

Contact details

Clinical inquiries should be directed to:

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

Laboratory inquiries should be directed to:

Division of Molecular Pathology
Institute of Medical and Veterinary
Science
Frome Road
Adelaide SA 5000

Tel: (08) 8222 3895

Fax: (08) 8222 3146

Email: jacqueline.carroll@imvs.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

Checklist:

- Q Blood sample collected-
10ml in EDTA
- Q Consent form completed
and signed
- Q Service agreement form
completed and signed
- Q All samples and paperwork
sealed in IMVS Path-o-Pak

**Samples should be kept at
room temperature at all times
and transported to the
laboratory within 24 hours
where practicable.**

Feb 2002

Genetic testing for von Hippel-Lindau Syndrome (VHL) for clinicians`



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**South
Australian
Familial
Cancer
Service**

Introduction

A person with an inherited mutation in the VHL gene is at a high risk of developing a number of tumours. Almost all carriers will manifest features by the age of 60 years. The average age of diagnosis is 26 years.

Retinal angiomas are usually the earliest manifestation of VHL syndrome and occur in 70% of patients. These are cytologically benign but can cause blindness. All patients with VHL syndrome require lifelong retinal surveillance.

The lifetime risk of **CNS haemangioblastomas** is 84%. Though histologically identical to a retinal angioma they can cause severe morbidity because of their location in the cerebellum (60%), the spinal cord (25%), or the brain stem (20%). Early treatment is associated with a better prognosis.

Renal cell carcinoma is the leading cause of death in VHL syndrome. It occurs in 60% of patients overall. Some patients have renal cysts identified but the relationship between cysts and carcinoma is not well defined.

The risk of **phaeochromocytoma** varies in different kindreds. The overall risk is approximately 15%.

Other less common complications include pancreatic endocrine tumours, hepatic cysts and angiomas, paragangliomas and endolymphatic sac tumours.

VHL Gene Testing

Germline mutations in the VHL gene can be detected utilizing current techniques in approximately 80% of people with the disease. Once a mutation has been identified in a patient, genetic testing of relatives is essentially 100% accurate.

Testing is conducted under the auspices of the South Australian Familial Cancer Service at the Institute of Medical and Veterinary Science, Adelaide.

Turnaround time for Testing

Analysis of the VHL gene involves screening the three exons of the gene for any small insertions, deletions or single-base substitution mutations by direct DNA sequencing.

Most results are available in 1-2 months.

Detection of an identified mutation can be extended to presymptomatic testing in other family members. This testing takes about 4 weeks.

Procedure For Testing

1. Collect 10ml fresh blood in EDTA. Please ensure that these samples are kept at room temperature at all times including transportation to the lab.
2. Ensure that a *Consent Form* from a Familial Cancer Clinic accompanies each of the samples to be tested. We require that informed consent be obtained and documented. (A suitable consent form is available from the laboratory if required).
3. Complete the *Service Agreement Form* indicating that payment can be guaranteed.
4. Place all paperwork and samples in the IMVS Path-O-PaK and complete the IMVS request form on the front.
5. Forward to the laboratory within 24 hours of collection where practicable.

Results

A full report will be issued on the outcome of the testing. Please indicate on the IMVS request form if additional reports are required to be issued to other specialists involved in the case.

Identification of a mutation in the VHL gene serves to aid both in the genetic counselling process and in early clinical management.