MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Expanded Cancer Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the gene(s) analyzed. NGS, multiplex ligation-dependent probe amplification (MLPA), and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the gene(s) analyzed. PCR and gel electrophoresis is performed to test for the presence of the 10 megabase inversion of coding exons 1–7 of the *MSH2* gene. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
AIP	NM_003977.4
ALK	NM_004304.5
APC	NM_000038.6
ATM	NM_000051.3
AXIN2	NM_004655.4
BAP1	NM_004656.4
BARD1	NM_000465.4
BLM	NM_000057.4
BMPR1A	NM_004329.2
BRCA1	NM_007294.4
BRCA2	NM_000059.3
BRIP1	NM_032043.3
BUB1B	NM_001211.5
CDC73	NM_024529.4
CDH1	NM_004360.5
CDK4	NM_000075.4
CDKN1B	NM_004064.4
CDKN2A	NM_000077.4
CHEK2	NM_007194.4
CTNNA1	NM_001903.5
DICER1	NM_177438.2
DIS3L2[b,c]	NM_152383.4
EGFR	NM_005228.5
ELP1	NM_003640.5
EPCAM[d]	NM_002354.3
EXT1	NM_000127.2
EXT2	NM_207122.1
FANCA	NM_000135.4
FH	NM_000143.3

Gene	Reference Transcript[a]
FLCN	NM_144997.7
GPC3	NM_004484.4
GREM1[e]	NM_013372.7
HOXB13	NM_006361.5
KIT	NM_000222.2
LZTR1	NM_006767.4
MAX	NM_002382.5
MC1R	NM_002386.3
MEN1	NM_130799.2
MET	NM_001127500.3
MITF[f]	NM_000248.3
MLH1	NM_000249.3
MLH3[c]	NM_001040108.1
MSH2	NM_000251.3
MSH3	NM_002439.5
MSH6	NM_000179.2
MUTYH	NM_001128425.1
NBN[c]	NM_002485.4
NF1	NM_000267.3
NF2	NM_000268.3
NTHL1	NM_002528.7
PALB2	NM_024675.4
PDGRFA[c]	NM_006206.6
PHOX2B	NM_003924.4
PMS2	NM_000535.7
POLD1	NM_002691.4
POLE	NM_006231.4
POT1[c]	NM_015450.3
PRKAR1A	NM_002734.4

Targeted Genes and Methodology Details for Hereditary Expanded Cancer Panel (continued)

Gene	Reference Transcript[a]
PTCH1	NM_000264.5
PTEN	NM_000314.8
RAD51B	NM_133509.4
RAD51C	NM_058216.3
RAD51D	NM_002878.3
RB1	NM_000321.2
RECQL4	NM_004260.3
REST	NM_005612.5
RET	NM_020975.6
RNF43	NM_017763.5
SDHA[b,c]	NM_004168.4
SDHAF2	NM_017841.2
SDHB	NM_003000.3
SDHC	NM_003001.3
SDHD	NM_003002.4

Gene	Reference Transcript[a]
SMAD4	NM_005359.6
SMARCA4[c]	NM_001128849.2
SMARCB1	NM_003073.5
SMARCE1[c]	NM_003079.5
STK11	NM_000455.5
SUFU	NM_016169.3
TMEM127	NM_017849.3
TP53	NM_000546.5
TRIP13	NM_004237.4
TSC1	NM_000368.5
TSC2	NM_000548.5
VHL	NM_000551.3
WRN	NM_000553.6
WT1	NM_024426.6

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively evaluated by sequencing as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences.

[c] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

[d] This gene is analyzed for the presence of copy number variants only. No sequencing analysis is performed.

[e] The *GREM1* gene is analyzed for the presence of copy number gains involving the upstream enhancer region only. No other sequencing or copy number variant analyses are performed.

[f] The *MITF* gene is analyzed for the presence of variant c.952G>A p.E318K (dbSNP rs149617956) only. No other sequencing or copy number variant analyses are performed for this gene.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.