

Using Genetic Testing to Diagnose Mutations in *VHL* as the Cause of Renal Cell Carcinoma

Indications:

- Multifocal or bilateral renal cell carcinoma
- Renal cell carcinoma in the presence of renal cysts or before age 50 years
- Renal cell carcinoma in the presence of other characteristic manifestations of von Hippel-Lindau disease (VHL)
- Family history of renal cell carcinoma or VHL

Benefits:

- Genetic testing for VHL can:
 - identify VHL as the cause of renal cell carcinoma.
 - 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:

- VHL is the primary cause of hereditary renal cell carcinoma, accounting for about 1.6% of all cases.¹
- VHL is a familial cancer syndrome associated with retinal, cerebellar, and/or spinal hemangioblastoma and/or pheochromocytoma in addition to renal cell carcinoma.²
- VHL is inherited in an autosomal dominant manner.²
- Frequent tumor screening has been recommended for patients with VHL and their at-risk family members.²
- Early diagnosis of VHL has been shown to increase survival.³
- VHL is due to loss-of-function mutations in the tumor suppressor gene *VHL*.⁴
- Up to 60% of VHL-associated mutations in *VHL* can be detected by gene sequencing.⁵

References: 1. Neumann HP, et al (1998) J Urol 160:1248-54. 2. Richard S, et al (2003) Expert Rev Anticancer Ther 3:215-33. 3. Richards FM, et al (1998) J Intern Med 243:527-33. 4. Latif F, et al (1993) Science 260:1317-20. 5. Stolle C et al (1998) Hum Mutat 12:417-23.

Ordering Information: please see other side

Ordering Information for the VHL DNA Sequencing Test

Symptoms Indicating This Syndrome

- Multifocal or bilateral renal cell carcinoma
- Renal cell carcinoma in the presence of renal cysts or before age 50 years
- Renal cell carcinoma in the presence of other characteristic manifestations of von Hippel-Lindau disease
- Family history of renal cell carcinoma or von Hippel-Lindau disease

Ordering Information

Gene(s)	Test Code
VHL	170101

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.