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Africa's people must be able to write their own genomics agenda

Genomics on the continent is finally getting the attention it deserves — but funding needs to come from more diverse sources.

An extensive study of genomic data from people in Africa has revealed more than three million previously undescribed genetic variants, many of them in populations whose DNA had never been sampled before.

The study is a milestone in genomics research. It begins to fill a gaping hole in the world's DNA databases, which have until now contained little data from Africa's people. At the same time, most of the study's authors are based at institutions in Africa. Both of these facts are to be welcomed.

The gap in the data should not be so wide. Africa, the most genetically diverse continent, is where modern humans, *Homo sapiens*, originated. Populations on other continents are descended from groups that migrated out of Africa many tens of thousands of years ago. These groups represent a fraction of the genetic diversity found on the African continent, and yet dominate studies of DNA — often with damaging consequences.

For many years, for example, researchers erroneously thought that the genetic disorder cystic fibrosis, which severely shortens life, was not present in African populations[1]. As a result, diagnostic kits looked only for genetic variants found in European populations, missing many other variants found in African populations.

On page 741 of this issue, researchers in the Human Heredity and Health in Africa (H3Africa) Consortium report the whole genome sequences of 426 people across 50 ethnolinguistic groups — those unified by both language and ethnicity — across Africa[2].

In addition to new genetic variants, the researchers found 62 new chromosomal locations that are under strong selection, meaning that they are currently evolving. Many are involved in viral immunity, DNA repair and metabolism, so could have applications in treating disease. The DNA also contains evidence of past migrations, as well as mixing between populations.

The study is a major step forwards for African genomics, which has long suffered from what study co-author Neil Hanchard, a geneticist at Baylor College of Medicine in Houston, Texas, calls "academic colonialism" — a reference to the fact that most past studies have been led by researchers and funding agencies based in the Northern Hemisphere.

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The consequences of this imbalance were highlighted in a 2011 study[3] led by Ambroise Wonkam, a medical geneticist at the University of Cape Town, South Africa. The team identified 50 genetic studies of people in Cameroon that were published between 1989 and 2009. They found that only 28% of the authors were in Cameroon. Moreover, the studies focused mainly on questions that were of interest to researchers — such as migration patterns — rather than those that might benefit the population. There was little research on public-health concerns, and almost all of the samples were banked outside Africa — meaning that the ultimate decisions about their use were being taken by non-Africans.

Alongside this are issues surrounding consent. People who gave their DNA for research studies were often not provided with enough information to fully understand — nor given much of a say in — how their data would be used, leading to a breakdown in trust between researchers and participants.

In recent years, individuals and organizations in Africa have been pushing back. Most dramatically, in 2017, the San people of South Africa issued their own code of research ethics. Partly as a result of such efforts, 23 of the 32 named authors on the latest study are at institutions in Africa, including South Africa, Egypt, Tunisia, Uganda, Nigeria and Morocco.

"A big thing is to change the narrative; to have this come from Africa," another co-author, Zané Lombard, a senior researcher in human genetics at the University of the Witwatersrand in Johannesburg, South Africa, told *Nature*.

The H3Africa Consortium comprises a US$150-million, 10-year initiative that supports institutes in 12 African countries. With the findings of the current paper, researchers have taken an important step in showing the potential of African genomics. The consortium's donors — the US National Institutes of Health (NIH) and the UK biomedical charity Wellcome — have also helped to empower a new generation of researchers.

This time last year, NIH director Francis Collins told a meeting of international research donors in Addis Ababa that the agency pledged to ensure African ownership of the research it funds on the continent. That's a crucial aim. But ownership also needs Africa's own funders — especially philanthropists and corporations — to create new funding opportunities for researchers, so that more research questions, priorities and intended outcomes can be framed locally, nationally and regionally.

African genomics holds significant potential for advancing research and improving understanding and treatment of diseases on the continent and beyond, given humans' shared ancestry. For that potential to be fully realized, a true partnership will be required, in which research funding comes from appropriate sources in Africa and from around the world.

1. Stewart, C. & Pepper, M. S. *Genet. Med.* **18**, 653–662 (2016).
2. Choudhury, A. *et al. Nature* **586**, 741–748 (2020).
3. Wonkam, A., Azabji Kenfack, M., Muna, W. F. T. & Ouwe-Missi-Oukem-Boyer, O. *Dev. World Bioeth.* **11**, 120–127 (2011).