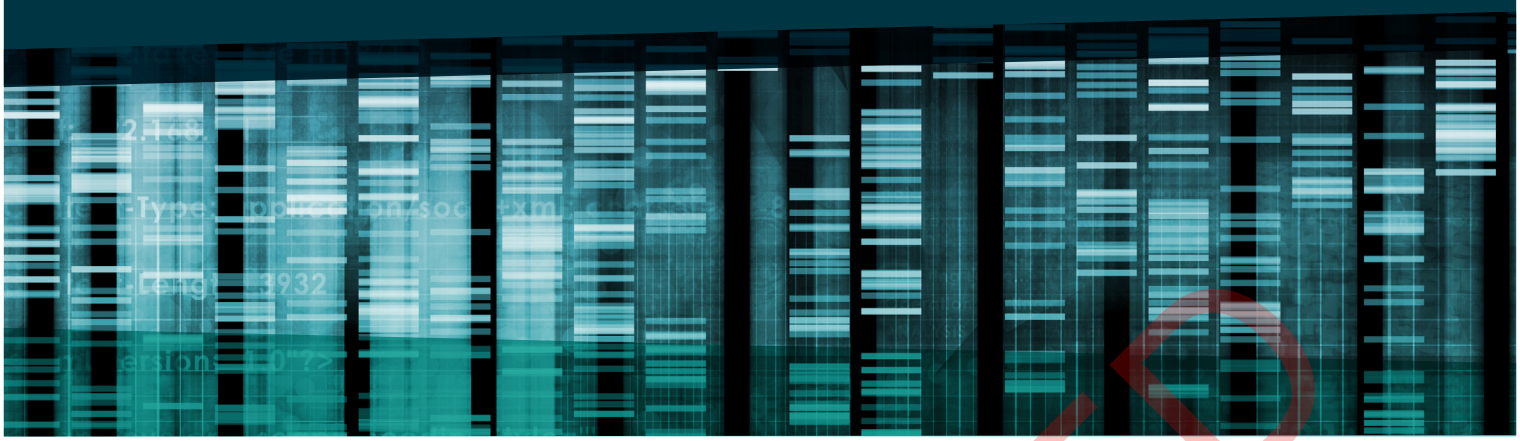
Australian Government
National Health and Medical Research Council



Personalised medicine and genetics

Many of us wonder why some people develop cancer while others do not and why a medication might work well for one person but be less effective or cause serious side effects in another. Although these differences are due to a number of important factors such as age, weight and lifestyle, our genetic make-up also plays a part. Using information about genetic predisposition to disease is a key part of personalised medicine.

What is meant by the term 'personalised medicine'?

The way genes interact with each other can sometimes predispose us to developing particular diseases. Scientists have identified specific links between genes and some diseases as well as between genes and the effectiveness of some medicines or treatments.

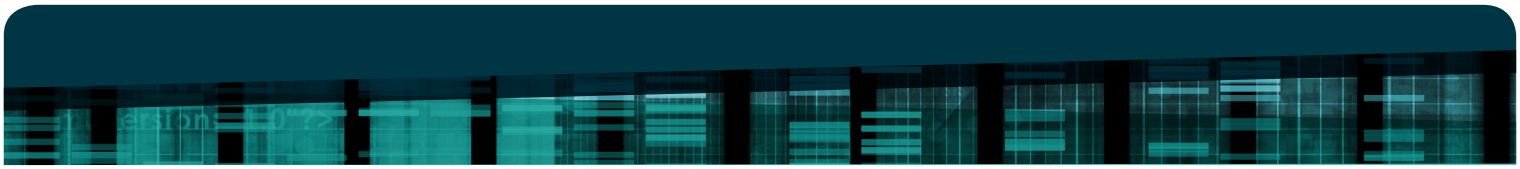
'Personalised medicine' (also known as stratified or precision medicine) uses this knowledge of genetics to predict disease development, to influence decisions about lifestyle choices or to tailor treatment to an individual.

Appropriate application of personalised medicine would be expected to result in better disease prevention and more accurate diagnosis of disease. Personalised medicine could also use knowledge of the way specific genes work with medicines to tailor more effective treatment of disease for each individual.

What impact does personalised medicine have on disease diagnosis?

In the past, disease diagnosis was based on symptoms that might be indicative of several diseases. Nowadays, diagnosis of some diseases has become more accurate because we are able to test for genes known to be associated with the disease. This method not only clearly identifies the presence of a particular disease; it can also precisely determine the subtype of the disease.

An example of this approach is identifying specific subtypes of acute promyelocytic leukaemia. Once the acute promyelocytic leukaemia subtype is identified by DNA testing, doctors can prescribe a treatment that specifically targets that subtype.



How can genetic information be used in disease prevention?

Individual genetic variation could increase the risk of an individual developing a particular disease or could determine how a person's body is likely to be affected by a disease. For example, a person's genetic make-up could indicate an increased risk of developing diabetes or heart disease.

Early identification of these variations and calculation of the risks associated with them are important in disease prevention. This knowledge could influence lifestyle choices and decisions about interventions that may prevent the disease from developing, delay disease onset or reduce the impact of the disease.

At this stage, there are a limited number of genetic tests available for this purpose but the field of personalised medicine is certain to expand rapidly in the near future.

What is pharmacogenetics?

Pharmacogenetics describes the science that explores how genetic differences can lead to differences in the way certain medicines interact with the human body. These interactions can affect both the effectiveness of the medication and any side effects.

Pharmacogenomics is another word that is used to describe personalised medicine in the context of treatment with medications. While the terms pharmacogenetics and pharmacogenomics are subtly different, they can both be used interchangeably for the purpose of this topic.

How can pharmacogenetics improve the effectiveness of medications?

At present, doctors use a 'one size fits all' approach to prescribe medicines for most diseases. Patients are first given a medication at an average dose and then the doctor makes adjustments to the treatment according to the response of the patient's body.

Applying pharmacogenetics ensures patients are prescribed the most effective drug or optimal dosage from the beginning of treatment. Pharmacogenetics can be used to minimise the likelihood of an adverse reaction to a medicine.

For example, the drug Abacavir is approved in Australia for the treatment of patients with HIV. However, approximately 5% of patients suffer a potentially fatal hypersensitive reaction to this medication. Research has found that with a particular variation of a gene people with a gene called HLA-B are more likely to develop a reaction to Abacavir. Clinicians are advised to consider testing patients for the presence of this gene variant before they prescribe Abacavir.

Another example involves testing the TPMT (thiopurine methyltransferase) gene. About 10% of people have a form of this gene which places them at risk of severe side effects following treatment with thiopurine medications such as azathioprine. These medications are used for the treatment of leukaemia, rheumatoid arthritis, and inflammatory bowel disease. The starting dose of thiopurine medications is usually reduced in patients with particular forms of the TPMT gene.