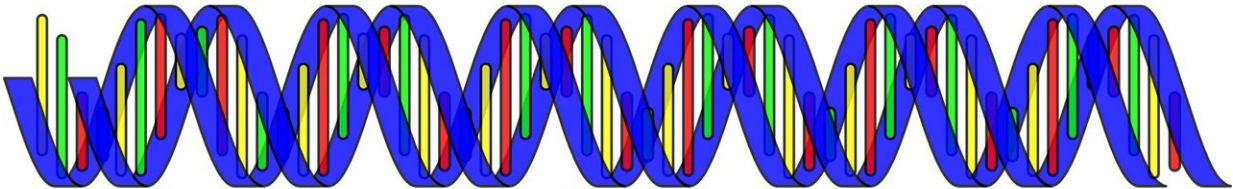


Sequencing African genomes yields new data resource with broad applicability

October 16 2019



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By collaborating globally in a new, large-scale effort, researchers have made strong progress in sequencing genomes from regions and countries across Africa. These findings will enable more broadly representative and relevant studies ranging from basic through clinical genetics. The researchers' new data and preliminary observations were presented as a featured plenary abstract at the American Society of Human Genetics 2019 Annual Meeting in Houston.

"There is a dearth of baseline [genetic data](#) for African populations," said Neil Hanchard, MD, DPhil, Assistant Professor at the Baylor College of Medicine, who presented the work. As part of the Human Heredity and Health in Africa (H3Africa) Consortium, a collaborative effort supported by the National Institutes of Health to conduct [genomic research](#) in Africa, Dr. Hanchard and his colleagues sequenced the whole genomes of 426 individuals from 13 African countries, whose ancestries represented 50 ethnolinguistic groups from across the continent.

Of the 426 genomes sequenced, 320 were analyzed at high depth. This allowed the researchers to examine rare genetic variants in an accurate and quantifiable way, in addition to the common variants that have been the focus of most of the previous genetic studies in Africans, Dr. Hanchard explained.

"We found an impressive breadth of genomic diversity among these genomes, and each ethnolinguistic group had unique genetic variants," he said. "There was a great deal of variation among people in the same region of Africa, and even among those from the same country. This reflects the deep history and rich genomic diversity across Africa, from which we can learn much about population history, environmental adaptation, and susceptibility to diseases."

Beyond the sheer amount of variation within and among the groups studied, the researchers were able to use the data to examine historic patterns and pinpoint migration events that were previously unknown.

"For the first time, our data from East and West Africa showed evidence of movement that took place 50 to 70 generations ago from East Africa to a region in central Nigeria. This movement is reflected in the genomes of a Nigerian ethnolinguistic group and is distinct from previous reports of gene flow between East and West Africa," said Adebowale Adeyemo, MD, deputy director of the Center for Research on Genomics and

Global Health at the National Human Genome Research Institute, and a senior author on the study. "This data gives us a more complete picture of the genetic history of Africa."

In addition to migration, the data showed that infectious disease has played a role in shaping the genomes of African populations. The researchers found many areas of the genome where variation indicated natural selection had taken place. Among these were 33 locations in genes known to be involved in viral infection and metabolism.

"When you consider which forces have shaped the genetic diversity of Africans, you tend to think of mosquito-transmitted diseases like malaria," Dr. Hanchard said. "Our study suggests that [viral infections](#) have also helped to shape genomic differences between people and groups, via genes that affect individuals' disease susceptibility."

The researchers hope their work will lead to wider recognition of the extent of uncatalogued genomic variation across the African continent, and of the need for continued studies of the many diverse populations in Africa. "Adding genomic data from diverse populations is essential to ensure that all global populations can benefit from the advances in health that precision medicine offers," said Zané Lombard, Ph.D., Associate Professor at the Division of Human Genetics of the University of the Witwatersrand, South Africa, and a senior author on the study.

The study authors noted that their findings have broad relevance, from population genetics research into human history and migrations, to basic research into the genetic structure of various groups, to clinical research into the impact of specific variants on health outcomes. Immediate next steps include further examination of their initial findings and leveraging their data to represent more African populations.

"Our findings provide a base for future H3Africa studies to build upon,

as well as the opportunity to include more African genetic sequences in the human reference [genome](#) databases and create a better reference for researchers around the world to use," said Dr. Adeyemo.

In moving the work forward, Dr. Hanchard cited the continuing importance of global collaboration in this research. "It is important to recognize the degree of involvement of scientists from countries across Africa to perform this work, and to raise the profile of African geneticists in all areas of human genetics and genomics research," he said.

More information: N Hanchard et al. (2019 Oct 15). Abstract: High-depth genome sequencing in diverse African populations reveals the impact of ancestral migration, cultural demography, and infectious disease on the human genome. Presented at the American Society of Human Genetics 2019 Annual Meeting. Houston, Texas.

Provided by American Society of Human Genetics

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