

Using Genetic Testing to Diagnose Mutations in *VHL* as the Cause of Retinal Hemangioblastoma

Indications:

- Retinal hemangioblastoma
- Family history of retinal hemangioblastoma or von Hippel-Lindau disease

Benefits:

- Genetic testing for von Hippel-Lindau disease (VHL) can:
 - identify VHL as the cause of retinal hemangioblastoma.
 - alert patients and physicians to the risk of other VHL-associated manifestations.
 - allow family screening for VHL, helping to identify family members who should undergo frequent tumor screening and family members who do not need frequent tumor screening.

Background:

- 30-60% of all retinal hemangioblastoma is due to VHL.^{1,2}
- VHL is a familial cancer syndrome associated with cerebellar, and/or spinal hemangioblastoma, pheochromocytoma, and or renal cell carcinoma in addition to retinal hemangioblastoma.³
- VHL is inherited in an autosomal dominant manner.³
- Frequent tumor screening has been recommended for patients with VHL and their at-risk family members.³
- Early diagnosis of VHL has been shown to increase survival.⁴
- VHL is due to loss-of-function mutations in the tumor suppressor gene *VHL*.⁵
- Up to 60% of VHL-associated mutations in *VHL* can be detected by gene sequencing.⁶

References: 1. Niemela M, et al (2000) Invest Ophthalmol Vis Sci 41:1909-15. 2. Singh AD, et al (2001) Ophthalmology 108:1907-11. 3. Richard S, et al (2003) Expert Rev Anticancer Ther 3:215-33. 4. Richards FM, et al (1998) J Intern Med 243:527-33. 5. Latif F, et al (1993) Science 260:1317-20. 6. Stolle C et al (1998) Hum Mutat 12:417-23.

Ordering Information: please see other side

Ordering Information for the *VHL* DNA Sequencing Test

Indications for Testing

- Retinal hemangioblastoma
- Family history of retinal hemangioblastoma or von Hippel-Lindau disease

Ordering Information

Gene(s)	Test Code
<i>VHL</i>	180201

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit,
please contact Client Services at 1-866-647-0735 or visit us on the web at
www.correlagen.com.**