APPRECIATING THE RAINBOW NATION'S GENETIC DIVERSITY

By: Galen Wright, Stellenbosch University

African soil – the home of diverse cultures, remarkable wildlife and alluring mineral wealth – has intrigued foreign nations for centuries. This year sees the eyes' of the world converge on the African continent once again, with the 2010 FIFA World Cup reaching our shores in June. South Africans, members of the Rainbow Nation, are eager to capitalise on the exposure that will hopefully boost our economy. There is, however, another untapped national treasure that could be used to benefit the South African community in the future, if utilised in the correct manner. This asset is tangled up in our bodies, locked tightly within the cells of South Africa's eclectic citizens and is known as genetic diversity. It is what makes everyone of us unique and by studying such information, overseas and local researchers wish to determine the biological basis of numerous diseases, provide optimal drug therapy and other intrinsic characteristics. Such findings have obvious commercial value and it is therefore imperative that we manage this wealth of genetic information correctly.

Our genomes are approximately 99% alike, yet genetic diversity exists within and between populations and is shaped by a variety of different mechanisms. Over time, heritable mutations accumulate in the DNA of populations and are the source of genetic variation, contributing towards observable traits. This natural process occurs in all organisms, from bacteria to humans, and although the term "mutation" usually has negative connotations; the effects are typically not detrimental and can occasionally have positive consequences. The migration patterns of modern humans have influenced genetic diversity, such that when a small group of individuals leaves a particular region, the new founder population will lose genetic diversity. Moreover, genetic diversity is lost when the death rate of a population increases during pandemics. The process of natural selection also plays a role, causing the enrichment of genetic traits that are favourable for survival in a particular region. Together, these mechanisms have formed the genetic makeup of populations over millennia, yet molecular biologists have only acquired the tools to efficiently study this in the last decades.

Long before extensive paleontological and DNA evidence, one of the most eminent figures in biological research, Charles Darwin, wrote in his work, *The Descent of Man* (1871): "...it is

somewhat more probable that our early progenitors lived on the African continent than elsewhere". The findings of recent research have confirmed that Africa was the home of early humans and is therefore the ultimate source of *Homo sapiens* genetic variation. South Africa is no exception, with 11 official languages testifying to our remarkable assortment of population groups. To date, the most comprehensive research involving the genetic structure of African populations was published last year in *Science*. Tishkoff and co-workers reported that Southern African populations, especially the ancient Khoisan population, contained a large number of mutations absent from non-African ones. It was also shown that Southern African Khoisan population contributed to the gene pool of the Xhosa population significantly, which is linguistically evident through their click constants. High levels of genetic contributions from populations from various continents were detected in the Cape Mixed Ancestry population. These findings illustrate that DNA evidence can be used to elucidate the history of African populations.

The intricate genetic diversity observed in South Africa should be celebrated in the same way that we appreciate our cultural diversity. Our country presents a unique set of challenges to geneticists, yet also offers the potential for tremendous rewards. Currently, genetic diagnostic tests designed in non-African populations have limited use in South Africa, as they do not account for genetic variation that is unique to Africa. Studying African populations could therefore facilitate in the identification of the majority of genetic risk factors for disease. A recent report by PricewaterhouseCoopers estimated that the market for personalised medicine, where an individual's genetic profile is taken into account, will grow to over $450 billion in the US alone by 2015, emphasising the importance of such research. These technologies would be extremely beneficial in sub-Saharan Africa, where diseases such as HIV/Aids, tuberculosis and malaria are major concerns. Mining the genomes of the Rainbow Nation will therefore uncover a wealth of genetic information, enabling this national treasure to benefit the South Africa population.