Editorial - Genes and the Future of Medicine

Clinicians lead busy lives and there is very little time outside of one's medical duties to devote to the understanding of a complex science such as molecular biology. However, the intrinsic role that genetics play in the vast majority of diseases and the explosion in genetic knowledge means that molecular biology is another discipline in which doctors must quickly become experts.

The stimulus for the increasing interest in the genetic basis of disease was the recent completion of a 'working draft' of the Human Genome Project (HGP), announced on the 26th of June 2000. Currently thousands of scientists are working worldwide on genetic material obtained from 50 individuals of both sexes and from several ethnicities, to characterise the 3 billion base pairs that comprise the human genome.

Completion of the sequence data was originally projected for 2005, but the project is remarkable for being both ahead of schedule, and running below budget. In view of the progress, the aim of the project has been changed to target the completion of a tenfold coverage of the entire genome by 2003, and the production of a database of the most common sequence variations that distinguish one person from another. Eventually the objective is to characterise all of the genetic differences for human disease.

Human disease is currently defined according to the clinical appearance rather than by the basic mechanism of disease. Soon knowledge of the human genome will provide the opportunity to define diseases instead by their molecular mechanism. In other words, it will change medicine from using a phenotypic classification of disease, to a genotypic classification. For example, asthma presently is described as 'a reversible narrowing of the bronchial airways', as opposed to, a possible future definition of 'a respiratory disease characterised by the substitution of valine for leucine on gene 322, combined with a deficiency of gene 7003'.

Molecular genetics in medicine is not a new concept. It has already been applied to identify single gene disorders such a Cystic Fibrosis and Polycystic Kidney Disease. The reason for the excitement associated with the HGP is because of the potential to elucidate the genetic basis of common diseases and multifactorial disorders.

Many scientists and cynics when asked why the human genome is being characterised may reply using the immortal words of Mallory when asked why he climbed Mt Everest; "Because its there!". However, in addition to the great scientific milestone and organisational achievement, there will undoubtedly be benefits for society and the individual.

Benefits that a greater knowledge of human genetics will bring include organogenesis. Tissues or organs which have failed may be replaced by substitutes grown *in vitro* using an individual's DNA, thus solving the current problems facing organ transplantation of organ availability, immunosuppression, and organ rejection. Thus in the future, there may be true harvesting of organs, although, the moral and ethical issues may restrict progress in human experimentation.

The concept and importance of pharmacogenetics has already been integrated into the undergraduate pharmacology course. However, a greater knowledge of the genetic basis for response to drugs will enable prediction of adverse reactions, as well as optimisation of pharmacotherapy for individuals. With the greater insight into gene function, it has been predicted that the 500 drugs currently used in clinical practice will increase to 3000 over the next 10 to 15 years, as drugs gain a greater specificity of function.

The most exciting aspect of the HGP is the unforeseen discoveries it may yield. People have postulated the potential benefits for the past 10 years but it is quite likely that there are some advances that have not even been imagined yet.

It is interesting to consider a few statistics generated by the HGP data. Firstly, we are all 99.8% the same, with only 2 in every 1000 base pairs varying between individuals. Secondly, 98% of the human genetic code is identical to that of the chimpanzee (and perhaps even higher in some of us). Lastly, to date 97% of the human genome has no known function, and it is in this last point that we hope to make some progress in the near future.

Undoubtedly the genetic information provided by the HGP is going to revolutionise medicine. Dr Collins, the Director of the National Human Genome Institute in the United States stated, "Virtually all disease, except some trauma, has a hereditary component". The integration of the genetic basis of disease with modern imaging techniques and traditional clinical skills will be a potent combination. In seeking excellence in clinical practice, it should still be remembered that we share 98% of the gene sequence of chimpanzees.

There are no perfect genetic specimens, only individuals.

Following the very professional production of The Trinity Student Medical Journal, Volume 1 in 2000, I am extremely honoured to be the Editor-in-Chief for the 2001 edition. It has been a thoroughly enjoyable experience reading the contributions and working with my fellow committee members. The TSMJ Committee have been enthusiastic, encouraging and supportive. Thank you very sincerely to all of the authors who contributed articles, and to all of the TSMJ Committee Members, particularly Sonia, John, Pete, Barbara, Ciaran, Myto and Deirdre. Very importantly I would like to thank Professors O'Brien and Feighery for their continued support.