

UM School of Medicine Researchers Participate in Landmark Study Detailing Sequencing of Full Human Genomes to Better Capture Genetic Diversity

64 Human Genomes Sequenced Will Serve as New Reference for Genetic Variation and Predisposition to Human Diseases

BALTIMORE, Feb. 25, 2021 – Researchers at the University of Maryland School of Medicine (UMSOM) co-authored a study, published today in the journal *Science*, that details the sequencing of 64 full human genomes. This reference data includes individuals from around the world and better captures the genetic diversity of the human species. Among other applications, the work will enable population-specific studies on genetic predispositions to human diseases as well as the discovery of more complex forms of genetic variation.

Twenty years ago this month, the International Human Genome Sequencing Consortium announced the first draft of the human genome reference sequence. The Human Genome Project, as it was called, required 11 years of work and involved more than 1000 scientists from 40 countries. This reference, however, did not represent a single individual, but instead was a composite of humans that could not accurately capture the complexity of human genetic variation.

Building on this, scientists have conducted several sequencing projects over the last 20 years to identify and catalog genetic differences between an individual and the reference genome. Those differences usually focused on small single base changes and missed larger genetic alterations. Current technologies now are beginning to detect and characterize larger differences – called structural variants – such as insertions of new genetic material. Structural variants are more likely than smaller genetic differences to interfere with gene function.

The new finding in *Science* announced a new and significantly more comprehensive reference dataset that was obtained using a combination of advanced sequencing and mapping technologies. The new reference dataset reflects 64 assembled human genomes, representing 25 different human populations from across the globe. Importantly, each of the genomes was assembled without guidance from the first human genome composite. As a result, the new dataset better captures genetic differences from different human populations.

"We've entered a new era in genomics where whole human genomes can be sequenced with exciting new technologies that provide more substantial and accurate reads of the DNA bases," said study co-author [Scott Devine, PhD](#), Associate Professor of Medicine at UMSOM and faculty member of IGS. "This is allowing researchers to study areas of the genome that previously were not accessible but are relevant to human traits and diseases."

Institute of Genome Science (IGS)'s Genome Resource Center (GRC) was one of three sequencing centers, along with Jackson Labs and the University of Washington, that generated the data using a new sequencing technology that was developed recently by Pacific Biosciences. The GRC was one of only five early access centers that was asked to test the new platform.

Dr. Devine helped to lead the sequencing efforts for this study and also led the sub-group of authors who discovered the presence of “mobile elements” (i.e., pieces of DNA that can move around and get inserted into other areas of the genome). Other members of the Institute for Genome Sciences (IGS) at the University of Maryland School of Medicine are among the 65 co-authors. [Luke Tallon, PhD](#), Scientific Director of the Genomic Resource Center, worked with Dr. Devine to generate one of the first human genome sequences on the Pacific Biosciences platform that was contributed to this study. Nelson Chuang, a graduate student in Dr. Devine’s lab also contributed to the project.

"The landmark new research demonstrates a giant step forward in our understanding of the underpinnings of genetically-driven health conditions," said [E. Albert Reece](#), MD, PhD, MBA, Executive Vice President for Medical Affairs, UM Baltimore, and the John Z. and Akiko K. Bowers Distinguished Professor and Dean, University of Maryland School of Medicine. "This advance will hopefully fuel future studies aimed at understanding the impact of human genome variation on human diseases."

About the University of Maryland School of Medicine

Now in its third century, the University of Maryland School of Medicine was chartered in 1807 as the first public medical school in the United States. It continues today as one of the fastest growing, top-tier biomedical research enterprises in the world -- with 45 academic departments, centers, institutes, and programs; and a faculty of more than 3,000 physicians, scientists, and allied health professionals, including members of the National Academy of Medicine and the National Academy of Sciences, and a distinguished two-time winner of the Albert E. Lasker Award in Medical Research. With an operating budget of more than \$1.2 billion, the School of Medicine works closely in partnership with the University of Maryland Medical Center and Medical System to provide research-intensive, academic and clinically based care for nearly 2 million patients each year. The School of Medicine has more than \$563 million in extramural funding, with most of its academic departments highly ranked among all medical schools in the nation in research funding. As one of the seven professional schools that make up the University of Maryland, Baltimore campus, the School of Medicine has a total population of nearly 9,000 faculty and staff, including 2,500 student trainees, residents, and fellows. The combined School of Medicine and Medical System (“University of Maryland Medicine”) has an annual budget of nearly \$6 billion and an economic impact more than \$15 billion on the state and local community. The School of Medicine, which ranks as the 8th highest among public medical schools in research productivity, is an innovator in translational medicine, with 600 active patents and 24 start-up companies. The School of Medicine works locally, nationally, and globally, with research and treatment facilities in 36 countries around the world.

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