

Other sources of information

Useful websites

Retinoblastoma International:

www.kidseyecancer.org

National Organisation of Rare Disorders:

www.rarediseases.org

Retinoblastoma website

www.wch.sa.gov.au/rb/rb.html

**Familial Cancer Unit
SA Clinical Genetics Service
Women's & Children's Hospital
North Adelaide SA 5006
Australia**

Tel: (08) 8161 7375

(08) 8161 7010

Fax: (08) 8161 6088

email: sacgs@mail.wch.sa.gov.au

Genetic Testing for Retinoblastoma for Families



Your local Cancer Council:

Retinoblastoma Support Group:

48 David Avenue
North Ryde
NSW 2113

Phone:(02) 9878 4267

Useful contact



Cancer Help Line

Phone: 13 11 20

Information, advocacy, counselling and support services can all be accessed through the Cancer Help Line

South Australian Familial Cancer Service

What is Retinoblastoma?

The retina, situated at the back of the eyeball, is a light sensitive tissue lining with a rich blood and nerve supply. It is responsible for the detection of light. Retinoblastoma is a very rare tumour of the immature cells of the retina in one or both eyes. This only occurs in babies or toddlers under the age of 5 years.

In 1996, 19 children were diagnosed with retinoblastoma in Australia. With early diagnosis this disease is treatable. In the developed world 97% of children survive their retinoblastoma but most have moderate to severe visual impairment.

What are the Signs and Symptoms of Retinoblastoma?

The commonest signs of retinoblastoma are:

- ❖ a white glow or glint in one or both eyes
- ❖ the presence of a white pupil in a colour photo
- ❖ crossed or misaligned eyes

There may be other symptoms that may not be so specific. Any visual problems or eye irritations should be investigated by a medical practitioner.

What causes Retinoblastoma?

Genes are working units of DNA, a complex pool of chemical information that carries a complete set of instructions to enable the cells of our body to function normally.

Most retinoblastoma is sporadic, with little chance that other family members will develop the cancer.

Some children have a familial form of retinoblastoma. The close relatives of these children may be at high risk of developing retinoblastoma (or a related cancer such as sarcoma).

It is usually possible to distinguish these two forms of retinoblastoma by genetic testing.

What is Genetic Testing for Retinoblastoma?

It is now possible to test a person's blood in order to detect whether they have inherited errors in the RB1 gene. The current techniques that are used can detect 90% of these faults (or mutations) in people with familial or bilateral retinoblastoma. In individuals with unilateral retinoblastoma it is feasible to confirm the sporadic nature of the disease in approximately 85% of cases where tumour is available for testing.

The advantages of Genetic Counselling and Testing

There are a number of advantages associated with genetic testing for an inherited mutation of the *RB1 gene*:

- ❖ The presence of a mutation identifies children who may be at risk of developing another retinoblastoma or other cancers later in life.
- ❖ Relatives who are at risk of developing retinoblastoma can be tested for the same mutation to determine whether they need regular screening.
- ❖ If an at-risk relative is found not to have inherited the mutation then no further screening is necessary. The emotional and financial costs of such surveillance can be avoided.

How does one access Genetic Counselling and Testing?

The Institute of Medical and Veterinary Science (IMVS) provides genetic testing for retinoblastoma. Genetic testing should be preceded by genetic counselling. In South Australia this is available through the Familial Cancer Unit. Similar services exist in other States or territories. We recommend that you discuss this with your doctor.