MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Rapid Hereditary Breast Cancer Treatment Decision Panel

The following applies to BRTP / Rapid Hereditary Breast Cancer Treatment Decision 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 June 2023 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	-	-
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	-
CDH1	NM_004360.5	-	-
CHEK2	NM_007194.4	-	-
PALB2	NM_024675.4	-	-
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	-	-