Understanding Africa's diverse gene pool can help fight lifestyle diseases

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Credit: AI-generated image (disclaimer)

Africa is home to about 16% of the world's population. That's 1.2 billion people. But the continent is disproportionately burdened by a double health challenge: infectious diseases and a recent increase in non-communicable diseases.

Non-communicable diseases such as hypertension, cardiovascular diseases and diabetes are on the march due to an ageing population, a transition to increased urbanisation, dietary changes, a more sedentary lifestyle and an increase in the prevalence of obesity.

Non-communicable diseases result in deaths everywhere in the world. But in Africa they are also a major reason for premature deaths, that is people dying between the ages of 40 and 70. In South Africa there is more than a 25% chance of dying prematurely from non-communicable diseases. On the rest of the continent it ranges between 15% and 24%. This compares to the average of less than 15% for the US and Europe.

The continent's health systems are struggling to bring these diseases under control. One of the key strategies explored elsewhere is the use of genomics for a precision medicine approach. This opens the door to understanding which genetic drivers are responsible for an increased risk to a particular disease and how genetic variants in a population dictate responses to treatment.

Once scientists understand which treatments have the largest impact they can target therapy accordingly, this known as precision public health.

This approach could help to alleviate the health burden in Africa too but implementing it is more difficult than elsewhere. This is because the continent has added challenges. It has a genomic spectrum that is more diverse than other continents. In addition it has a wide range of different environments, cultures and levels of poverty.

That's not to say it's impossible. A precision public health approach would be possible if it was driven by research at a population level with large cohorts. This could help scientists understand how genes respond in the presence of certain environments, and interact with them (known as gene-environment interactions). Cracking this would open a new frontier

in the drive against rising non-communicable diseases.

Genomic research challenges

There are four main problems with advancing genomic research in Africa.

Firstly, there is sparse data on genomics and gene-environment interactions in African populations. Scientists still do not know how populations with a particular genetic variant spectrum react to changes in the environment, such as an increase in poverty or lifestyle change during urbanisation, and what the likely impact of a particular genetic variants is.

In addition, scientists are prone to using interpretations based on research conducted elsewhere. There's a particular bias, for example, to apply Eurocentric interpretations. In fact, people's genetic background could have a profound effect on the way people react to their environment and to treatments. Applying a Eurocentric approach therefore doesn't make sense. For example, sickle cell disease would not be very relevant in a European setting, but is very common in many regions of Africa and causes an enormous disease burden.

The second challenge is around the regulatory framework and how good practice guidelines are implemented. In many African countries privacy and genetic information is not protected or legislated. There is therefore the potential for harm.

Thirdly, there is a lack of resources to conduct primary research to inform precision public health approaches. These include money, people, infrastructure and electronic public health records. All are critical.

Implementing a precision public health approach is costly and it needs to be reviewed and updated continuously as understanding deepens and the environments that people live in change.

The fourth challenge is around informing people about the approach and what's involved. Without this there is unlikely to be any buy in.

First steps

Genomic research has gained considerable momentum on the continent over the past decade. Two initiatives are boosting the capacity for genomic research on African populations. These are expected to benefit health initiatives elsewhere in the world too.

The International Network for the Demographic Evaluation of Populations and Their Health (INDEPTH) does two things: it collects data on populations. In addition it has launched a new initiative to collect biological specimens from populations. On the basis of this the project, known as CHESS, can provide data on diseases, pathogens and causes of death in specific populations.

The second initiative, the Human Heredity and Health in Africa (H3Africa) Consortium, studies infectious and non-communicable diseases from a genomics point of view.

These initiatives are important because they are studying populations that have been under-represented.

Longterm goals

There are several examples for successful use of precision medicine in the developed world (for example in some cancers).

Before Africa can boast its own examples it will first need to generate knowledge and data. This will take time which means that a precision public health approach to tackle disease won't be yielding immediate results.

Many people on the continent do not get the treatment they need. In the short-term genomic research on drug responses could make a difference by providing governments with guidelines for what effective medication they should be giving their populations.

For longer term impact, researchers need to understand how genetic predisposition works in Africa. Only then will we begin to know how to treat the diseases more effectively.

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