DIAGNOSTIC TESTING RESULTS

Patient name:	Yvonne K Franklin	**Sample type:**	Blood	**Report date:**	04-OCT-2026
DOB:	17-JUN-1979	**Sample collection date:**	25-SEP-2026	**Invitae #:**	RQ8460236
Sex assigned at birth:	Female	**Sample accession date:**	26-SEP-2026	**Clinical team:**	Kari Thorsen Rachel Yung
Gender:					
Patient ID (MRN):	U4772842				

Reason for testing

Diagnostic test for both a personal and a family history of disease

Test performed

Sequence analysis and deletion/duplication testing of the 13 genes listed in the Genes Analyzed section.

- Invitae Hereditary Breast Cancer Guidelines-Based Panel

RESULT: NEGATIVE

About this test

This diagnostic test evaluates 13 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

Next steps

- This test did not identify any pathogenic variants known to cause disease. This result does NOT exclude a genetic diagnosis and should be discussed with a healthcare provider, such as a genetic counselor, to learn about the appropriate next steps for further evaluation. Clinical follow up may still be warranted. This result should be interpreted within the context of additional laboratory results, family history and clinical findings.
- Genetic counseling is available to discuss the potential clinical implications of this result. To access Labcorp Genetic Counselors please visit **https://schedulegc.labcorp.com/SRC** or call (855) GC-CALLS (855-422-2557).

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Clinical summary

No reportable genetic variants were identified by this analysis, however, this individual may still be at risk for certain medical conditions based on other factors such as family history, genetic causes not evaluated with this test, or other environmental influences. Follow up of this individual and surveillance of their family members may still be indicated.

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Genes analyzed

This table represents a complete list of genes analyzed for this individual, including the relevant gene transcript(s). If more than one transcript is listed for a single gene, variants were reported using the first transcript listed unless otherwise indicated in the report. An asterisk (*) indicates that this gene has a limitation. Please see the Limitations section for details. Results are negative unless otherwise indicated in the report. Benign and Likely Benign variants are not included in this report and in specific scenarios variants of uncertain significance in the requisitioned gene(s) may not be included in this report.

GENE	TRANSCRIPT
ATM*	NM_000051.3
BARD1	NM_000465.3
BRCA1	NM_007294.3
BRCA2	NM_000059.3
CDH1	NM_004360.3
CHEK2	NM_007194.3
NF1*	NM_000267.3
PALB2	NM_024675.3
PTEN*	NM_000314.4
RAD51C	NM_058216.2
RAD51D	NM_002878.3
STK11	NM_000455.4
TP53	NM_000546.5

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Methods

- Genomic DNA obtained from the submitted sample is enriched for targeted regions using a hybridization-based protocol, and sequenced using Illumina technology. Unless otherwise indicated, all targeted regions are sequenced with >=50x depth or are supplemented with additional analysis. Reads are aligned to a reference sequence (GRCh37), and sequence changes are identified and interpreted in the context of a single clinically relevant transcript, indicated in the Genes Analyzed table. Enrichment and analysis focus on the coding sequence of the indicated transcripts, 20bp of flanking intronic sequence, and other specific genomic regions demonstrated to be causative of disease at the time of assay design. Promoters, untranslated regions, and other non-coding regions are not otherwise interrogated. For some genes only targeted loci are analyzed. Exonic deletions and duplications are called using an in-house algorithm that determines copy number at each target by comparing the read depth for each target in the proband sequence with both mean read-depth and read-depth distribution, obtained from a set of clinical samples. Markers across the X and Y chromosomes are analyzed for quality control purposes and may detect deviations from the expected sex chromosome complement. Such deviations may be included in the report in accordance with internal guidelines. Variants are reported according to the Human Genome Variation Society (HGVS) guidelines. Confirmation of the presence and location of reportable variants is performed as needed based on stringent criteria using one of several validated orthogonal approaches (PubMed ID 30610921). Sequencing is performed by Labcorp Genetics (1400 16th Street, San Francisco, CA 94103, #05D2040778). Confirmatory sequencing is performed by Labcorp Genetics (1400 16th Street, San Francisco, CA 94103, #05D2040778). RNA sequencing is performed by Labcorp Genetics (1400 16th Street, San Francisco, CA 94103, #05D2040778).

  The following additional analyses are performed if relevant to the requisition. For PMS2 exons 12-15, the reference genome has been modified to force all sequence reads derived from PMS2 and the PMS2CL pseudogene to align to PMS2, and variant calling algorithms are modified to support an expectation of 4 alleles. If a rare SNP or indel variant is identified by this method, both PMS2 and the PMS2CL pseudogene are amplified by long-range PCR and the location of the variant is determined by Pacific Biosciences (PacBio) SMRT sequencing of the relevant exon in both long-range amplicons. If a CNV is identified, MLPA or MLPA-seq is run to confirm the variant. If confirmed, both PMS2 and PMS2CL are amplified by long-range PCR, and the identity of the fixed differences between PMS2 and PMS2CL are sequenced by PacBio from the long-range amplicon to disambiguate the location of the CNV. For C9orf72 repeat expansion testing, hexanucleotide repeat units are detected by repeat-primed PCR (RP-PCR) with fluorescently labeled primers followed by capillary electrophoresis. Interpretation Reference Ranges: Benign (Normal Range): <25 repeat units, Uncertain: 25-30 repeat units, Pathogenic (Full Mutation): >=31 repeat units (PMID: 21944779, 22406228, 23111906, 28689190, 31315673, 33168078, 33575483). A second round of RP-PCR utilizing a non-overlapping set of primers is used to confirm the initial call in the case of suspected allele sizes of 22 or more repeats. For RNA analysis of the genes indicated in the Genes Analyzed table, complementary DNA is synthesized by reverse transcription from RNA derived from a blood specimen and enriched for specific gene sequences using capture hybridization. After high-throughput sequencing using Illumina technology, the output reads are aligned to a reference sequence (genome build GRCh37; custom derivative of the RefSeq transcriptome) to identify the locations of exon junctions through the detection of split reads. The relative usage of exon junctions in a test specimen is assessed quantitatively and compared to the usage seen in control specimens. Abnormal exon junction usage is evaluated as evidence in the Sherloc variant interpretation framework. If an abnormal splicing pattern is predicted based on a DNA variant outside the typical reportable range, as described here, the presence of the variant is confirmed by targeted DNA sequencing.

- A PMID is a unique identifier referring to a published, scientific paper. Search by PMID at http://www.ncbi.nlm.nih.gov/pubmed.
- An rsID is a unique identifier referring to a single genomic position, and is used to associate population frequency information with sequence changes at that position. Reported population frequencies are derived from a number of public sites that aggregate data from large-scale population sequencing projects, including ExAC, gnomAD, and dbSNP.
- A MedGen ID is a unique identifier referring to an article in MedGen, NCBI's centralized database of information about genetic disorders and phenotypes. Search by MedGen ID at http://www.ncbi.nlm.nih.gov/medgen. An OMIM number is a unique identifier referring to a comprehensive entry in Online Mendelian Inheritance in Man (OMIM). Search by OMIM number at http://omim.org/.
- The laboratory uses information from individuals undergoing testing to inform variant interpretation. If "internal data" is cited as a reference in the variant details this may refer to the individual in this requisition and/or historical internal observations.

Limitations

Based on validation study results, this assay achieves >99% analytical sensitivity and specificity for single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications. This test detects insertions and deletions larger than 15bp but smaller than a full exon but sensitivity for these may be marginally reduced. In some situations, single-exon copy number events may not be confidently determined due to inherent sequence properties or isolated reduction in data quality. Certain types of variants, such as structural rearrangements (e.g. inversions, gene conversion events, translocations, etc.) or variants embedded in sequence with complex architecture (e.g. short tandem repeats or segmental duplications), may not

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be detected. Additionally, it may not be possible to fully resolve certain details about variants, such as mosaicism, phasing, or mapping ambiguity. Unless explicitly guaranteed, sequence changes in the promoter, non-coding exons, and other non-coding regions are not covered by this assay. Please consult the test definition on our website for details regarding regions or types of variants that are covered or excluded for this test. This report reflects the analysis of an extracted genomic DNA sample. While this test is intended to reflect the analysis of extracted genomic DNA from a referred patient, in very rare cases the analyzed DNA may not represent that individual's constitutional genome, such as in the case of a circulating hematolymphoid neoplasm, bone marrow transplant, blood transfusion, chimerism, culture artifact or maternal cell contamination. Genomic DNA derived from tissue cultured for extended periods of time are at increased risk for cell culture artifacts. Interpretations are made on the assumption that any clinical information provided, including specimen identity, is accurate. RNA analysis is not designed for use as a stand-alone diagnostic method and cannot determine absolute RNA levels. Results from the RNA analysis may not be informative for interpreting copy number events. Additionally, sensitivity to detect RNA splicing events may be reduced in regions that do not meet RNA detection thresholds, which may include variants in the first donor site of each gene and complex events, among others.

ATM: Sequencing analysis for exons 6, 24, 43 includes only cds +/- 10 bp. NF1: Sequencing analysis for exons 2, 7, 25, 41, 48 includes only cds +/- 10 bp. PTEN: Sequencing analysis for exons 8 includes only cds +/- 10 bp.

Disclaimer

DNA studies do not constitute a definitive test for the selected condition(s) in all individuals. It should be realized that there are possible sources of error. Errors can result from trace contamination, rare technical errors, rare genetic variants that interfere with analysis, recent scientific developments, and alternative classification systems. This test should be one of many aspects used by the healthcare provider to help with a diagnosis and treatment plan, but it is not a diagnosis itself. This test was developed and its performance characteristics determined by Labcorp Genetics. It has not been cleared or approved by the FDA. The laboratory is regulated under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high-complexity clinical tests (CLIA ID: 05D2040778). This test is used for clinical purposes. It should not be regarded as investigational or for research.

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Genes analyzed

ATM*, BARD1, BRCA1, BRCA2, CDH1, CHEK2, NF1*, PALB2, PTEN*, RAD51C, RAD51D, STK11, TP53

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This report has been released utilizing a validated procedure approved by:

Jeana DaRe

Jeana DaRe, Ph.D., FACMG
Laboratory Director

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GENERAL GUIDELINES
NEGATIVE RESULTS GUIDE

This document is not part of the Invitae® clinical report and does not represent medical advice. These are general guidelines that are not specific to your result and may not represent all relevant international recommendations. You can use this guide to talk to your healthcare provider about your test results, clinical history, and the most current guidelines. We recognize that individuals have diverse gender and sexual identities. In this guide, the terms female, male, women, and men refer to sex assigned at birth.

What is a negative result?



A negative test result means that no significant genetic changes ("pathogenic" or "likely pathogenic" variants) were found. Risk for disease can still be influenced by a combination of personal, lifestyle, environmental, and/or unidentified genetic factors.

Create a plan with a healthcare provider



It is important to share these results with a healthcare provider to determine appropriate next steps. The chance for an individual to develop a disease is not usually determined by genetic test results alone.

What does this result mean for family members?



Parents, siblings, children, and other relatives have their own genetic makeup. Although these results did not find a significant genetic change, family members can discuss their own potential health and/or reproductive risks and the option of genetic testing with their own healthcare providers.

Resources



Genetic counseling can help individuals understand their genetic test results and options for next steps. Reviewing test results with a genetic counselor or other healthcare provider is recommended. Local or telehealth genetic counselors can be identified using the Find a Genetic Counselor search tool at nsgc.org (US and Canada). Individuals with an Invitae test result can also log in to their patient portal (invitae.com) to view their results, contact a genetic counselor, or join the Invitae Patient Insights Network (PIN) (pin.invitae.com), an online platform where individuals can share information about their health and experiences to help advance research and drug development.

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