

Are you ready for genomic medicine?



In October 1989, the much-publicised Human Genome Project was initiated. The main aims of the project were to chart the complete human genome by determining the sequences of the three billion chemical base pairs that make up human DNA and identify all genes in human DNA.

This critical information would be

stored in databases readily available to researchers, and the tools for analysing such data would be improved in order to facilitate and develop further research in the field of genetics. Key technology developed through this United States Federal Government-funded project would also be licensed to the private sector in order to kick-start genetics-related industries. It is common knowledge that the project was finished ahead of time in 2003, due to some key technological advances, coinciding with the 50th anniversary of Watson and Crick's description of the fundamental structure of DNA.

I can remember, as a medical student during the late 1970s, that very little attention was given to genetics in our teachings. We learned about DNA and genetic conditions like Down's syndrome and a few others, but genetics was mostly regarded by us medical students as some esoteric academic speciality and of very little value to pass our exams (alas the hidden curriculum). Will I ever forget the pea experiments of Mendel? But how have things changed! Today, we are learning a completely new language. "Genetics" is the study of single genes and their effects, but "genomics" is the study, not just of single genes, but of the functions and interactions of all the genes in the genome. Common conditions, such as breast cancer, colorectal cancer and HIV infection, apparently display a complex interaction between many genes and certain environmental factors.¹

The present growth of this field is astounding. It is estimated that the human genome includes approximately 30 000 to 35 000 genes. One can already have your full genome (DNA sequence) charted for \$10 000, and the aim is to bring the cost down to less than \$1 000 within five years. Interestingly, Emeritus Archbishop Desmond Tutu is the first South African whose full genome has been charted. Every week, a number of new disease-linked genes are discovered. The current known disease-linked gene pool is already more than

2 850. Even the Neanderthal genome has been charted and, according to preliminary sequences, 99.7% of the base pairs of the modern human and Neanderthal genomes are probably identical.

The field of genetics holds much promise to improve health and alleviate human suffering. Despite all these achievements and discoveries, and perhaps a good dose of journalistic hyperbole, medicine as we practise it today is unlikely to be transformed radically in the short to medium term. Some people, and particularly our patients reading the newspapers and magazines, are expecting miracles, such as "the cure for cancer". However, the full potential of a DNA-based transformation of medicine will only be realised gradually over the course of decades to come, as researchers unravel the content of our genomes and, most important, the physiological consequences of variations in their sequence.

Are we family doctors ready for this new era of genomic medicine? First, one would urge medical schools and family medicine departments to look critically at the curriculum to make sure that new generations of doctors will be well prepared to understand and deal with the practical and clinical implications of genomics. All physicians will soon need to understand the concept of genetic variability, its interactions with the environment, and its implications for patient care. Practising physicians should attempt to learn about the genes and genomes that will progressively change medical practice as we know it. The practice of medicine has already entered an era in which the individual patient's genome will help to determine the optimal approach to care, whether it is preventive, diagnostic, or therapeutic.

There are just not enough genetic counsellors out there, and doctors and nurses will have to play a major part for many years to come. Perhaps our main role, at present, would be to assist our patients to understand and interpret the information they get from the popular media. We can also warn and help protect them against quackery and false expectations.

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Reference

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