Test Definition: VHLKP

Von Hippel-Lindau (VHL) Gene, Known Mutation

Reporting Title: VHL Known Mutation
Performing Location: Rochester

Specimen Requirements:

Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send specimen in original tube.
Additional Information:
1. This test can only be performed if a mutation has previously been identified in a family member of this individual. Documentation of the specific familial mutations must be provided with the specimen in order to perform this test. If the familial mutation is a large or exonic deletion, order VHLD/89211 Von Hippel-Lindau (VHL) Deletion Detection. If a familial mutation has not been previously identified, order VHLSP/89083 Von Hippel-Lindau (VHL) Gene, Full Gene Analysis.
2. Transfusions will interfere with testing for up to 4 to 6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte-reduced. Wait 4 to 6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.
Forms:
1. VHL Gene Testing Patient Information Sheet (Supply T641) in Special Instructions is required.
2. Informed Consent for Genetic Testing (Supply T576) in Special Instructions is required.
3. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

Specimen Type	Temperature	Time
Whole Blood EDTA	Refrigerated (preferred)	
	Ambient	
	Frozen	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
28626	Reason For Referral	Alphanumeric		42349-1
28627	Method	Alphanumeric		In Process
28628	Result	Alphanumeric		N/A
28629	Interpretation	Alphanumeric		69047-9
28630	Comment	Alphanumeric		N/A
28631	Reviewed By	Numeric		N/A
VHLSQ	Sequencing	Alphanumeric		34502-5

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
VHLKM	VHL Known Mutation			No	No
VHLMQ	VHL Known Mutation Sequencing			No	No

CPT Code Information:

81403-Known familial variant, not otherwise specified, for gene listed in Tier 1 or Tier 2, DNA sequence analysis

Reference Values:

An interpretive report will be provided.