MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Gastrointestinal 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]
APC	NM_000038.6
ATM	NM_000051.3
AXIN2	NM_004655.4
BMPR1A	NM_004329.2
CDH1	NM_004360.5
CHEK2	NM_007194.4
CTNNA1	NM_001903.5
EPCAM[c]	NM_002354.3
GREM1[d]	NM_013372.7
KIT	NM_000222.2
MLH1	NM_000249.3
MLH3[b]	NM_001040108.1
MSH2	NM_000251.3

Gene	Reference Transcript[a]
MSH3	NM_002439.5
MSH6	NM_000179.2
MUTYH	NM_001128425.1
NTHL1	NM_002528.7
PDGFRA[b]	NM_006206.6
PMS2	NM_000535.7
POLD1	NM_002691.4
POLE	NM_006231.4
PTEN	NM_000314.8
RNF43	NM_017763.5
SMAD4	NM_005359.6
STK11	NM_000455.5
TP53	NM_000546.5

[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 analyzed for the presence of copy number variants.

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

[d] 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.

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