Exploring African DNA can transform understanding of human genetics – what does that mean for medicine?

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Why African genomics holds the key to precision medicine and global health equity.

Africa is home to more human genetic diversity than anywhere else on Earth. The average African genome shows nearly a million more genetic variants than the average non-African genome.

But this wealth of data remains largely unexplored, with African populations historically underrepresented in research. Less than 3% of global genomic datasets and genome-wide association studies include participants of African descent.

Renowned African researchers have released a new book to explain what we know about the continent's remarkable genetic diversity, showcase on-the-ground research being carried out today and set out a roadmap for future discoveries.

Genomics in Africa is designed to support the next generation of African scientists, providing vital information for researchers, funders and policymakers working to advance genomics and precision medicine in the continent.

It is co-edited by Professor Segun Fatumo of Queen Mary University of London, and Dr Tinashe Chikowore of Harvard Medical School, who believe there is an urgent need to invest in African genomics research.

Professor Fatumo says: "Genetic research on the African continent could create more effective precision medicine, improve global drug development, and help scientists understand everything from our evolutionary history to why some people develop particular diseases."

Why is Africa so genetically diverse?

While there is a lot we don't know about human evolution and background, it is accepted that early *Homo sapiens* evolved in Africa approximately 300,000 years ago. Over millennia, we spread across the continent and settled all over it.

At some point between 50,000 and 70,000 years ago, a small band of people left Africa. This might have been a single tribe or group of tribes, and it likely had only a few thousand people of childbearing age.

There have been other migrations, but this particular one – known as the Southern Route or Southern Dispersal – is believed to be the founder population which colonised the rest of the world and interbred with archaic humans such as Neanderthals and Denisovans.

The small size of this founder population means all modern humans with ancestry outside Africa

share a high degree of genetic similarity.

While we can't know exactly how many people in total were living on the African continent at that time, we do know that the population the group left was much larger and much more genetically diverse. This means many gene variants not present in the handful of people who left Africa have been passed down and are thriving in people of African genetic heritage.

What would more research tell us?

Ambroise Wonkam, Professor of Medical Genetics at Johns Hopkins School of Medicine and current President of the African Society of Human Genetics said of the work: "Increasing the number of sequenced African genomes will refine tools for predicting disease-related variants, improve diagnoses, and enhance precision medicine.

"Ultimately, investing in African genomics promotes both scientific innovation and health equity worldwide."

Professor Segun Fatumo and Dr Tinashe Chikowore suggest that further research into African genomics could also develop and improve treatments of inherited diseases which predominantly affect African populations, such as sickle cell disease.

African researchers are currently exploring research into infectious diseases, cardiometabolic traits, cancer, mental health traits and medicine sensitivities – but the authors argue more investment both in research and infrastructure is needed to unlock the potential of these studies.

They propose a ten-year strategic roadmap for research leaders, funders, and governments planning future genomic initiatives on the continent.

"Research into these areas is essential to ensure that people with African genetic heritage can experience the full benefit of precision medicine and gene therapies," Professor Fatumo explains. "Genetic variation could mean they react differently to treatments or medicines developed for the population outside Africa, so for true health equity there is an urgent case to expand on current scientific knowledge."

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