



**H3Africa**  
Human Heredity & Health in Africa

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## **Millions of new variants in African genomes discovered**

An estimated three million novel genetic variants in over 300 genomes have been discovered through a new, large-scale African collaboration between researchers from the Human Heredity and Health in Africa (H3Africa) consortium, which includes academics from the University of Cape Town (UCT).

The findings, published in the prestigious journal [Nature](#), are paving the way for more broadly representative and relevant studies ranging from basic through clinical genetics.

The study, titled "High-depth African genomes inform human migration and health" is a move towards remedying the lack of representation of populations that are not usually the subject of much coverage. These factors are significant as they enable a more detailed study of the genetic landscape of African populations, which are very diverse.

"We need a better understanding of Africans to enable research on the genetic basis of diseases in these populations," said Professor Nicola Mulder, head of the computational biology division in the Department of Integrative Biomedical Sciences in UCT's Faculty of Health Sciences.

Whole genome sequencing of 426 individuals from 13 African countries was carried out, of which 314 were analysed at high depth. This allowed the researchers to examine rare genetic variants in an accurate and quantifiable way.

Together with collaborators, Mulder's team applied expertise in the processing of large-scale human genome sequence data and the analysis and interpretation of variants. Their findings indicated extensive genomic diversity among these genomes, even within countries and regions, with unique variants identified in each ethnolinguistic group.

"This reflects the long history and rich genomic diversity across Africa, and suggests enormous scope for identifying novel variants with additional sequencing," said Mulder.

According to Associate Professor Emile Chimusa, from the Division of Human Genetics who worked on the selection section of the paper, the study shows that functionally important genetic variants associated with physiological traits are highly relevant to infectious diseases, and geographically restricted to the local adaptation of a particular lifestyle or environment.

"These findings are extremely informative for understanding the frequency of many genetic traits and their links to infectious disease, including those that cause disease susceptibility or resistance among African populations and populations of recent African descent," he said.

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, their data 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.

The researchers found more than 100 areas of the genome with evidence of being under natural selection. A sizable proportion of these regions were associated with genes related to immunity. 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.

***ENDS***

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