

How does one access genetic counselling and testing for VHL?

The Institute of Medical and Veterinary Science (IMVS) provides genetic testing for VHL.

Genetic counselling should precede genetic testing. 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.

Familial Cancer Unit
SA Clinical Genetics Service
Women's and Children's Hospital
North Adelaide
SA 5006
AUSTRALIA

Tel: (08) 8161 7375
Fax: (08) 8161 6088
Email: sacgs@mail.wch.sa.gov.au

Useful website:

VHL Family Alliance:
www.vhl.org

Useful contact:

**Association of Genetic Support of
Australasia (AGSA)**
66 Albion Street
SURREY HILLS NSW 2010
Email: agsa@ozemail.com.au
Website: www.agsageneticsupport.org.au

Useful contact:

Cancer Help Line
13 11 20

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

Genetic testing for von Hippel-Lindau syndrome (VHL) for families



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What is VHL?

Von Hippel-Lindau Syndrome, or VHL is a rare inherited disease that can present in many different ways in family members.

In people with VHL, very small blood vessels, or capillaries, knot together to form abnormal growths called angiomas. These angiomas may cause little or no problem in some family members, whereas others may have very serious health problems.

A person with VHL may develop angiomas and other tumours in one or more parts of the body :

- ❖ **Retinal angioma**, affecting the retina of the eye
- ❖ **Haemangioblastoma**, in the brain or the spinal cord
- ❖ **Phaeochromocytoma**, a tumour of the adrenal glands, found on top of the kidneys
- ❖ Cysts or cancers in the kidneys and in the pancreas

How do people get VHL?

DNA is a complex set of genetic information within the body's cells that instructs them to work normally. Genes are the working units of DNA. VHL is a disease that results from an error, or a mutation, in a gene called the VHL gene.

Genes come in pairs and a child inherits one gene from each parent. The mutation may be transmitted from a parent to a child following a dominant pattern of inheritance. This means that if one parent has a mutation of the VHL gene their child has a 50% chance of inheriting that mutation.

If a person is the first family member affected by VHL, the siblings and parents are at low risk of having VHL, but that person's child has a 50% chance of inheriting VHL.

What is Genetic Testing for VHL?

It is possible to test a person's blood in order to find out whether they have an inherited mutation in the VHL gene. The current laboratory techniques that are used can detect over 80% of VHL mutations. Once a mutation has been

identified in one affected family member, genetic testing for VHL for other family members is 100% accurate. It is important to have genetic counselling before undergoing genetic testing.

What are the advantages of genetic counselling and testing for VHL?

- ❖ The presence of a mutation in the VHL gene identifies those people who may be at risk of developing angiomas or cancer and derive the greatest benefit from regular screening.
- ❖ Relatives of those with VHL can also be tested for the same mutation to determine whether they also would benefit from regular screening.
- ❖ If a relative is found not to have inherited the mutation then no further screening is necessary. The emotional and financial costs of such screening can therefore be avoided.