



Test Definition: B RTP

Rapid Hereditary Breast Cancer Treatment
Decision Panel, Varies

Overview

Useful For

Establishing a diagnosis of a hereditary breast cancer syndrome allowing for surgical and management decision making

Determining therapeutic eligibility with poly (adenosine diphosphate-ribose) polymerase inhibitors based on certain gene alterations (eg, *BRCA1*, *BRCA2*) in selected tumor types

Evaluating patients with breast cancer who have a personal history suggestive of a hereditary breast or gynecological cancer syndrome

Identifying genetic variants associated with increased risk for breast cancer, allowing for predictive testing and appropriate screening of at-risk family members

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No
CULAF	Amniotic Fluid Culture/Genetic Test	Yes	No
_STR1	Comp Analysis using STR (Bill only)	No, (Bill only)	No
_STR2	Add'l comp analysis w/STR (Bill Only)	No, (Bill only)	No
MATCC	Maternal Cell Contamination, B	Yes	No

Genetics Test Information

This test utilizes next-generation sequencing to detect single nucleotide and copy number variants in 11 genes associated with hereditary breast cancer: *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN* (including promoter), *RAD51C*, *RAD51D*, *STK11*, and *TP53*. See [Targeted Genes and Methodology Details for Rapid Hereditary Breast Cancer Treatment Decision Panel](#) and Method Description for additional details.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, recurrence risk assessment, familial screening, and genetic counseling for hereditary breast cancer.

Testing Algorithm

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

For more information see [Breast, Gynecological and Prostate Cancer Testing Algorithm](#).

Special Instructions

- [Molecular Genetics: Inherited Cancer Syndromes Patient Information](#)
- [Informed Consent for Genetic Testing](#)
- [Informed Consent for Genetic Testing \(Spanish\)](#)
- [Breast, Gynecological and Prostate Cancer Testing Algorithm](#)
- [Targeted Genes and Methodology Details for Rapid Hereditary Breast Cancer Treatment Decision Panel](#)

Method Name

Sequence Capture and Targeted Next-Generation Sequencing (NGS) followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

NY State Available

Yes

Specimen**Specimen Type**

Varies

Ordering Guidance

This test is for patients diagnosed with cancer for whom results may impact treatment. A rapid turnaround time supports surgical and management decision making. For patients with cancer who do not need rapid results, order BRGYP / Hereditary Breast/Gynecologic Cancer Panel, Varies or COMCP / Hereditary Common Cancer Panel, Varies depending on the patient's personal and family history.

This test is **not appropriate for** patients who do not have cancer. If testing is needed based on a previous diagnosis of cancer or family history of cancer, order either BRGYP / Hereditary Breast/Gynecologic Cancer Panel, Varies or COMCP / Hereditary Common Cancer Panel, Varies, depending on the patient's personal and family history.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. For more information see FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Testing minors for adult-onset predisposition syndromes is discouraged by the American Academy of Pediatrics, the American College of Medical Genetics and Genomics, and the National Society of Genetic Counselors.

Specimen Required

Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top (ACD)

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. **Do not aliquot.**
3. Whole blood collected postnatal from an umbilical cord is also acceptable. See Additional Information.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

Additional Information:

1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
2. To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
3. For postnatal umbilical cord whole blood specimens, maternal cell contamination studies are recommended to ensure test results reflect that of the patient tested. A maternal blood specimen is required to complete maternal cell contamination studies. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on both the cord blood and maternal blood specimens under separate order numbers.

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

Supplies:

DNA Saliva Kit High Yield (T1007)

Saliva Swab Collection Kit (T786)

Container/Tube:

Preferred: High-yield DNA saliva kit

Acceptable: Saliva swab

Specimen Volume: 1 Tube if using T1007 or 2 swabs if using T786

Collection Instructions: Collect and send specimen per kit instructions.

Specimen Stability Information: Ambient (preferred) 30 days/Refrigerated 30 days

Additional Information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Extracted DNA

Container/Tube:

Preferred: Screw Cap Micro Tube, 2 mL with skirted conical base

Acceptable: Matrix tube, 1 mL

Collection Instructions:

1. The preferred volume is at least 100 µL at a concentration of 75 ng/µL.
2. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated

Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Forms

1. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:

-[Informed Consent for Genetic Testing](#) (T576)

-[Informed Consent for Genetic Testing \(Spanish\)](#) (T826)

2. [Molecular Genetics: Inherited Cancer Syndromes Patient Information](#) (T519)

3. If not ordering electronically, complete, print, and send a [Oncology Test Request](#) (T729) with the specimen.

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Clinical & Interpretive

Clinical Information

Breast cancers occur in about 12% of the general population.(1) In some cases, breast cancer may be attributed to a hereditary cancer syndrome.(2-5) Evaluation of the genes on this panel may be useful for patients with breast cancer to determine surgical and management decision making. Rapid turnaround time testing allows for informative results to be returned to patients and providers prior to surgery or initiation of other treatment.

Hereditary breast and ovarian cancer syndrome (HBOC), caused by disease-causing variants in the *BRCA1* and *BRCA2* genes, accounts for the majority of hereditary breast cancer.(2,4) HBOC is predominantly characterized by early-onset breast cancer and ovarian cancer. Individuals with HBOC are also at increased risks for prostate, pancreatic, and male breast cancers.(2,4)

There are other genes known to increase risk for breast cancer that are included on this panel.(2) The risk for developing cancer associated with these syndromes varies.(2) Some individuals with a disease-causing variant in one of these genes develop multiple primary cancers or bilateral cancers.(2)

The National Comprehensive Cancer Network and the American Cancer Society provide recommendations regarding the

medical management of individuals with hereditary breast cancer syndromes.(2,3,6,7)

Reference Values

An interpretive report will be provided.

Interpretation

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations.(8) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance

Cautions

Clinical Correlations:

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data.

Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

If testing was performed because of a clinically significant family history, it is often useful to first test an affected family member. Detection of a reportable variant in an affected family member would allow for more informative testing of at-risk individuals.

To discuss the availability of additional testing options or for assistance in the interpretation of these results, contact Mayo Clinic Laboratories genetic counselors at 800-533-1710.

Technical Limitations:

Next-generation sequencing may not detect all types of genomic variants. In rare cases, false-negative or false-positive results may occur. The depth of coverage may be variable for some target regions; assay performance below the minimum acceptable criteria or for failed regions will be noted. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder. If a specific clinical disorder is suspected, evaluation by alternative methods can be considered.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis due to technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. Confirmation of select reportable variants will be performed by alternate methodologies based on internal laboratory criteria.

This test is validated to detect 95% of deletions up to 75 base pairs (bp) and insertions up to 47 bp. Deletions-insertions (delins) of 40 or more bp, including mobile element insertions, may be less reliably detected than smaller delins.

This analysis targets single and multi-exon deletions/duplications; however, in some instances single exon resolution cannot be achieved due to isolated reduction in sequence coverage or inherent genomic complexity. Balanced structural rearrangements (such as translocations and inversions) may not be detected.

Deletion/duplication events that extend past the genes included on the panel may occur. In these instances, genes included in the ordered test are provided on the report and interpreted, and genomic breakpoints are reported if they are confirmed. However, copy number variants for genes not listed in the Method Description are typically not reported or interpreted for haploinsufficiency/triplosensitivity. CMACB / Chromosomal Microarray, Congenital, Blood; WESPR /

Panel to Whole Exome Sequencing Reflex Test, Varies; or WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies is recommended for a full interpretation of deletions/duplications predicted to extend past the genes included on the panel.

This test is not designed to detect low levels of mosaicism or differentiate between somatic and germline variants. If there is a possibility that any detected variant is somatic, additional testing may be necessary to clarify the significance of results.

Genes may be added or removed based on updated clinical relevance. For the most up to date list of genes included in this test as well as detailed information regarding gene specific performance and technical limitations, see Method Description or contact a laboratory genetic counselor.

If the patient has had an allogeneic hematopoietic stem cell transplant or a recent non-leukocyte reduced blood transfusion, results may be inaccurate due to the presence of donor DNA. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

Reclassification of Variants:

Currently, it is not standard practice for the laboratory to systematically review previously classified variants on a regular basis. The laboratory encourages healthcare professionals to contact the laboratory at any time to learn how the classification of a particular variant may have changed over time. Due to broadening genetic knowledge, it is possible that the laboratory may discover new information of relevance to the patient. Should that occur, the laboratory may issue an amended report.

Variant Evaluation:

Evaluation and categorization of variants are performed using published American College of Medical Genetics and Genomics and the Association for Molecular Pathology recommendations as a guideline.⁽⁸⁾ Other gene-specific guidelines may also be considered. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance. Variants classified as benign or likely benign are not reported.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and periodic updates to these tools may cause predictions to change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgment.

Rarely, incidental or secondary findings may implicate another predisposition or presence of active disease. Incidental findings may include, but are not limited to, results related to the sex chromosomes. These findings will be carefully reviewed to determine whether they will be reported.

Clinical Reference

1. Howlader N, Noone AM, Krapcho M. SEER Cancer Statistics Review, 1975-2018, National Cancer Institute. Updated April 2021. Accessed November 13, 2025. Available at https://seer.cancer.gov/csr/1975_2018/
2. Daly MB, Pal T, Berry M, et al. NCCN Guidelines Insights: Genetic/familial high-risk assessment: breast, ovarian, and pancreatic, version 2.2021. J Natl Compr Canc Netw. 2021;19(1):77-102

3. Gupta S, Provenzale D, Llor X, et al. NCCN Guidelines Insights: Genetic/familial high-risk assessment: colorectal, version 2.2019. *J Natl Compr Canc Netw*. 2019;17(9):1032-1041
4. Petrucelli N, Daley MB, Pal T. *BRCA1*- and *BRCA2*-associated hereditary breast and ovarian cancer. In: Adams MP, Feldman J, Mirzaa GM, et al, eds. *GeneReviews* [Internet]. University of Washington, Seattle; 1998. Updated March 20, 2025. Accessed November 13, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1247/
5. Idos G, Valle L. Lynch syndrome. In: Adam MP, Feldman J, Mirzaa GM, et al, eds. *GeneReviews* [Internet]. University of Washington, Seattle; 2004. Updated February 2, 2021. Accessed November 13, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1211/
6. Saslow D, Boetes C, Burke W, et al. American Cancer Society guidelines for breast screening with MRI as an adjunct to mammography. *CA Cancer J Clin*. 2007;57(2):75-89
7. Smith RA, Andrews KS, Brooks D, et al. Cancer screening in the United States, 2019: A review of current American Cancer Society guidelines and current issues in cancer screening. *CA Cancer J Clin*. 2019;69(3):184-210
8. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med*. 2015;17(5):405-424

Performance

Method Description

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 genes analyzed, as well as some other regions that have known disease-causing variants. The human genome reference GRCh37/hg19 build was used for sequence read alignment. At least 99% of the bases are covered at a read depth over 30X. Sensitivity is estimated at above 99% for single nucleotide variants, above 94% for deletions/insertions (delins) less than 40 base pairs (bp), above 95% for deletions up to 75 bp and insertions up to 47 bp. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed.

There may be regions of genes that cannot be effectively evaluated by sequencing or deletion and duplication analysis due to technical limitations of the assay, including regions of homology, high guanine-cytosine (GC) content, and repetitive sequences. See [Targeted Genes and Methodology Details for Rapid Hereditary Breast Cancer Treatment Decision Panel](#) for details regarding the targeted genes analyzed for each test and specific gene regions not routinely covered. (Unpublished Mayo method)

Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genes analyzed: *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *PTEN* (including promoter), *RAD51C*, *RAD51D*, *STK11*, and *TP53*

PDF Report

Supplemental

Day(s) Performed

Varies

Report Available

10 to 14 days

Specimen Retention Time

Whole blood: 25 days (if available); Extracted DNA: 3 months, Saliva: 30 days (if available)

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81432

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
B RTP	Rapid Hereditary Breast Cancer Test	97656-3

Result ID	Test Result Name	Result LOINC® Value
619958	Test Description	62364-5
619959	Specimen	31208-2
619960	Source	31208-2
619961	Result Summary	50397-9
619962	Result	82939-0
619963	Interpretation	69047-9
619964	Resources	99622-3
619965	Additional Information	48767-8
619966	Method	85069-3
619967	Genes Analyzed	82939-0
619968	Disclaimer	62364-5

Test Definition: B RTP

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