

Incorporating Clinical Genetics in Routine Patient Care

"One general law, leading to the advancement of all organic beings, namely, multiply, vary, let the strongest live." Charles Darwin, The Origin of Species 1859 (1)

The discovery of the double helical deoxyribonucleic acid (DNA) structure by Watson, Crick & Wilson, for which they received the Nobel Prize for Physiology/Medicine in 1962, heralded the start of unravelling the mysteries of the human genome. The genome of an organism is responsible for encoding of all information necessary for its existence and propagation of life. All living organisms on earth can trace their origin to 3.8 billion years ago, when our universal ancestor resided in water (2). Subsequent evolutionary changes led to diversification of the phylogenetic tree with the emergence of new and distinct species. It is due to having common ancestry that humans share 96% and 40% of the genetic material with chimpanzees and bananas respectively (3). Human species are 99.9% identical, with the least diversity observed among monozygotic twins.

In order to decode the human genome, the National Institutes of Health undertook the Human Genome Project (HGP) and mapped out the entire nucleotide sequence consisting of 3 Billion base pairs contained in 23 chromosomes in the nucleus and mitochondria in the cytoplasm. Of these, only 1% of the human genome encodes for proteins, with the other 99% not yet fully understood. The human genome encodes for approximately 20,000 proteins, which averages 1,000 proteins per chromosome. The HGP formed the basis of understanding human genetics and its role in physiological and disease states. Subsequent advances in medical and population genetics have led to the discovery of genetic variants and their relationship to physiologic and disease states (4). Initial HGPs had little representation of Africans residing in Africa and there are ongoing efforts to decipher the genome of the African.

With the subsequent advancement of genetics, medical genetics has evolved from a purely lab based discipline to an established subspecialty in internal

medicine, paediatrics, pathology and fetomaternal medicine. Medical genetics has immense potential for altering medical diagnostics for patients and the at risk relatives, treatment options and reproductive choices. This has been most evident in oncology where genetic testing is used in tailoring patient treatment as per somatic genetic mutations identified.

With the emergent good molecular laboratory infrastructure, and current ongoing works in medical and population genetics in Kenya, there exists a tremendous unmet need for routine genetic counselling and testing in everyday clinical practice. With the ever decreasing costs, genetic test requisition may become as common place as a haemogram.

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