

Von Hippel Lindau syndrome (VHL)

Gene: VHL

Locus: 3p26-p25

OMIM: 193300

OMIM: 608537

SERVICE: mutation and dosage analysis of the VHL gene

TESTING: **Diagnostic*:** clinically affected patients
Presymptomatic patients at risk of developing VHL (known mutation)
Prenatal at risk of having an affected child (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Clinical Geneticists only
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£295 (sequencing + dosage)
Presymptomatic:	2 weeks	£145
Prenatal:	3 days	£350 (including maternal contamination studies)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-3 and splice site boundaries of the VHL gene
 - Multiplex ligation dependent probe amplification analysis of exons 1-3 of the VHL gene
- Intragenic mutations account for 60-70%, and deletion of one or more VHL exon accounts for 30-40% of cases

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**
- Samples should be accompanied by a FULLY completed request card (available from the laboratory)
- Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

CONTACT DETAILS

Genetics Laboratories, Box 143
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Cambridge CB2 0QQ
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of disease(s):	Von Hippel Lindau syndrome
Name of gene(s):	von Hippel-Lindau tumour suppressor; VHL

Patient name:	Date of birth:
Patient postcode:	NHS number:
Name of referrer:	
Title/Position:	
Department/Hospital:	
Contact email/telephone number:	

Referrals will only be accepted from one of the following:
(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you.
Consultant Clinical Geneticist	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
Two VHL-associated lesions including: <ul style="list-style-type: none"> • retinal angiomas • spinal or central haemangioblastoma • Retinal or pancreatic cysts, phaeochromocytoma, renal clear cell carcinoma • Epididymal cystadenomas • Elevated urine or blood catecholamine OR one of the following: <ul style="list-style-type: none"> • Single retinal or CNS haemangioblastoma • Single case of renal cell carcinoma or phaeochromocytoma with features of inherited predisposition (e.g. multiple tumours or relatively young age of onset) • Single case with Endo Lymphatic Sac Tumour • One lesion and FH of VHL associated lesion 	<input type="checkbox"/>
OR At risk family members with a known mutation	<input type="checkbox"/>

If the patient does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory