innovation in genomic technology to increase diagnostic yield and help additional AGHI participants
AGHI scientists looked at 96 people with rare diseases who were strongly suspected of having a genetic cause, but whose earlier genetic testing using standard short-read DNA sequencing had found nothing useful. AGHI investigators were among the first to use and show that long-read genome sequencing provides additional diagnostic value. In this group, an additional 7% of patients, that standard sequencing missed, received a genetic diagnosis. We expect the usefulness of long-read sequencing to increase further as the technology, and interpretation methods continue to improve.

AGHI is also helping advance the use of long-read genome sequencing by generating data and ensuring accurate interpretation of genetic variation. To support this effort, AGHI scientists helped establish CoLoRSdb (the Consortium of Long Read Sequencing Database), a global collaboration that brings together long-read sequencing data from researchers around the world. By sharing data and expertise, this initiative is creating a resource far larger and more powerful than any single institution could build alone, accelerating discoveries and improving the ability to diagnose rare genetic conditions.

Innovation in genomic assessment: DNA Methylation
AGHI researchers are exploring a variety of other ways to improve the power of genetics to diagnose rare disease. One example is through the analysis of DNA "methylation". DNA methylation is an essential process where small chemical tags (methyl groups) are added to DNA. This modifies gene activity without changing the underlying DNA sequence. It acts like a dimmer switch, telling genes exactly when to turn on or off.

Advances in sequencing technology now allow researchers to analyze methylation patterns alongside genetic data, providing an additional layer of information that can reveal disease-causing changes that might otherwise go undetected. This approach has already contributed to several rare disease diagnoses and is expected to drive many new discoveries as the field continues to evolve.


Anna Hurst, MD, MS (UAB)


Raven Hildreth, BS (UAB)


Hannah Frerichs, MS, LCGC (UAB)


Susan Hiatt, PhD (HAIB)


Greg Cooper, PhD (HAIB)


Meagan Cochran, MS, CGC (HAIB)

Glossary

A few definitions that may help you better understand genomics:

- **Genes** are sections of DNA that are important for building and operating a human body.
- **Genome** is the complete set of genes or genetic material present in every cell in your body.
- **A genetic variant** is a difference in the genetic code of DNA. All people have differences in their DNA. Most are not harmful and do not affect your health. Some can be harmful and cause health conditions.
- **Pharmacogenomics (PGx)** is the study of genes and how they affect response to medications. This information can help physicians choose the right kind and dose of medications for each patient, so that these medications have a good chance to work well with the minimum risk of side effects.
- **Pharmacogenetically actionable findings** may affect the way your body processes certain drugs.
- **Medically actionable gene:** Researchers have proven that there is a connection between a single genetic variant (monogenic) and an increased risk for a specific health condition. If you are found to have one of these genetic differences, there are actions you and your doctor may take to help reduce your risk.
- **Genetic carrier** is a person who has one harmful genetic variant for a condition that requires two harmful variants to cause an effect.
- **Undiagnosed diseases** are conditions where the cause of a patient's symptoms remains unknown after a thorough medical workup. They can involve a wide range of symptoms and may affect many organ systems.
- **Genome sequencing (GS)** is a laboratory process that determines the nearly 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.



A Guide to AGHI Participation and Referral

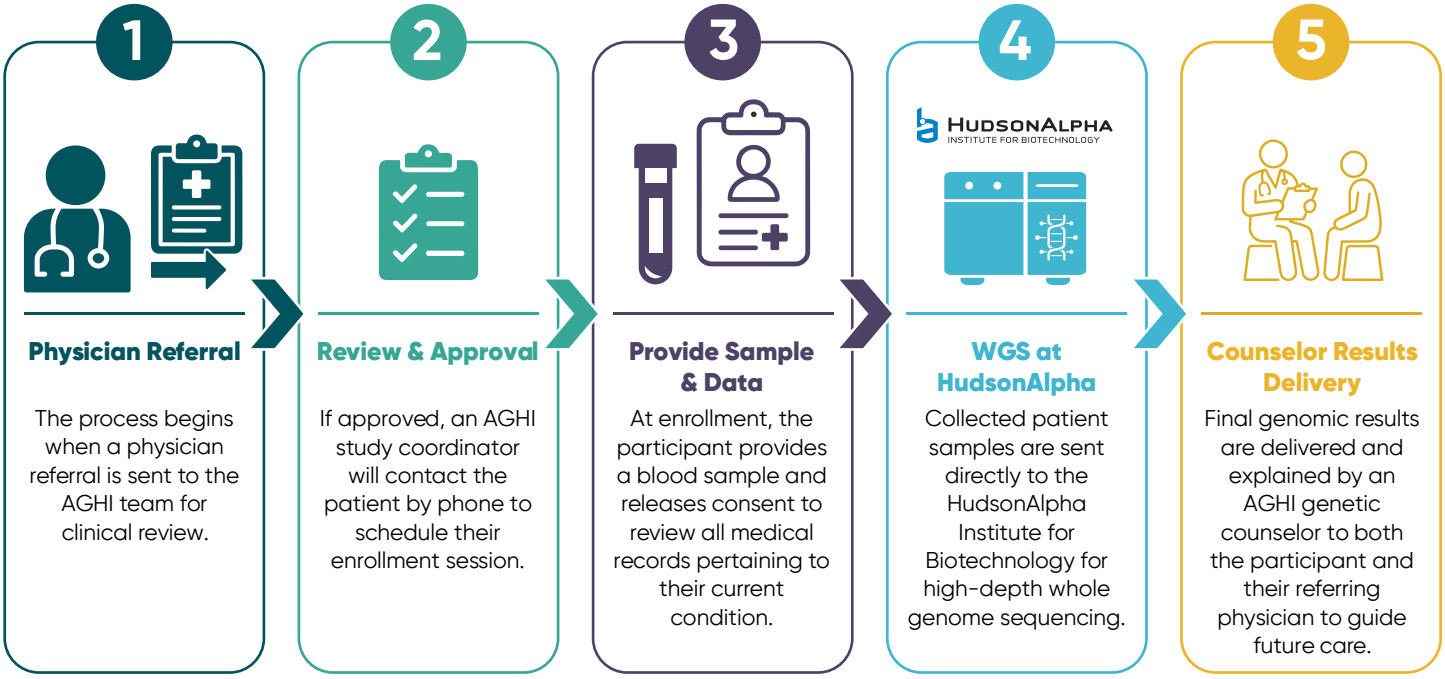
AGHI Alabama Genomic Health Initiative

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**.

HudsonAlpha Institute for Biotechnology · UAB Medicine The University of Alabama at Birmingham · UAB The University of Alabama at Birmingham.

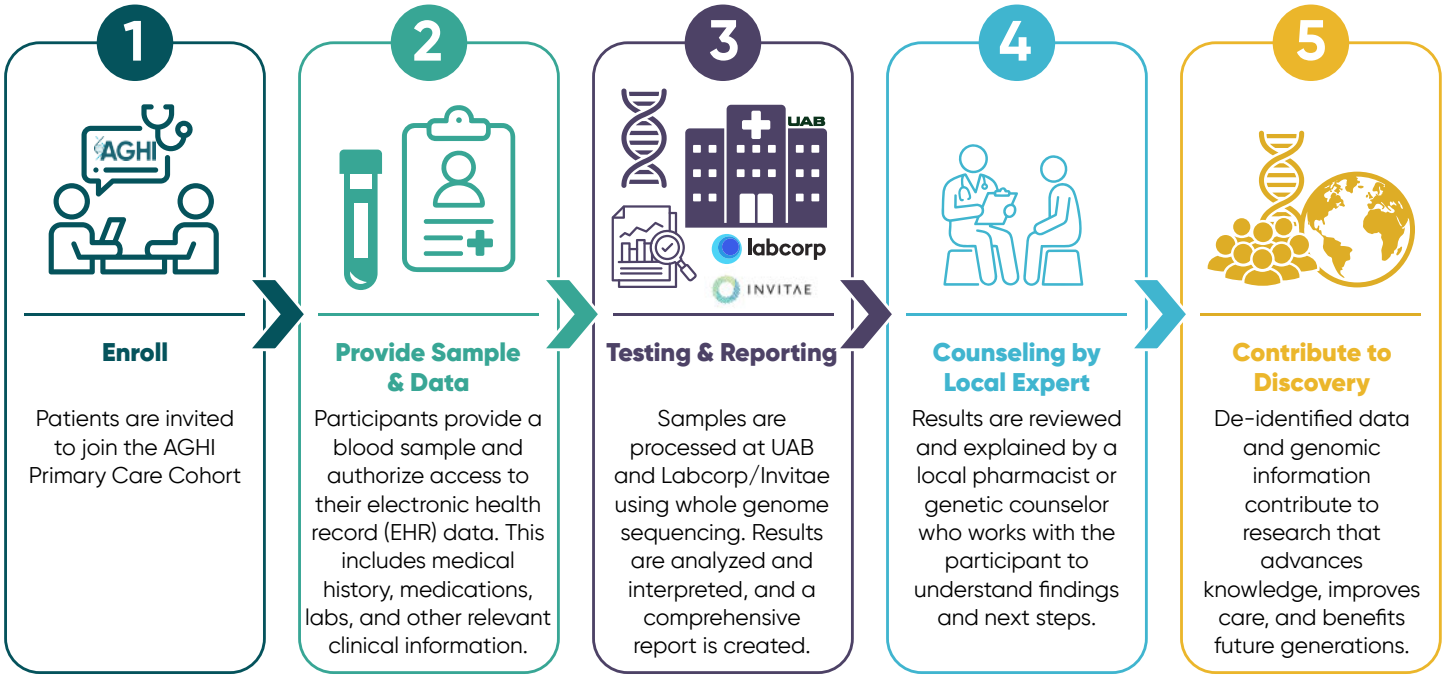
AGHI Alabama Genomic Health Initiative

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

AGHI 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 individuals 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).



Shaping the Future of Genomic Medicine Together, Generating Insights Into Clinical Integration of Genomic Medicine



Shaping the Future of Genomic Medicine Together

Our efforts have long been informed by community advisors, whose perspectives help ensure that our research is ethical, culturally appropriate, and responsive to community needs. Over the past year, we further strengthened these partnerships, deepening our commitment to conducting research with respect, fairness, and transparency while expanding the reach of genomic medicine.

We partnered with our **Center for Clinical and Translational Science (CCTS) Community and Scientific Action Board (CSAB)** comprising 7 researchers and 8 community members—to strengthen connections between scientists and the public and foster the trust needed to support participation in clinical research. The CSAB provided guidance to ensure that informed consent documents, educational materials, and study communications were clear, accessible, and culturally appropriate. They also advised on respectful and effective approaches for returning genetic test results to participants.

This collaboration extends our reach across the CTSA Partner Network (Figure to the right) through the Partner Network Community Coalition (PNCC). Through this network, AGHI investigators can engage with and learn from a broader and more diverse community, ensuring that community perspectives inform study design, implementation, and dissemination.



Guided by Participant Perspectives

To ensure that our efforts understand the patient/participant perspective, we have created a Genomic Medicine Participant Advisory Board. This allows participants who have received genetic test results to share their experiences. This helps us ensure accurate communication, manage emotional and ethical implications, and ensure all our work is rooted in the lived experience of the people impacted.



Time, Tech, and Training: Empowering Clinics for Genomic Medicine

Through AGHI, we evaluated the implementation of genomic medicine in three Alabama primary care clinics (light teal pins in the map below), surveying and interviewing physicians, nurses and clinic managers to assess readiness, barriers, and strategies for integrating genomic testing into routine patient care.

Primary care clinicians see value in genomic medicine, but broader adoption will depend on targeted investments in education, infrastructure, and implementation support to make genomic information practical and actionable in everyday patient care. The study suggests that successful integration of genomic medicine into primary care will require investments in workforce training, health information technology, clinical decision-support tools, and access to genetic specialists.







Ready for the Future? Funding Infrastructure for FQHC Genomic Care

As part of a pilot study led by the AGHI Community Engagement team, healthcare providers and administrative staff working for Alabama Regional Medical Services (ARMS) clinics (dark teal pins in the map below), were interviewed to assess readiness, interest and barriers for genomic medicine implementation. Findings, as reported in a 2026 in Public Health Genomics article, include that while staff are committed to genomic medicine, barriers such as limited budgets, staffing shortages, and language challenges hinder readiness. Successful implementation for underserved populations requires dedicated leadership, targeted staff training, and culturally appropriate patient resources.





Mona Fouad, MD (UAB)



Stephen Sodeke, PhD, DBE (Tuskegee)



Edna Fields, BS (UAB)



Lori Bateman, PhD, RD (UAB)



Tiffany Osborne, (UAB)



Larry Hearld, PhD (UAB)



Clifford Kennon, EdS, MPA (UAB)



Emory Heffernan, PharmD (UAB)



Fueling the Future: Advancing Genomic Research and Empowering the Next Generation of Scientists



Fueling the Future: Advancing Genomic Research and Empowering the Next Generation of Scientists

Alabama's investment in the Alabama Genomic Health Initiative continues to produce meaningful returns by supporting research that improves health outcomes, attracts additional funding, and advances precision medicine. Through access to genomic data, biospecimens, and associated clinical information, researchers can address important health challenges more efficiently while maximizing the value and impact of previously collected resources. These resources support various ancillary research projects, foster new collaborations, generate new research opportunities, and help position the state as a leader in biomedical research.

The projects highlighted below demonstrate how AGHI resources are contributing to scientific discovery and supporting the development of more personalized approaches to disease prevention and treatment.

Advancing precision medicine to improve the Safety of Blood Thinner Medications
Blood thinners are medications that prevent blood clots from forming or getting larger. They do not dissolve existing clots but are essential for preventing life-threatening conditions like heart attacks, strokes, and pulmonary embolism. By analyzing samples from AGHI participants who consented to future research, Dr. Davis is studying how genomic, metabolomic, and clinical factors affect risk for bleeding complications among patients on blood thinner medications. This will support the development of more personalized approaches to prescribing and monitoring blood thinner therapies.



Brittney Davis, PharmD

Advancing Precision Medicine for Multiple Sclerosis
Multiple sclerosis (MS) is a chronic, autoimmune disease of the central nervous system. The immune system mistakenly attacks the protective sheath covering nerve fibers in the brain, spinal cord, and optic nerves. This damage disrupts electrical signals, causing a wide range of neurological symptoms.

By understanding how clinical, genomic, and metabolomic data affects MS, Dr. Davis (above) and Dr. Rodriguez can shed light on biological processes associated with the disease and treatment response to MS therapies and develop personalized treatment strategies.



Kevin Rodriguez, PharmD

Waking Up the Brain's Defense Against Alzheimer's
Alzheimer's disease is a progressive, irreversible brain disorder that accounts for 60% to 80% of dementia cases. It slowly destroys memory and thinking skills by causing abnormal protein buildup that damages and eventually kill nerve cells in the brain.

Risk for Alzheimer's disease is about 50% due to genetics. Risk genes influence the likelihood of developing the disease. TREM2 is one such gene. TREM2 is like a radar system on the brain's cellular cleanup crew (called microglia) that detects toxic garbage, triggers a rapid cleanup response, and builds a protective shield around dangerous plaques to keep them from spreading and destroying healthy brain cells.

By using the genetic data from AGHI and recontacting participants who agreed to this, Drs. Roberson and Cochran are studying Alabamians with genetic changes in TREM2 so that we can develop therapies to delay or slow onset of cognitive impairment and Alzheimer's disease.



Erik Roberson, MD



Nick Cochran, PhD

Genetics of Inflammation in Gout



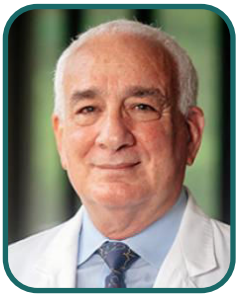
Tony Merriman, PhD

Gout is a common form of arthritis that affects about 5% of adults in the United States. It causes sudden, painful attacks of joint inflammation called "flares." These flares happen when needle-like uric acid crystals build up in joints and trigger the body's immune system.

Not everyone with high uric acid levels develops gout, and researchers want to understand why some people are more likely to get gout than others. Previous genetic studies have identified hundreds of genetic differences linked to gout risk. Some of these genetic changes may affect how the immune system responds to uric acid crystals. It is hypothesized that certain genetic factors may increase gout risk by making immune cells overly sensitive to uric acid crystals, leading to stronger inflammatory responses and more gout flares.

By studying immune cells from AGHI participants who had agreed to participate in future research, researchers can compare gene activity in immune cells from people who carry many versus few gout-risk genes. This can help us identify biological pathways that drive gout.

The Stiff Heart and the Swollen Joint: ***Deciphering the Shared Genetic Architecture of Gout and HFpEF*****.**



Louis Dell'Italia, MD

Heart failure affects nearly 6.7 million adults. About 1 in 4 people will develop it in their lifetime. Heart failure means your heart muscle isn't pumping blood as well as it should. To find out why, your doctor measures your ejection fraction (EF), the percentage of blood your heart pushes out with each beat. Based on the EF, heart failure falls into two main types:

- About 45-50% of heart failure cases have a low EF because the heart muscle is too weak and stretched out to push blood forward effectively (squeezing problem).
- About 35-45% of heart failure cases have a preserved EF (HFpEF). The heart muscle is too thick and stiff to relax and fill with enough blood between heart beats (filling problem).

Xanthine oxidase (XO) is a natural enzyme in the body that acts like a factory producing uric acid and harmful molecules. In patients with HFpEF, this XO factory goes into overdrive creating a large quantity of harmful molecules that cause blood vessels to narrow, increasing inflammation. This in turn makes the heart get even stiffer.

Drs. Merriman (above) and Dell'Italia are studying how genes affect XO expression and activity using bio-banked samples from AGHI participants who had consented for future research

These projects are not just advancing science, they are also providing training opportunities, fostering new collaborations, and supporting the development of future research initiatives. These efforts are helping build a foundation for precision medicine approaches that may improve care and outcomes for Alabamians.

Polygenic Risk Scores: Prediciting Disease Before it Starts

Chronic diseases are the leading driver of illness, disability, and healthcare spending in the United States. Today, 60% of adults live with at least one chronic disease, and 42% have two or more. These conditions account for more than 90% of the nation's $4.5 trillion annual healthcare costs. Most healthcare dollars are spent treating disease after symptoms appear rather than preventing it in the first place. New advances in genetics offer an opportunity to change that.

The Alabama Genomic Health Initiative (AGHI) is helping identify people who may be at higher risk for serious diseases long before symptoms develop. Researchers are studying both rare genetic changes and combinations of many small genetic variations that can increase a person's risk for common conditions such as diabetes, cancer, and kidney disease.

One promising tool is the **Polygenic Risk Score (PRS)**. A PRS combines thousands of small genetic markers into a single score that estimates a person's inherited risk of developing a disease. This information can help doctors act earlier- such as recommending cancer screenings at younger ages or creating personalized plans for diet, exercise, and medication.

AGHI researchers are applying this approach to one of Alabama's most pressing health challenges: diabetic kidney disease. Kidney disease is a common and serious complication of diabetes that often develops silently for years. By the time it is diagnosed, significant and irreversible damage may already have occurred.

Supported by AGHI, researchers analyzed health information from more than 81,000 participants with type 2 diabetes. Their goal was to identify who was most likely to develop kidney disease before symptoms appeared. The team combined information already available in medical records, including laboratory results, medications, healthcare visits, and factors such as access to care.

The results were striking. The advanced prediction model identified more than twice as many people at risk compared with traditional approaches, while minimizing false alarms.



Ahmed Khattab, MD, PhD



Marguerite Irvin, PhD, MS



Hemant Tiwari, PhD

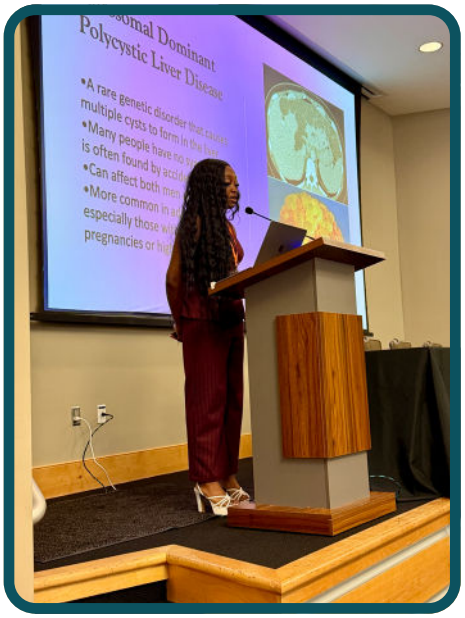


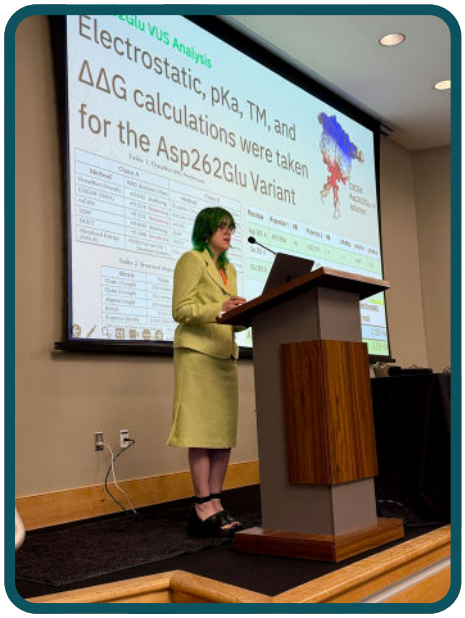
Nita A. Limdi, PharmD, PhD, MSPH, FAHA, FACTS

Advancing Genetics Education Through Real-World Case Studies

AGHI is also a key source for genetics education, including to promote understanding the process of rare disease testing and diagnosis. As part of this effort, AGHI genetic counselors (GCs) at HudsonAlpha develop case studies that present patient symptoms, medical histories, genetic testing results, and their health implications. These cases help students understand how genomic information influences patient care, family health, and clinical decision-making. These case studies provide real-world scenarios with which learners at a wide range of levels, from middle school to graduate level, can engage. For example, the "CODE" program ("Characterizing our DNA Exceptions", https://www.hudsonalpha.org/code/) uses these case studies to introduce college students to the process of analyzing "variants of uncertain significance" (or "VUSs"), which often arise during genomic testing of rare disease (AGHI alone has yielded over 100 VUSs).

This past year (2025-2026), AGHI counselors have developed an all new series of case studies based on recent data from AGHI, including long-read genome sequencing data and recently discovered disease genes. This work ensures that genetics education materials evolve and advance to reflect the changes happening on the research and clinical levels. They have used those new case studies to train genetic counseling graduate students at UAB, and shared them with training programs at other universities including Vanderbilt, Sarah Lawrence, and BayPath. In this way, **AGHI blends its cutting-edge research and medical innovations into educational efforts to empower future generations of Alabamians and beyond to best benefit from, and build on, those innovations in the future**.







A Chance Invitation, A Career in Genomic Medicine

by Brittney Davis

My connection to the Alabama Genomic Health Initiative (AGHI) began in 2017 with an AGHI postcard. Little did I know at the time that this postcard would shape my career trajectory and ultimately lead me to a career in genomic medicine.



I was entering my final year of pharmacy training, and I had developed an interest in genetics through genealogy research. I was intrigued by AGHI's innovative approach to identifying individuals with genetic variants associated with serious but potentially preventable health conditions. Recognizing the opportunity to contribute to a groundbreaking statewide research effort, I enrolled as a participant.

AGHI provided my first real exposure to genomic medicine and demonstrated how genetic information could be used to improve patient care. Seeing how genomic information could help identify health risks, guide prevention and treatment strategies, and support more personalized healthcare sparked a passion that continues to drive my research today.

After completing pharmacy school, I sought specialized training in pharmacogenomics, or the use of genetic information to guide medication selection and dosing. This led me to pursue a fellowship through the UAB-HudsonAlpha Genomic Medicine T32 training program under the mentorship of Nita Limdi, PharmD, PhD, MSPH.

During my fellowship, I collaborated with AGHI investigators at UAB and HudsonAlpha to evaluate the potential impact of pharmacogenomics across Alabama. Through this work, I helped determine how many Alabamians are prescribed medications for which genetic information could improve treatment decisions and how common genetic differences that influence medication effectiveness and safety are within the state's population. These findings helped demonstrate the potential for genomic medicine to improve medication use and patient outcomes across Alabama.

AGHI resources have continued to serve as a foundation for my independent research career, supporting ancillary studies that led to the successful award of a National Institutes of Health (NIH) K23 Career Development Award focused on improving anticoagulation outcomes through precision medicine. Additionally, AGHI data and resources have also supported a pilot project investigating multiple sclerosis treatment response, creating new opportunities to translate genomic discoveries into improved patient care.

My journey from AGHI participant to trainee and to a federally funded investigator illustrates the long-term impact of Alabama's investment in genomic medicine. AGHI has not only generated valuable scientific and clinical insights but has also directly helped train and retain the next generation of scientific leaders in genomic medicine. These investigators continue to bring external funding, innovation, and precision medicine expertise to the state while generating new opportunities to improve health outcomes for Alabamians.



Brittney Davis, PharmD (UAB)

AGHI Publications

PUBLICATION TITLE	PUBMED ID
Integrating genomic medicine into primary care -examining perceptions of community advisory board members.	J Community Genet. 2026 https://pubmed.ncbi.nlm.nih.gov/42303954/
Implementing Genomic Medicine in Primary Care: A Convergent Mixed Method Case Study.	J Eval Clin Pract. 2026 https://pubmed.ncbi.nlm.nih.gov/41773642/
Implementing Genomic Medicine in a Federally Qualified Health Center: Assessing Readiness through a Mixed-Methods Approach.	Public Health Genomics. 2026 https://pubmed.ncbi.nlm.nih.gov/41802142/
Monoallelic and biallelic KDM5A variants identified in patients with autism spectrum disorder.	HGG Adv. 2026 https://pubmed.ncbi.nlm.nih.gov/41889174/
Where risk becomes visible: a layered fixed-policy framework for diabetic kidney disease screening in type 2 diabetes.	medRxiv. 2026 https://pubmed.ncbi.nlm.nih.gov/42078401/
Heterozygous CECR2 variants support a distinct neurodevelopmental syndrome with features overlapping cat eye syndrome	HGG Adv. 2026 https://pubmed.ncbi.nlm.nih.gov/41964217/
Factors influencing trustworthiness and perceived biases of medical information and genetic testing for Black and White Americans.	PLoS Genet. 2025 https://pubmed.ncbi.nlm.nih.gov/41171888/
PTBP1 variants displaying altered nucleocytoplasmic distribution are responsible for a neurodevelopmental disorder with skeletal dysplasia.	Clin Invest. 2025 https://pubmed.ncbi.nlm.nih.gov/40965981/
Long-read genome sequencing and variant reanalysis increase diagnostic yield in neurodevelopmental disorders.	Genome Res. 2024 https://pubmed.ncbi.nlm.nih.gov/39299904/
Key contextual factors involved with participation in medical and genomic screening and research for African American and White Americans: a qualitative inquiry.	J Community Genet. 2024 https://pubmed.ncbi.nlm.nih.gov/39495399/
De novo variants predicting haploinsufficiency for DIP2C are associated with expressive speech delay.	Am J Med Genet 2024 https://pubmed.ncbi.nlm.nih.gov/38421105/
Variants in LRRC7 lead to intellectual disability, autism, aggression and abnormal eating behaviors.	Nat Commun. 2024 https://pubmed.ncbi.nlm.nih.gov/39256359/
Does genetic testing offer utility as a supplement to traditional family health history intake for inherited disease risk?	Fam Pract. 2023 https://pubmed.ncbi.nlm.nih.gov/36856778/
Deleterious, protein-altering variants in the transcriptional coregulator ZMYM3 in 27 individuals with a neurodevelopmental delay phenotype.	Am J Hum Genet. 2023 https://pubmed.ncbi.nlm.nih.gov/36586412/

PUBLICATION TITLE	PUBMED ID
Parents' Perspectives on the Utility of Genomic Sequencing in the Neonatal Intensive Care Unit.	J Pers Med. 2023 https://pubmed.ncbi.nlm.nih.gov/37511639/
Genome sequencing as a first-line diagnostic test for hospitalized infants.	Genet Med. 2022 https://pubmed.ncbi.nlm.nih.gov/34930662/
The seventh international RASopathies symposium: Pathways to a cure-expanding knowledge, enhancing research, and therapeutic discovery.	Am J Med Genet A. 2022 https://pubmed.ncbi.nlm.nih.gov/35266292/
Medical and psychosocial outcomes of state-funded population genomic screening.	Clin Genet. 2023 https://pubmed.ncbi.nlm.nih.gov/37340305/
Poison exon annotations improve the yield of clinically relevant variants in genomic diagnostic testing.	Genet Med. 2023 https://pubmed.ncbi.nlm.nih.gov/37161864/
The Therapeutic Odyssey: Positioning Genomic Sequencing in the Search for a Child's Best Possible Life.	AJOB Empir Bioeth. 2021 https://pubmed.ncbi.nlm.nih.gov/33843487/
Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls.	Genet Med. 2021 https://pubmed.ncbi.nlm.nih.gov/32989269/
Phenotypic expansion of the BPTF-related neurodevelopmental disorder with dysmorphic facies and distal limb anomalies.	Am J Med Genet A 2021 https://pubmed.ncbi.nlm.nih.gov/33522091/
Recruiting diversity where it exists: The Alabama Genomic Health Initiative.	J Genet Couns. 2020 https://pubmed.ncbi.nlm.nih.gov/32220047/
Evaluation of population-level pharmacogenetic actionability in Alabama.	Clin Transl Sci. 2021 https://pubmed.ncbi.nlm.nih.gov/34121327/
Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation.	Genet Med. 2021 https://pubmed.ncbi.nlm.nih.gov/34012067/
A state-based approach to genomics for rare disease and population screening.	Genet Med. 2021 https://pubmed.ncbi.nlm.nih.gov/33244164/
Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients	Sci Adv. 2020 https://pubmed.ncbi.nlm.nih.gov/33268356/
Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis	Neurol Genet 2020 https://pubmed.ncbi.nlm.nih.gov/32802946/

The AGHI Team and Timeline



Leadership



Nita A. Limdi, PharmD PhD MSPH FAHA FACTS (UAB)
Dr. Ray L Watts Heersink Endowed Chair in Neurology
Associate Dean for Genomic Medicine
Director, Alabama Genomic Health Initiative
Director, Genomic Medicine T32 Training program



Greg Cooper, PhD (HAIB)
Faculty Investigator, HudsonAlpha Institute for Biotechnology
Co-director Alabama Genomic Health Initiative
Co-director Genomic Medicine training Program



Matthew Might, PhD (UAB)
Director of UAB Hugh Kaul Precision Medicine Institute
Co-director Alabama Genomic Health Initiative

Senior Advisors



Bruce R. Korf, MD PhD
Distinguished Emeritus Professor



Greg Barsh, MD PhD (HIAB)
Faculty Investigator, HudsonAlpha Institute for Biotechnology

Informatics Bioinformatics



Jim Cimino, MD (UAB)
Director of UAB Informatics Institute



David Crossman, PhD (UAB)
Professor, Dept of Genetics



Chrisly Dillon, MD MHA (UAB)
Senior Research Scientist

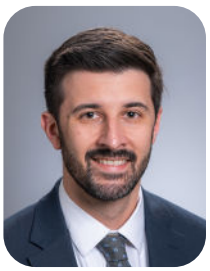
Pharmacogenetics



Brittney Davis, PharmD (UAB)
Assistant Professor, Translational Pharmacogenomics Program



Blake Goff, PharmD (UAB)
Pharmacogenomics Pharmacist, Translational Pharmacogenomics Program



Josh Cortopassi, PharmD (UAB)
Pharmacogenomics Pharmacist, Translational Pharmacogenomics Program

Implementation Science



Larry Hearld, PhD (UAB)
Associate Professor, Director of Research



Lori Bateman, RD PhD (UAB)
Associate Professor UAB General Internal Medicine and Population Science



Ria Hearld, PhD (UAB)
Professor and Director, PhD in Administration-Health Services Program

Medical Genetics & Primary Care



Nathaniel Robin, MD (UAB)
Professor, Departments of Genetics, Pediatrics, and Otolaryngology



Irfan Asif, MD (UAB)
Heersink Endowed Professor of Sports & Exercise Medicine
Chair, Family & Community Medicine



Anna Hurst, MD MS (UAB)
Professor, Clinical Genetics



Erin Delaney, MD (UAB)
Neil E. Christopher, M.D., Endowed Chair in Family & Community Medicine



Stuart Cohen, MD (UAB)
Professor of Medicine
Chief, UAB Primary Care

Community Engagement, Ethical and Social Implications



Mona Fouad, MD (UAB)
Senior Associate Dean for Access & Engagement



Thomas May, PhD (HAIB, WSU)
Research Professor
Medical Ethics Program Director



Stephen Sodeke, PhD DBE (Tuskegee)
Bioethicist and Professor of Bioethics and Allied Health Sciences, Center for Biomedical Research



Mariko Nakano, PhD (UAB)
Assistant Professor of Bioethics at UAB School of Medicine



Lori Bateman, RD PhD (UAB)
Associate Professor UAB General Internal Medicine and Population Science



Tiffany Osborne, (UAB)
Staff Director, Partnerships & Engagement Program

Genetic Counseling



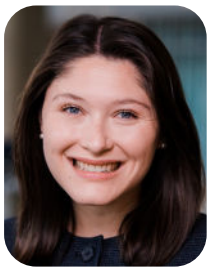
Kelly East, MS CGC (HAIB)
Certified Genetic Counselor, HudsonAlpha Institute for Biotechnology



Whitley Kelley, MS, CGC (HAIB)
Certified Genetic Counselor, HudsonAlpha Institute for Biotechnology



Jessica Denton, PhD MS CGC (UAB)
Program Director, Genetic Counseling Assistant Professor



Hannah Frerichs, MS LCGC (UAB)
Genetic Counselor General Adults, Prenatal Genetics



Meghan Bartos, MS LCGC (UAB)
Clinical and Research Genetic Counselor (GC)



Meagan Cochran, MS CGC (HAIB)
Director and Certified Genetic Counselor, HudsonAlpha Institute for Biotechnology

Biobanking and Genotyping / Sequencing



Jeffrey Edberg, PhD (UAB)
Professor in UAB Division of Clinical Immunology and Rheumatology



Yunjia Chen, PhD FACMG (UAB)
Assistant Professor Clinical Genetics



Shuko Harada, MD (UAB)
Professor, Genomic Diagnostics and Bioinformatics



Jay Holliday, MS (UAB)
Lead Medical Technologist (Diagnostic Molecular Biology)



Zach Bonnstetter, PhD (HAIB)
Computational Biologist



Susan Hiatt, PhD (HAIB)
Research Faculty Investigator



James Lawlor, (HAIB)
Computational Biologist and Bioinformatician



Don Latner, PhD (HAIB)
Senior Scientist

Administration and Recruitment



Stephanie Carroll, BS CRA (UAB)
Manager Research Administrtion



Jennifer Mahaffey, BS (UAB)
Clinical Research Regulations



Irene P. Moss, MA MPH (UAB)
Operations Administrator, UAB Cystic Fibrosis Research Center



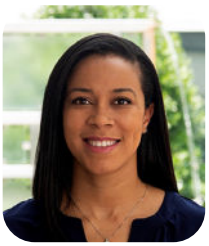
Raven Hildreth, BS (UAB)
Clinical Research Coordinator



Edna Fields, BS (UAB)
Clinical Research Coordinator



Julie Schach, BS (UAB)
Staff Co-Director, Participant Access to Research Core Staff Director



Candice Finnila, PhD (HAIB)
Clinical Research Coordinator for HudsonAlpha Institute for Biotechnology



Aida El Kholi Starling, PhD (HAIB)
Director of Research Administration at HudsonAlpha Institute for Biotechnology

AGHI TIMELINE
2017 – 2024

MAY 2017
AGHI Begins Enrollment
Participant enrollment begins at The Kirklin Clinic of UAB Hospital in Birmingham, marking the launch of the Alabama Genomic Health Initiative

JUNE 2017
First WGS Participant
First whole genome sequencing participant enrolled at North Alabama Children's Specialists

NOVEMBER 2017
Precision Medicine Milestone
The first diagnosis is returned in the whole genome sequencing cohort, and the first medically actionable risk variant is identified in the population cohort.

AUGUST 2018
Expanded Enrollment Access
Enrollment expands to include the UAB Montgomery Regional Medical Campus and UAB Selma Family Medicine, increasing access across Alabama.

APRIL 2019
Major Enrollment Milestone
AGHI surpasses 5,000 enrolled participants, marking a significant milestone in the growth of Alabama's genomic medicine and research cohort

MARCH 2020
Enrollment Suspended
Population cohort enrollment is temporarily suspended due to the COVID-19 pandemic while analysis of previously enrolled participants continues.

SEPTEMBER 2020
Enrollment Resumed
Whole genomic sequencing enrollment resumes at the Children's Health Research Unit

JANUARY 2021
Rare Diseases Symposium
AGHI co-sponsors 8th Annual Rare Disease Symposium: Next Generation Technology to Manage Rare Disease

JULY 2021
Pharmacogenomic Testing Added
Pharmacogenomic testing is incorporated into AGHI to help personalize medication selection and improve patient care

SEPTEMBER 2021
Major Enrollment Milestone
AGHI surpasses 7,500 enrolled participants, reflecting continued statewide growth and engagement

JANUARY 2022
Cardiogenomics Clinic Opens
The new Cardiogenomics Clinic at UAB opens to provide specialized follow-up care and genetic counseling for AGHI participants identified with inherited cardiovascular disease risks

FEBRUARY 2022
Rare Diseases Symposium
AGHI co-sponsors 9th Annual Rare Disease Symposium: Telehealth and Therapeutic Advances in Rare Disease Care

OCTOBER 2022
National Poster Presentations
AGHI presents "Integrating Genomics into Primary Care" at the 2022 CPIC and ASHG Annual Meetings, highlighting its innovative approach to genomic medicine implementation

FEBRUARY 2024
Rare Diseases Symposium
AGHI co-sponsors the 11th Annual Rare Disease Symposium focusing on AI, genomics, and healthcare policy

FEBRUARY 2024
Long-Read Sequencing
The first participant undergoes long-read sequencing through the Undiagnosed Disease Cohort at UAB Medicine, expanding diagnostic capabilities for rare diseases

MARCH 2024
PGx Results in Under 14 Days
UAB offers in-house pharmacogenomic (PGx) testing with a turnaround time of under 14 days

APRIL 2024
PGx Clinical Decision Support
Pharmacogenomic clinical decision support is integrated into the electronic health record, providing medication guidance to clinicians at the point of care

APRIL 2024
Expert-Guided Reporting
Pharmacists (PharmDs) develop tailored reports for both physicians and patients and provide access through a user-friendly online portal

JUNE 2024
Expansion in Care
Whole genome sequencing services expand to include both pediatric and adult participants

JULY 2024
$2.9M NIH Grant Awarded
UAB receives a $2.9 million NIH grant to support AGHI participants through the Undiagnosed Disease Program and advance genomic discovery

AGHI TIMELINE

2024 - Present

FEBRUARY 2025

Leadership Change

Dr. Nita Limdi assumes the role of Principal Investigator for the AGHI

MARCH 2025

Disease Risk Testing

Implemented disease risk testing with Invitae, providing clinically actionable reports. Positive findings were identified in 9% of participants, enabling personalized follow-up care

APRIL 2025

Clear, Counselor-Guided Results

Genetic counselor–led result disclosure helps patients understand their findings and provides physicians with clear next steps and consultation summaries

JUNE 2025

Family History Integration & Training

Integrated family history assessment while providing genetic counseling students with hands-on research experience

JULY 2025

Community-Engaged Partnership

AGHI partners with the Center for Clinical and Translational Science Community and Science Action Board to ensure community voices shape meaningful action

AUGUST 2025

Major Enrollment Milestone

AGHI surpasses 10,000 enrolled participants, demonstrating the program's continued statewide impact and growth

AUGUST 2025

Expanding Genomic Screening

Awarded a $4.87 million Population Genomic Screening grant to broaden access to genomic screening and enable earlier, personalized healthcare

SEPTEMBER 2025

UAB-HAIB Leadership Meeting

Bringing leaders together to discuss research priorities and foster collaboration

OCTOBER 2025

Streamlined Remote Participation

Expanded access through remote consent, making participation easier for patients and families

DECEMBER 2025

Genomic Participant Advisory Board

Bringing participant experiences to the forefront and helping shape the future of genomic medicine

FEBRUARY 2026

Expanded Clinical Laboratory Capabilities

Added Broad Clinical Laboratory capabilities to enhance genomic assessment for both research and clinical care

APRIL 2026

Featured at Keystone Symposium

AGHI was highlighted as a model for genomic and precision population health at Keystone Symposia - Closing the Gap in Global Genomics Keystone

How Alabama is Leading the Way in Genomic Medicine

Hockey legend Wayne Gretzky famously advised, "**Skate to where the puck is going, not to where it has been.**" That philosophy perfectly captures what the Alabama Genomic Health Initiative (AGHI) has enabled Alabama to do in genomic medicine.

From its inception, AGHI positioned Alabama ahead of national and international trends in genomic healthcare. When much of the scientific community was sequencing only 1–2% of the human genome to diagnose disease, AGHI was investing in whole genome sequencing, providing a far more comprehensive view of genetic variation and disease risk. As whole genome sequencing became the standard, AGHI adopted advanced long-read sequencing technologies capable of detecting significant genomic changes that were previously invisible to conventional approaches.

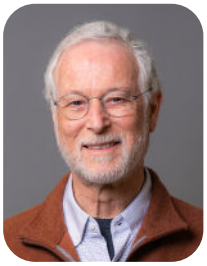
AGHI has also transformed how genomics is used in healthcare. While many programs focused exclusively on patients who already exhibited symptoms of disease, AGHI integrated genomic screening into primary care settings. This proactive approach has identified individuals at risk for serious but preventable conditions long before symptoms develop, creating opportunities for earlier intervention, improved outcomes, and reduced healthcare costs.

Beyond its direct impact on patient care, AGHI has built the infrastructure, workforce, expertise, partnerships, and public trust necessary to establish Alabama as a national leader in genomic medicine. By creating a statewide genomic ecosystem that integrates healthcare providers, researchers, health systems, and communities, AGHI developed capabilities that few states possess.

These investments have enabled Alabama institutions to compete successfully for and lead major federally funded genomic medicine programs and research initiatives, including the NIH All of Us Research Program, the Electronic Medical Records and Genomics (eMERGE) Network, Implementing Genomics in Practice (IGNITE), the Undiagnosed Diseases Network (UDN), population genomic screening initiatives, and the Regional Genomic Medicine eConsult Program. These programs have brought substantial federal investment to the state, expanded access to cutting-edge genomic services for Alabama patients, accelerated research discoveries, and strengthened Alabama's reputation as a destination for genomic innovation.

We are grateful for the support from the State of Alabama for the Alabama Genomic Health Initiative and look forward to continuing to ensure that Alabama is at the forefront of genomic medicine, improving the health of our citizens while strengthening the state's position as a national leader in biomedical innovation, precision medicine, and economic development.

Simply put, AGHI has allowed Alabama to skate to where the puck is going, not where it has been. It has allowed us to capitalize on that foresight for the benefit of patients, communities, researchers, and the state's future.



Bruce R. Korf, MD, PhD



Nita A. Limdi, PharmD, PhD, MSPH, FAHA, FACTS

Skating to Where the Puck Is Going in Genomics: Charting Our Path Forward



As we reflect on the remarkable progress of the Alabama Genomic Health Initiative, we extend our deepest gratitude to the leaders (especially Drs. Korf and Barsh and Ms. Moss), state legislators, healthcare providers, researchers, community partners, and thousands of participating Alabamians who made this journey possible.

The success of AGHI is rooted in the foresight of leaders who recognized that genomics would transform healthcare and in the commitment of policymakers who invested in that vision. Their support enabled Alabama to become a national leader in genomic medicine, delivering tangible benefits to patients while creating a foundation for scientific discovery and innovation.

Today, we stand at an exciting new crossroads. Building on nearly a decade of success and leveraging our leadership in national programs and collaborations, AGHI is evolving toward an even greater opportunity: creating the Alabama Genome-Phenome Resource. By securely linking genomic information with electronic medical record data, we can establish a powerful research and clinical resource that accelerates discovery, improves patient care, and strengthens Alabama's position as a leader in precision health.

This next phase of **AGHI will enable researchers and clinicians to better understand how genetic, environmental, and clinical factors influence health and disease across our populations.** It will also support the development of Polygenic Risk Scores for common chronic conditions such as diabetes, kidney disease, heart disease, cancer, and Alzheimer's disease, helping identify individuals at elevated risk before disease develops and enabling earlier, more effective intervention.

At the same time, this resource will **strengthen Alabama's clinical research enterprise** by making clinical trials more efficient, precise, and accessible. Specifically, it will help:

- Assess trial feasibility by identifying populations that meet study criteria.
- Connect researchers with individuals who have agreed to be contacted about future research opportunities and may be eligible for clinical trials.
- Enrich clinical trials through genomic and phenotype data, enabling identification of participants with specific genetic signatures that are critical for precision medicine studies and targeted therapies.

These capabilities will attract new research investments, expand access to innovative treatments, and position Alabama as a preferred destination for precision medicine research and clinical trials.

The vision that launched AGHI remains unchanged: improving the health of Alabamians through the responsible use of genomic medicine. What has changed is the scale of opportunity before us.

Thank you for your continued partnership and support as we enter this next chapter. Together, we can build a future where discovery happens faster, prevention begins earlier, treatments are more precise, and every Alabamian benefits from the promise of genomic medicine.



Nita A. Limdi, PharmD, Ph.D, MSPH, FAHA, FACTS
Ray L Watts Heersink Endowed Chair in Neurology
Associate Dean for Genomic Medicine
Director, Alabama Genomic Health Initiative
Director, UAB-HAIB Genomic Medicine Training Program

UAB MEDICINE
The University of Alabama at Birmingham

HudsonAlpha
INSTITUTE FOR BIOTECHNOLOGY

412 • 500 22nd Street South
1720 2nd Avenue South
Birmingham, AL 35294-0104

AGHI

11,000+

Have Received Genomic Insights From All 67 Counties

9,299 Participants Received Genetic Risk Information for Common Conditions. For **9%**, testing uncovered hidden risks for chronic disease, guided proactive care

2,768 received pharmacogenomics results. For **23%**, testing personalized current medications. Results can personalize prescriptions throughout life for all

628 patients and **856** family members received whole genome sequencing. For **22%**, sequencing provided a diagnosis and a plan for healthcare

Clinical-Genomic Database

11,000+ participants, **41,000+** biological specimens. **90%** consented to future research. Of these, **99%** also have data from genome-wide testing.

Driving Innovation. Improving Lives. Strengthening Alabama.