

Von Hippel Lindau syndrome (VHL)

OMIM 193300

Gene: VHL Locus: 3p25-p26 OMIM: 608537

SERVICE: mutation and dosage analysis of the VHL gene

TESTING: Diagnostic: clinically affected patients
Presymptomatic[#]: relatives of clinically affected patients (known VHL mutation)
Prenatal[#]: at risk of having an affected child (known VHL mutation)

*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Consultant Physicians and Consultant Clinical Geneticists[#] only

TARGET REPORTING TIME AND COSTS

(Non NHS patients are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	8 weeks	£474 (sequencing + dosage)
Presymptomatic:	2 weeks	£195 (sequence one exon)
		£157 (dosage only)
Prenatal:	3 days	£555

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
- Linkage analysis, if definite clinical diagnosis and sufficient family members available (please discuss)

Intragenic mutations account for 60-70%, and deletion of one or more VHL exon accounts for 30-40% of cases

SAMPLE REQUIREMENTS

- 5ml blood in EDTA or 50ul DNA
- 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.

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.

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UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Von Hippel Lindau syndrome (VHL)

Referrals only will 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	
Consultant Physician	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:

Criteria	Tick if this patient meets criteria
1. 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• Urine or blood catecholamine or	
2. One lesion and family history of a VHL-associated lesion	

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