MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Paraganglioma/Pheochromocytoma Panel

The following applies to HPGLP / Hereditary Paraganglioma/Pheochromocytoma 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 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 2021 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
FH	NM_000143.3	-	-
MAX	NM_002382.5	-	-
NF1	NM_000267.3	-	Analysis for sequence variants beyond coding exons +/- 10 base pairs of adjacent intronic sequence will not be performed
RET	NM_020975.6	-	-
SDHA	NM_004168.4	-	-
SDHAF2	NM_017841.2	-	-
SDHB	NM_003000.3	-	-
SDHC	NM_003001.3	-	-
SDHD	NM_003002.4	-	-
TMEM127	NM_017849.3	-	-
VHL	NM_000551.3	-	-

Effective Date	Version	Synopsis of Test Change
June 2023	V2	Updated format (additional columns: Additional Evaluations and Technical Limitations)