Personalized medicine in the era of the personal genome

Personalized medicine as a field was developed with the purpose of relating the chances of a particular SNP being associated with a particular disease and/or drug reaction. In fact, an industry has been generated whereby, for a fee, SNPs are determined and risks of particular diseases are given to those who supply a sample of DNA. There have been concerns regarding the value of this to individuals. However, a new dawn in personalized medicine is coming, owing to the falling prices of sequencing and, ultimately, the US\$1000 or \$100 genome.

In the foreseeable future, everyone will have their genome (or exome) sequenced on a chip, and they will be able to read it gene by gene. There are two major outcomes besides the clear industrial activity:

The first is that individuals or their advisor (or a computer) will be able to look through the genes for variations. When one is found, for example, in the colon cancer gene, the individual will want to know if it is harmful or not. In order to obtain an answer, a complete catalogue of mutations and their effects will be needed.

The second is that when two individuals are to be married and plan to have children, they will be able to put their genome chips into a computer and with reference to the complete catalog of mutations and their effects, they might be told by the computer that "...you both have a serious defect in (for example) the maple syrup urine disease gene, we suggest you consult a genetic counselor". This activity has a precedent in the globin gene causing thalassemia in the Mediterranean area, particularly in Cyprus, where premarital testing is carried out to alert a couple of the chance of potentially having an affected child. This has resulted in the almost complete elimination of this disease in the area. Extension of this to the whole genome promises to eliminate a considerable amount of human suffering.

The promise of this strategy underlines the importance of collecting all mutations in all the genes that cause disease. This is the objective of the Human Variome Project [101]. This project was initiated at a meeting of world experts and representatives of the United Nations Educational, Scientific and Cultural Organization, European Commission, WHO and the Organisation for Economic Co-operation and Development in 2006 [1,2], and specific projects and integration of activities will be developed at a meeting in Paris, France, taking place on 10–14 May 2010 [102].

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Richard GH Cotton^{1,2,3}

¹Human Variome Project

²Genomic Disorders Research Centre,
Florey Neuroscience Institutes,
Carlton South, VIC 3053, Australia

³Faculty of Medicine, Dentistry & Health Sciences,
University of Melbourne, Parkville, VIC 3010, Australia
Tel.: +61 392 882 980
Fax: +61 392 882 989
cotton@unimelb.edu.au