

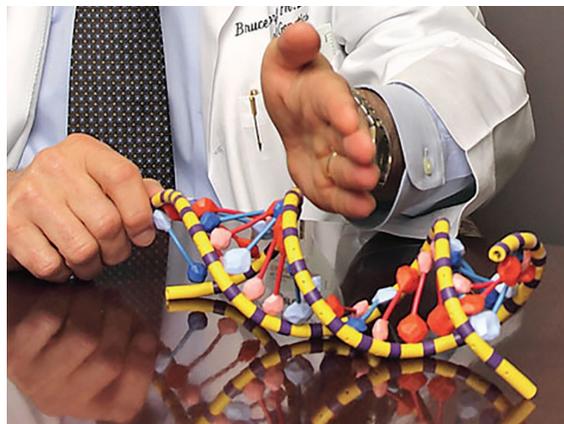


# INSIDE PEDIATRICS

## Recruitment under way for Alabama Genomic Health Initiative UAB, Children's, HudsonAlpha spearhead \$2 million project

In order to better meet health needs across the state, the Alabama Genomic Health Initiative (AGHI) recently launched as a partnership among Children's of Alabama, the University of Alabama at Birmingham (UAB) and the HudsonAlpha Institute for Biotechnology.

Full scale recruitment for the AGHI has begun at UAB clinics in Birmingham. The AGHI, funded by a \$2 million appropriation from the Alabama legislature to UAB, is one of the nation's first statewide efforts to harness the power of genomic analysis to identify those at high risk for genetic disease and provide a basis for continuing research into genetic contributors to health and disease. It will recruit a diverse group of participants from every county in Alabama and provide genomic analysis and interpretation to this group free of charge.



Following the conclusion in May of a pilot project that recruited the first 100 participants, the AGHI has now opened widespread recruitment with the intent of adding 2,000 individuals in the first year. Over a five-year period, the goal will be to increase the number of participants to more than 10,000 persons.

"This project will result in immediate health benefits to some participants, and in the long term will help to address problems of chronic disease and rising health care costs in the state," said Bruce Korf, M.D., Ph.D., chair of the UAB and Children's Department of Genetics and co-director of the AGHI. "It will also position Alabama as a leader at the forefront of 21st century medicine."

For some, the results will indicate an increased risk of a disease for which preventive or treatment strategies exist. Those participants will receive genetic counseling and be linked to appropriate medical care. The initiative will also feature a public education campaign about genomic medicine and create a DNA biobank for research.

Korf anticipates that those who choose to participate will fall into one of two major categories. Most will be generally healthy, or will be receiving medical care for one or more conditions not recognized to have a genetic cause. The other group will be those with a recognized genetic issue whose origin is undetermined. Both groups will provide blood samples that will undergo genomic analysis at HudsonAlpha in Huntsville.

Pediatric patients will be eligible for whole genome analysis. This smaller portion of patients will be identified due to an undetermined or undiagnosed condition. At Children's, testing will be offered in the Genetics Clinic on the sixth floor of the Dearth Tower and at the North Alabama Children's Specialists office in Huntsville.

The blood samples from the larger group — those not known to be affected by a genetic condition — will undergo a genotyping array test, assessing some 650,000 identified genomic biomarkers. The genotyping test will look for the presence of variants in 59 specific genes, referred to as actionable genes by the American College of Medical Genetics and Genomics. An ACMG committee, of which Korf is a member, compiled the list in 2013 after an exhaustive search of medical literature. The 59 genes are those that are known to contribute to disease and for which the potential for prevention or treatment exists.

Ethical, legal and social issues will also be addressed by the AGHI, which has formed a bioethics working group to ensure the initiative conforms to the highest ethical principles. Bioethicists from HudsonAlpha, Tuskegee University and UAB will review all plans and procedures to ensure that appropriate safeguards and protections are in place and will guide the initiative on matters such as privacy, security and informed consent. The AGHI will establish an ethics review panel to investigate and respond to potential issues.

To learn more about the Alabama Genomic Health Initiative, visit [www.aghi.org](http://www.aghi.org). To participate, call 1-855-462-6850.