MAYO CLINIC LABORATORIES

# Targeted Genes and Methodology Details for Hereditary Renal Cancer Gene Panel

The following applies to RENCP / Hereditary Renal Cancer 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 noncoding variants. Next-generation sequencing, multiplex ligation-dependent probe amplification (MLPA), 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 December 2024 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 |
|---|---|---|---|
| *BAP1* | NM_004656.4 | - | - |
| *DICER1* | NM_177438.2 | - | - |
| *FH* | NM_000143.3 | - | - |
| *FLCN* | NM_144997.7 | CNV in noncoding exons 1–3 | - |
| *MET* | NM_001127500.3 | - | - |
| *MITF* | NM_000248.3 | - | Analyzed only for the presence of c.952G>A p.E318K (rs149617956), analysis for other sequence variants and CNV will not be performed |
| *PTEN* | NM_000314.8 | Promoter: c.-1302 to c.-589 | - |
| *SDHA* | NM_004168.4 | - | - |
| *SDHAF2* | NM_017841.2 | - | - |
| *SDHB* | NM_003000.3 | - | - |
| *SDHC* | NM_003001.3 | - | - |
| *SDHD* | NM_003002.4 | - | - |
| *SMARCA4* | NM_001128849.2 | - | CNV in exon 27 will not be detected or reported |
| *SMARCB1* | NM_003073.5 | - | - |
| *TMEM127* | NM_017849.3 | - | - |
| *TP53* | NM_000546.5 | - | - |
| *TSC1* | NM_000368.5 | - | - |
| *TSC2* | NM_000548.5 | - | - |
| *VHL* | NM_000551.3 | - | - |

| Effective Date | Version | Synopsis of Test Change |
|---|---|---|
| June 2023 | V2 | Updated format (additional columns: Additional Evaluations and Technical Limitations) |
| December 2024 | V3 | *FLCN*: Added CNV detection of noncoding exons 1–3 |