

USEFUL TERMS

Source: National Health and Medical Research Council. (2010). *Medical Genetic Testing: information for health professionals*. (E99).

<http://www.nhmrc.gov.au/guidelines/publications/e99>.

Genetic material

Any source of DNA or RNA that can be tested to obtain genetic information. This includes cells, whether as single cells, or as part of tissues, and extracted DNA and RNA.

Genetic test

A genetic test may be performed using DNA, RNA or protein (the 'gene product'), or involve measurement of a substance that indirectly reflects gene function or by analysing chromosomes. A genetic test reveals genetic information.

Genomics

The genome is the complete genetic material in an organism. Hence, genomics is the study of the structure of the genome including its DNA sequence. In contrast to genetics, which predominantly focuses on one gene – one disease, genomic-based technologies allow many (even hundreds or thousands) genes or gene markers to be assessed simultaneously.

Personalised medicine

The US President's Council of Advisors on Science and Technology in its Sept 2008 report *Priorities for Personalized Medicine* defines personalised medicine as: "The ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease or their response to a specific treatment. Preventative or therapeutic interventions can then be concentrated on those who will benefit, sparing expense and side effects for those who will not."

Pharmacogenomics

The study of variations of DNA and RNA characteristics as related to drug response i.e. the focus is the study of multiple genes and/or their function.

Variant

Changes in the DNA sequence (usually single base changes) are called variants.

Whole genome sequencing

Refers to methods that allow the whole DNA sequence of an organism to be identified. In people, this will require the sequencing of about 3 billion 'bases' (A, T, C and G).

Epigenetics

The study of heritable changes in gene function that occur without a change in the DNA sequence.