

GENETICS: OUR PAST AND OUR FUTURE

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In his book "What is Life?" the great physicist Erwin Schrödinger pronounced that the study of a humble locust is more complex than to study a whole galaxy. Fortunately, Charles Darwin provided us with an explanation for the origin of this complexity. His theory explains how simple laws generate complexity; how random variation combined with natural selection brings about complex life forms and structures. In 2009, the world celebrated the 200th anniversary of Darwin's birth. I can only imagine the excitement he would have experienced if he'd lived to witness the state of modern biology. The fields of genetics and molecular biology have expanded at such rates that today almost all approaches in biology revolve around these fields and they perhaps offer some of the best evidence for evolution.

Studies of human population genetics provide us with insights into human migration and history. It is truly astounding how much can be learnt about pre-history from modern DNA. Preserved within our DNA is an accurate record of great detail. When we compare our DNA to the DNA of our cousin species on this planet, we discover extraordinary details of our past. We, for example, possess dozens of damaged genes that mice use for scent. We can recognise these genes in our genome by comparison with mouse genomes; however, sometime during mankind's evolution these genes became unnecessary for our survival and have subsequently become corrupted like a damaged computer file.

In 2003 the entire human genome was sequenced and this major effort has taught us much about our species and it has also inspired numerous technological developments. The goal of human geneticists today is to map the extent of human genetic variation and many of these technological marvels are being put to good use to achieve this. The goal is not simply academic but rather geneticists intend to discover the variants which contribute towards human diseases and hopefully to use these discoveries to better advise medical treatments ultimately aiming for better and more personalised drug development. In the last few years large international studies have been completed which link common variants in the genome to common traits; from human height and bodyweight to susceptibility to diabetes, heart disease, mental disorders and a number of infectious diseases.

So where does this leave us in Africa? Long before descendants of man were found, Darwin postulated that man originated in Africa. We now know that he was correct and the consequence of this fact for geneticists is that people of African descent display greater genetic variability than those who underwent the exodus from the mother continent. This is

due to the effect of genetic isolation on the people who left Africa and is in fact good evidence for the “Out of Africa” theory. Unfortunately, this makes the job of identifying disease causing variants in African populations more tricky and the international scientific community has as yet not sufficiently accounted for this variability. Current projects into human variability use only a few families from the Yoruban region of Nigeria to represent African people, yet most South African geneticists consider this to be insufficient to account for African genetic variability. If we are to identify the genetic variants associated with local health problems then South African scientists need to intensively pursue genome-wide studies of variation.

To date, a few genome-wide studies into HIV, malaria and tuberculosis susceptibility have been performed. But for the most part the technological processing of these studies has been internationally out-sourced despite the fact that within South Africa we possess the capability of performing genome-wide scans for variability and even for sequencing of whole genomes. If more genetic variants are to be discovered ideally these studies should be expanded upon and South African scientists should play a greater part in future studies. For the most part large genomic studies have not been performed in South Africa due to a paucity of funding and expertise. Both situations could be remedied if South African policy makers take a leaf out of President Obama’s book by heavily investing in science.

Scientists in Africa should consider themselves fortunate to have ready access to so many biological wonders; from human diversity, to our fauna and flora and to our critical place in human evolution. Let us hope that the mysteries of Africa will be uncovered by Africans.