MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Breast/Gynecologic Cancer Panel

The following applies to BRGYP / Hereditary Breast/Gynecologic 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 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. Polymerase chain reaction and gel electrophoresis is performed to test for the presence of the 10 megabase (Mb) 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.

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
ATM	NM_000051.3	-	-
BARD1	NM_000465.4	-	-
BRCA1	NM_007294.4	+/- 20 base pairs of adjacent intronic sequence on either side of the coding exons	-
BRCA2	NM_000059.3	+/- 20 base pairs of adjacent intronic sequence on either side of the coding exons	-
BRIP1	NM_032043.3	-	-
CDH1	NM_004360.5	-	-
CHEK2	NM_007194.4	-	-
EPCAM	NM_002354.3	-	Analysis for sequence variants will not be performed
MLH1	NM_000249.3	c.-27C>A (rs587779001) c.454-13A>G (rs267607749) c.1990-16_1990-2del (rs267607881)	-
MSH2	NM_000251.3	10 Mb inversion of exons 1–7	-
MSH6	NM_000179.2	-	-
NBN	NM_002485.4	-	CNV in exon 16 will not be detected or reported
NF1	NM_000267.3	-	Analysis for sequence variants beyond coding exons +/- 10 base pairs of adjacent intronic sequence will not be performed
PALB2	NM_024675.4	-	-
PMS2	NM_000535.7	-	-
PTEN	NM_000314.8	Promoter: c.-1302 to c.-589	-
RAD51C	NM_058216.3	-	-
RAD51D	NM_002878.3	-	-
STK11	NM_000455.5	-	-
TP53	NM_000546.5	-	-

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