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NEW STUDY REVEALS MAJOR RACIAL BIAS IN LEADING GENOMICS DATABASES

Discovery by University of Maryland School of Medicine researchers could lead to more cost-effective genetic diagnoses, targeted treatments.

Baltimore, Md., October 11, 2016 – Led by investigators at the University of Maryland School Of Medicine, a national group of researchers has confirmed for the first time that two of the top genomic databases, which are in wide use today by clinical geneticists, reflect a measurable bias toward genetic data based on European ancestry over that of African ancestry. The results of their study were published today in the journal *Nature Communication*.

The research team was led by Principal Investigator **Timothy O'Connor**, PhD, assistant professor at the University of Maryland School of Medicine (UM SOM) and a faculty member of the school's Institute for Genome Sciences. He is also a specialist in the areas of Human Evolutionary Genomics, Genotype/Phenotype Architecture, and Computational Biology. Other members of the study included researchers from UM SOM's Department of Medicine and the Program in Personalized and Genomic Medicine, and from the Johns Hopkins University, the University of Colorado, and the Henry Ford Health System.

This deficit in African ancestry genomic data was identified during an 18-month long study conducted under the auspices of the larger Consortium on Asthma among African-Ancestry Populations in the Americas (CAAPA). To create a benchmark for comparison to current database results, the researchers first created the largest, high-quality non-European genome data set ever assembled. Genetic samples of 642 subjects from the African diaspora, including representatives from US, African, and Afro-Caribbean populations, were sequenced in order to produce this unique data set. Then, when compared with current clinical genomic databases, researchers found a clearer preference in those databases for European genetic variants over non-European variants.

“By better understanding the important role of African ancestry in clinical genetics, we can begin to actually identify a disease that has been forgotten or is not part of an individual's self-identification,” says O'Connor. “For example, if an African-American patient walks in the door, he might have 20 percent European ancestry, while another might have 20 percent African ancestry. That difference will dramatically change how many variants are found in their genome, and what disease risks they might encounter. That's why we need to expand these databases to include a broader range of ancestries, in order to produce more accurate medical genetic diagnoses.”

O'Connor also points out that this shortfall in genomic data also comes at a financial cost. “If you translate the review time it takes for each one of these variants to be sequenced in terms of cost in a clinical setting, you're looking at a difference of about \$1,000 more to analyze an African American's genome than a European American's genome—and you still receive less accurate results,” he notes.



“This groundbreaking research by Dr. O’Connor and his team clearly underscores the need for greater diversity in today’s genomic databases,” says UM SOM Dean **E. Albert Reece**, MD, PhD, MBA, who is also Vice President of Medical Affairs at the University of Maryland and the John Z. and Akiko Bowers Distinguished Professor at UM SOM. “By applying the genetic ancestry data of all major racial backgrounds, we can perform more precise and cost-effective clinical diagnoses that benefit patients and physicians alike.”

About the University of Maryland School of Medicine

The University of Maryland School of Medicine was chartered in 1807 and is the first public medical school in the United States and continues today as an innovative leader in accelerating innovation and discovery in medicine. The School of Medicine is the founding school of the University of Maryland and is an integral part of the 11-campus University System of Maryland. Located on the University of Maryland’s Baltimore campus, the School of Medicine works closely with the University of Maryland Medical Center and Medical System to provide a research-intensive, academic and clinically based education. With 43 academic departments, centers and institutes, and a faculty of more than 3,000 physicians and research scientists and more than \$400 million in extramural funding, the School is regarded as one of the leading biomedical research institutions in the U.S. with top-tier faculty and programs in cancer, brain science, surgery and transplantation, trauma and emergency medicine, vaccine development and human genomics, among other centers of excellence. The School is not only concerned with the health of the citizens of Maryland and the nation, but also has a global presence, with research and treatment facilities in more than 35 countries around the world.

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