

## **Southern African Human Genome Programme: A Programme for Southern Africa by Southern Africans**

In 2011, the ground-breaking initiative of the Southern African Human Genome Project (SAHGP) kicked off with the support from the Department of Science and Technology (DST). The project aims to understand the genetic variation in southern Africans and how this translates into understanding our susceptibility to disease, and developing more effective methods for disease diagnosis and treatment in southern Africans.

### Why look at Southern Africa genetic diversity?

Up to now, scientific studies have shown that great variation exists in the genetic makeup of southern African populations, to the extent that scientists believe southern Africa may be the location of the origin of modern human populations. The genetic makeup of indigenous southern African populations has been shown to differ significantly from European, Asian and West African populations.

It is not only large variation in indigenous populations that should be considered when one looks at the genetic makeup of southern Africans, but also the relatively smaller differences between more similar population groups. For example, it is apparent that there is a unique mutation spectrum in the BRCA1 and BRCA2 genes for breast cancer patients in the Afrikaner population, which differs from that in European patients.

Up to now, southern Africans have been poorly represented in studies that look at genetic susceptibility to disease, as well as in drug trials where the success of a drug may be dependant on genetic makeup of the participants in the trial. Some diagnostic tests for genetic predisposition to diseases that have been developed based on this type of research may not be accurate in southern African populations as they are based largely on variants that have been detected in Caucasian and Asian populations. Some drugs that are on the market may not work effectively in southern Africans. It is important therefore to have a database of the genetic variation in these populations as a resource to apply to these studies.

### The purpose of SAHGP

The vision of the SAHGP requires the development of skills and infrastructure in genomic research at a national level, as well as a coordinated effort to be able to manage the quantity of information. Skills are needed to gather genetic information and to analyse the enormous volumes of data that will be generated. Research efforts will be pooled through the programme through collaboration between scientists at Higher Education Institutions, science councils,

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government and industry.

The programme acknowledges a responsibility to the concept of “Genomic sovereignty”, in which the ownership, planning and execution of the project and process remains in southern Africa, so that the genetic resources remain in southern Africa and benefit can be shared with the people of the region, including the communities from which the genetic material is sourced.

The first phase of the Programme is aimed at bringing together all the relevant stakeholders at a national level, in order to formulate a well-represented strategy for the way forward. The first stakeholder meeting was held at the end of January this year and produced some significant outcomes, in which consensus was reached on the relevance and importance of the programme and its need to be retained in southern Africa through capacity development to the benefit of southern African populations.

The first phase of the programme is coordinated jointly by Prof Michael Pepper and Prof Michele Ramsay, who can be contacted for further details about the programme on [michael.pepper@up.ac.za](mailto:michael.pepper@up.ac.za) and [michele.ramsay@nhls.ac.za](mailto:michele.ramsay@nhls.ac.za).