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Genetic Support Council of WA Inc

Gene Advocate

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Personalised medicine and genetics

Have you ever wondered why some people develop cancer while others don't?

Or why a medicine may work well for you, but may be less effective or cause serious side effects in someone else?

Although the differences are due to a number of important factors such as age, weight and lifestyle, our genetic makeup also plays a part.

What is meant by the term 'personalised medicine'?

The way genes interact with each other can sometimes make us more likely to develop particular diseases. Research scientists have found a number of specific links between some genes and the development of certain diseases, as well as between some genes and the effectiveness of certain medicines.

'Personalised medicine' uses this knowledge of genetics to predict disease development, to influence decisions about lifestyle choices and/or to tailor medical treatment to an individual.

The result of personalised medicine is better disease prevention, more accurate diagnosis of disease and more effective treatment of disease by understanding the way specific genes work with medicines.

What impact does 'personalised medicine' have on disease diagnosis?

In the past, disease diagnosis was based on symptoms. However, several conditions can share similar symptoms, making diagnosis difficult. Nowadays, diagnosis of some diseases has become more accurate because we are able to test for genes that are known to be associated with the disease. Genetic testing can not only identify the presence of particular diseases,.it can also determine which subtype of a disease a person has.

Knowing the subtype of a disease can help determine an accurate outcome for the patient. Identification of the disease subtype also allows doctors to prescribe a drug therapy that will specifically target the subtype through the use of pharmacogenetics.

What is pharmacogenetics and how can it improve the effectiveness of drug treatment?

At present, doctors use a 'one size fits all' approach to prescribe medicines for most

diseases. Patients are first given a medicine at an average dose. The doctor then makes adjustments to the dose of medicine according to the patient's response.

Pharmacogenetics (or pharmacogenomics) aims to match the best available drug or dose to a person, based on his or her genetic makeup. A process known as gene expression profiling identifies the active genes in a patient. Profiling can tell the doctor how a medicine may be broken down by the patient's body and whether or not it will be effective.

Applying pharmacogenetics ensures patients are prescribed the most effective drug right from the beginning. Pharmacogenetics also minimises the number of adverse reactions to medicines as it can show whether a patient is likely to have a toxic response.

As scientists gain a better understanding of genetic variations, new subtypes of diseases could be identified and more specific medicines that target the new subtypes may be manufactured.

How can genetic information be used in disease prevention?

Individual variations (mutations) that may be present in our genes can determine how our bodies are likely to be affected by a disease, or affect our risk of developing a particular disease. Some variations can be protective and decrease risks. Other variations can increase the likelihood of developing a particular disease. For example, a person's genetic makeup 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 be used to influence lifestyle choices and decisions about possible interventions that may prevent the disease from developing, to delay disease onset or to reduce the impact of the disease.

There are currently a limited number of genetic tests available for this purpose. However, the field of personalised medicine is certain to expand rapidly in the near future.

When will personalised medicine be a reality?

Although the science is still in the early stages, pharmacogenetics is already being used to treat some diseases such as breast cancer. Before being prescribed the drug Herceptin®, breast cancer patients are tested to examine whether a particular gene, human epidermal growth factor receptor 2 (HER2), is present in the cancer cells.

Herceptin® is only effective when cancer cells have extra copies of the human epidermal growth factor receptor 2 (HER2) gene, or too many HER2 receptors. Testing to find this out allows doctors to predict the patient's response to Herceptin®, enabling a more appropriate medicine to be prescribed immediately.

Source: NHMRC
http://www.nhmrc.gov.au/your-health/egenetics/personalised-medicine