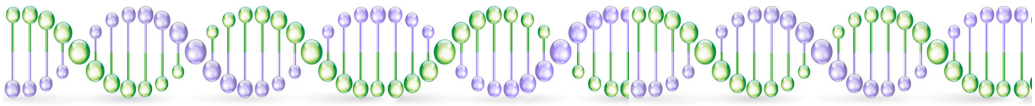
Information Sheet



Fanconi Anaemia and Breast and Ovarian Cancer Risk Genes

Fanconi anaemia is rare, affecting 1 in 160,000 people.

What causes Fanconi anaemia?
It is usually caused by inheriting 2 pathogenic (disease causing) mutations in the same DNA repair gene. This is called biallelic inheritance (an allele refers to the particular version of a gene) and the condition is referred to as a recessive condition. In a recessive condition or syndrome you need 2 mistakes in the same gene, one from each parent, before the condition or syndrome occurs.

When was Fanconi anaemia discovered?
Fanconi anaemia is named after the Swiss paediatrician Giuseppe Fanconi, who first described the syndrome in 1927.

What are the signs and symptoms of Fanconi anaemia?
Fanconi anaemia is very serious and usually results in bone marrow failure in childhood, requiring a bone marrow transplant. It also causes growth retardation, problems with the development of the thumb and/or forearm, a weakened immune system and other problems that may be detected at a very young age.

What causes Fanconi Anaemia?
There are more than 20 genes known to cause Fanconi anaemia. They are usually named FANC + a letter of the alphabet. The following genes, known to increase breast and/or ovarian cancer risk when one mutation is inherited (autosomal dominant), cause Fanconi anaemia when biallelic mutations are inherited (autosomal recessive). You can read more about them in the “genes” section of our website. They include: BRCA1 (FANCS), BRCA2 (FANCD1) BRIP1 (FANCJ), PALB2 (FANCN) and RAD51C (FANCO)

Can Fanconi anaemia be cured?
Fanconi anaemia cannot be cured, although some of the problems it causes can be treated.

Is Fanconi anaemia inherited?
Yes. Fanconi anaemia is a hereditary cancer syndrome caused by inheriting a mutation in both copies of a particular Fanconi gene.

Because Fanconi anaemia affects children and many of the signs and symptoms are not treatable, some couples undertake preconception testing of a partner. That is, if someone carries a BRCA2 mutation, their partner may have genetic testing to make sure they don't carry a BRCA2 mutation as well. If they do, the chance of Fanconi anaemia is 1 in 4.

Some couples use preimplantation genetic diagnosis (PGD) in the setting of in vitro fertilisation (IVF) to prevent the known familial mutation from either parent from being passed on to future generations.

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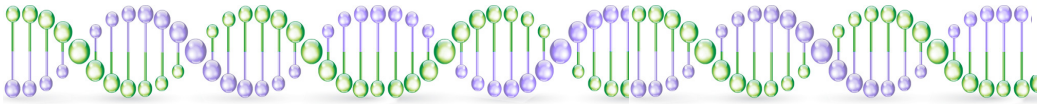

SYDNEY CANCER GENETICS

Does this sound like you or your family?
Has a BRCA1, BRCA2, BRIP1, PALB2 or RAD51C mutation been detected in you, your partner or a blood relative?

Make an appointment with Dr Hilda High at Sydney Cancer Genetics. It is a confidential opportunity to discuss your personal and family history of cancer and genetic testing can be organised, if needed.

More information is available on our website, including links to the following:

- Fanconi Anaemia Support Australasia is a support group for individuals and families affected by Fanconi Anaemia. It is based in Sydney but runs activities throughout Australia and New Zealand.
- The Cancer Genetics section of the Cancer Institute's eviQ website provides up-to-date Australian-based management guidelines for hereditary cancer syndromes
- The US National Library of Medicine website has more information about this syndrome.

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