MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Gene Panel

The following applies to HHTGG / Hereditary Hemorrhagic Telangiectasia and Vascular Malformations Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from November 2022 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
ACVRL1	NM_000020.3	-	-
BMPR2	NM_001204.7	chr2:g.203395505T>G (c.968-12T>G)	-
CCM2	NM_031443.3	-	-
ENG	NM_001114753.2	chr9:g.130616761G>A (c.-127C>T); chr9:g.130578359A>G (c.1742-22T>C)	-
EPHB4	NM_004444.5	-	-
GDF2	NM_016204.4	-	-
GLMN	NM_053274.3	-	CNV may not be detected in exons 2, 10, 16, and 17
KRIT1	NM_194456.1	chr7:g.91856011A>T (c.990-15T>A); chr7:g.91855154G>C (c.1147-13C>G); chr7:g.91830749T>C (c.2026-12A>G)	-
PDCD10	NM_145860.1	-	-
RASA1	NM_002890.3	-	-
SMAD4	NM_005359.6	-	-
TEK	NM_000459.4	-	-

Effective Date	Version	Synopsis of Test Change
6/4/2025	V2	• Added Additional Evaluations for *BMPR2, ENG,* and *KRIT1* • Added Technical Limitation for *GLMN* exon 16

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