

Genetics and Cancer

Genetics is an exciting field of science in the cancer area. It may have implications for the diagnosis, prognosis and treatment of cancer. However there is still a long way to go and many questions in the genetic field remain unanswered. This fact sheet provides an introduction to this topic and answers some common questions.

What have genes got to do with cancer?

Our bodies are vast collections of cells. Each cell has a nucleus which contains 23 pairs of chromosomes which we inherit from our parents. Chromosomes are made up of thousands of genes, which are the basic units of inheritance, and genes are made up of DNA (deoxyribonucleic acid). Each gene determines a single human characteristic and together they contain all the information that controls how our bodies work and how we look, for example whether we have brown or blue eyes, dark or fair hair.

Of the 30000 to 40000 genes within a human cell, only a handful are known to regulate cell growth and division. Every time a cell divides, the chromosomes also reproduce so that each cell has the same genetic information. Sometimes, a mistake can happen when the chromosomes are copied during cell division. In some cases this may be the first step that could lead to abnormal cell growth or cancer.

DNA can also be damaged by exposure to carcinogens such as ultraviolet radiation from the sun, tobacco (smoking) and asbestos.

Most cancers occur due to changes in our DNA that happen during our lifetime. This is known as a 'somatic' mutation. However, defective genes can also be inherited, or passed on, from parents to children, though only a small proportion of cancers (about 5%) occur for this reason. When mutations are passed through a family, these mutations are known as 'germline' mutations and they affect every cell in the body as they are already present when you are a single-celled embryo.

What is genetic pre-disposition?

Genetic make-up is passed on from parents to children. In families with known germline mutations, each child has a 50% chance of inheriting the cancer susceptibility mutation. However having a gene mutation does not necessarily mean that a person will develop cancer and, mostly likely, a trigger or promoter is also required for cancer to develop.

How do I know if the cancers in the family are inherited?

Cancer is a disease of ageing; therefore, older people are more likely to develop a cancer. The majority of these cancers are chance happenings, and many families will have at least one relative with a cancer. This does not necessarily indicate a hereditary risk.

The first step to assessing risk is to look at the number of cancers in your family by compiling a family history of your extended family, noting those who have had a cancer. Where possible write in information about the age at diagnosis and the type or location of the cancer.

Familial cancers account for just a small percentage of all cancers. Less than one in 20 people affected by cancer have inherited an altered gene.

The following features suggest a hereditary disposition to develop cancer:

- family member affected at a relatively young age (eg less than 50 years old)
- multiple cancers in one affected individual; and
- several family members on the same side of the family affected.

Can I be tested to see if I have a cancer gene?

At the moment genetic testing is only available on a limited basis to individuals with a strong personal or family history of cancer. Testing can also be offered to blood relatives when a mutation has been identified in another affected family member. Genetic counselling is offered before and after genetic testing to discuss the medical, social and emotional implications for an individual and their family. Testing can only be done with prior assessment and counselling at a recognised genetic centre. In some cases it is not always possible to detect an altered gene in the family. However, a regular surveillance program is recommended for individuals based on the family history and assessed risk of developing cancer.

Will genetic research improve treatment?

Possibly one advantage to knowing your inherited risk for cancer is it allows you to undergo special surveillance to improve the chances of detecting cancer early when it is potentially curable. In addition, there are currently over 600 approved clinical gene therapy trials running world-wide. Gene therapy may be used to replace a faulty gene or introduce a new gene whose function is to cure or modify the course of a disease. However, this form of therapy remains an experimental discipline and much research needs to be done to realise its potential. New approaches being studied include therapeutic cloning of stem cells (a controversial therapy used to create specific cells); and pharmacogenetics (study of how drugs work in the body). There are numerous legal, ethical issues to be considered before any of these treatments are routinely offered.

Internet information

The following sites are worth visiting for more information on genetics and cancer:

<http://gslc.genetics.utah.edu/>
www.genetics.com.au
www.cancercouncil.com.au/editorial.asp?pageid=645
(The Cancer Council NSW)

What help is available in WA?

Your general practitioner is a good place to start. They can refer you to a specialist service if appropriate. The following services operate in WA:

Genetic Services of WA, Familial Cancer Program - a state-wide service providing counselling, diagnostic and laboratory services. Phone (08) 9340 1603.
<http://www.kemh.health.wa.gov.au/services/genetics/index.htm>

Familial Cancer Registry - offers assistance with surveillance follow-up. Phone (08) 9340 1603.

Office of Population Health Genomics, Department of Health, service providing education to health professionals and the community. Phone (08) 9323 6600.