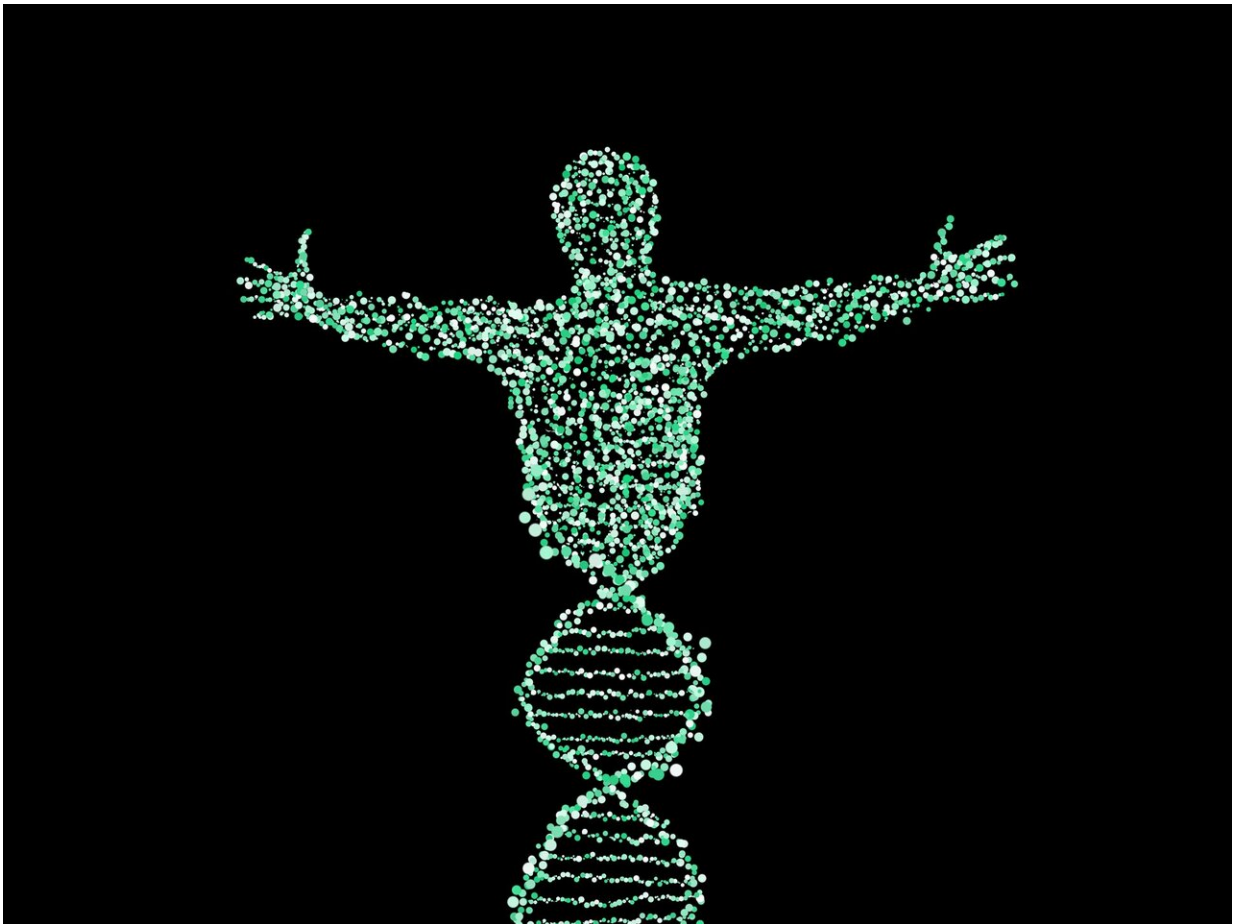


# Major new study unveils complexity and vast diversity of Africa's genetic variation

November 23 2020, by Zané Lombard

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Africa is the [cradle of humankind](#). All humans are descendants from this

common pool of ancestors. Africa and its multitude of ethnolinguistic groups are therefore fundamental to learning more about humankind and our origins.

A human [genome](#) refers to the complete set of genetic information found in a human cell. We inherit our genomes from our parents. Studying the variations in different people's genomes gives important clues to how genetic information influences people's appearance and health. It can also tell us about our ancestry. To date, very few African individuals have been included in studies looking at genetic variation. Studying African genomes not only fills a gap in the current understanding of human genetic variation, but also reveals new insights into the history of African populations.

My colleagues and I, who are all members of the [Human Heredity and Health \(H3Africa\)](#) consortium, contributed to a [landmark genetics study](#). This study focused on 426 individuals from 13 African countries. More than 50 different ethnolinguistic groups were represented in the study—one of the most diverse groups of Africans ever to be included in such an investigation. We sequenced the whole genome of each of these individuals—this means we could read every part of the genome to look for variation.

This study contributes a major, new source of African genomic data, which showcases the complex and vast diversity of African genetic variation. And it will support research for decades to come.

Our findings have broad relevance, from learning more about African history and migration, to [clinical research](#) into the impact of specific variants on health outcomes.

## **Gaining new insights**

One of the key outcomes was the discovery of more than three million new genetic variants. This is significant because we are learning more about human genetic diversity in general, and discovering more differences that could be linked to disease or traits in the future.

This study also adds details to what is known about the migration and expansion of groups across the continent. We were able to show that Zambia was most probably an intermediate site on the likely route of migration from west Africa to east and south Africa. Evidence supporting movement from east Africa to central Nigeria between 1,500 and 2,000 years ago was also revealed, through the identification of a substantial amount of east African ancestry in a central Nigerian ethnolinguistic group, the Berom.

The study also enabled us to reclassify certain variants that were previously suspected to cause disease. Variants that cause serious genetic diseases are often rare in the general population, mostly because their effect is so severe that a person with such a variant often does not reach adulthood. But we observed many of these variants at quite common levels in the studied populations. One wouldn't expect that these types of disease-causing variants would be this common in healthy adults. This finding helps to reclassify these variants for clinical interpretation.

Finally, we found a surprising number of regions with signatures of natural selection that have not been previously reported. Selection means that when individuals are exposed to environmental factors like a viral infection, or a drastic new dietary component, some gene variants may confer an added adaptive advantage to the humans that bear them in their genome.

Our best interpretation of these findings is that as humans across Africa were exposed to different environments—sometimes as a result of migration—these variants were likely important to surviving in those

new conditions. This has left an "imprint" on the genome and contributes to genomic diversity across the continent.

Our data has shown that we have not yet found all the variation in the [human genome](#). There is more to learn by adding new, unstudied population groups. We know that [less than a quarter](#) of participants in genomics research are of non-European ancestry. Most available genetic data come from just [three countries](#) – the UK (40%), the US (19%) and Iceland (12%).

It is essential to keep adding more genomic data from all global populations—including Africa. This will ensure that everyone can benefit from the advances in health that precision medicine offers. Precision medicine refers to the customisation of healthcare to fit the individual. Including personal genetic information could radically change the nature and scope of healthcare options that would work best for that individual.

### **Next steps**

The Human Heredity and Health consortium is now in its eighth year of existence, and supports more than 51 diverse projects. These include studies focusing on diseases like diabetes, HIV and tuberculosis. The reference data generated through our study are already being put to use by many of the consortium's studies.

Next, we are planning to take an even deeper look at the data to better understand what other types of [genetic variation](#) exist. We are also hoping to add further unstudied populations to grow and enrich this data set.

Building capacity for genomics research on the African continent is a key goal of Human Heredity and Health. An important aspect of this

study is that it was driven and conducted by researchers and scientists from the African continent. Researchers from 24 institutions across Africa participated and led this investigation. This study showcases the availability of both infrastructure and skills for large-scale genomics research on the continent. It also highlights the prospect of future world-class research on this topic from Africa.

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