Integrated BRACAnalysis® with MyRisk™ Hereditary Cancer Test

MyRisk Genetic Result

MyRisk™ Hereditary Cancer Test

RECEIVING HEALTHCARE PROVIDER

Test HCP, MD
Test Medical Center
6609 BLANCO RD STE 200
SAN ANTONIO, TX 78216

SPECIMEN

Specimen Type:	Blood
Draw Date:	Apr 22, 2024
Accession Date:	Apr 22, 2024
Report Date:	Apr 22, 2024

PATIENT

Legal Name:	Pt Last Name, Pt First Name
Date of Birth:	Apr 22, 1989
Patient ID:	Patient id
Sex at Birth:	F
Accession #:	07283333-BLD
Requisition #:	47624589

GENETIC RESULT: POSITIVE - CLINICALLY SIGNIFICANT MUTATION IDENTIFIED

Note: "CLINICALLY SIGNIFICANT," as defined in this report, is a genetic change that is associated with the potential to alter medical intervention.

CLINICAL HISTORY ANALYSIS: NO ADDITIONAL MANAGEMENT GUIDELINES IDENTIFIED BASED ON THE CLINICAL HISTORY PROVIDED

Other clinical factors may influence individualized management. This analysis may be incomplete if details about cancer diagnoses, ages, family relationships or other factors were omitted or ambiguous. If this patient also has a clinically significant mutation, the recommendations based on the clinical history analysis should be considered in light of the possibility that this mutation explains all or some of the cancer history in the family.

GENE	MUTATION	INTERPRETATION
BRCA2	c.32_33delinsA (p.Phe11Tyrfs*14) Heterozygous	**High Risk** This patient has *BRCA2*-associated hereditary breast and ovarian cancer syndrome.

DETAILS ABOUT: *BRCA2* c.32_33delinsA (p.Phe11Tyrfs*14): NM_000059.3; (aka: 260delTTinsA)

Functional Significance: Deleterious - Abnormal Protein Production and/or Function

The heterozygous germline *BRCA2* mutation c.32_33delinsA is predicted to result in the premature truncation of the *BRCA2* protein at amino acid position 24 (p.Phe11Tyrfs*14).

Clinical Significance: High Risk

This mutation is associated with increased cancer risk and should be regarded as clinically significant.

ADDITIONAL FINDINGS: NO VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) IDENTIFIED

Details About Non-Clinically Significant Variants: All individuals carry DNA changes (i.e., variants), and most variants do not increase an individual's risk of cancer or other diseases. When identified, variants of uncertain significance (VUS) are reported. Likely benign variants (Favor Polymorphisms) and benign variants (Polymorphisms) are not reported and available data indicate that these variants most likely do not cause increased cancer risk. Present evidence does not suggest that non-clinically significant variant findings be used to modify patient medical management beyond what is indicated by the personal and family history and any other clinically significant findings.

Variant Classification: Myriad's myVision™ Variant Classification Program performs ongoing evaluations of variant classifications. In certain cases, healthcare providers may be contacted for more clinical information or to arrange family testing to aid in variant classification. When new evidence about a variant is identified and determined to result in clinical significance and management change, that information will automatically be made available to the healthcare provider through an amended report.

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ADDITIONAL INFORMATION

Genes Analyzed: Sequencing (seq) and large rearrangement analyses were performed for all coding exons in the following genes, unless otherwise indicated:
APC, *ATM*, *AXIN2*, *BAP1*, *BARD1*, *BMPR1A*, *BRCA1*, *BRCA2*, *BRIP1*, *CDH1*, *CDK4*, *CDKN2A*, *CHEK2*, *CTNNA1*, *FH*, *FLCN*, *HOXB13* (seq only), *MEN1*, *MET*, *MLH1*, *MSH2*, *MSH3* (excluding repetitive portions of exon 1), *MSH6*, *MUTYH*, *NTHL1*, *PALB2*, *PMS2*, *PTEN*, *RAD51C*, *RAD51D*, *SDHA*, *SDHB*, *SDHC*, *SDHD*, *SMAD4*, *STK11*, *TP53*, *TSC1*, *TSC2*, *VHL*.
Limited promoter regions may also be analyzed for large rearrangements.

Sequencing (seq) and/or large rearrangement (LR) analyses were performed only for the gene portions indicated in parenthesis for the following genes:
EGFR (exons 18-21, seq and LR), *EPCAM* (exons 8-9, LR only), *GREM1* (exon 1 and upstream regulatory regions, LR only), *MITF* (c.952, seq only), *POLE* (exonuclease domain, seq only), *POLD1* (exonuclease domain, seq only), *RET* (exons 5, 8, 10, 11, 13-16 seq and LR), *TERT* (promoter region 71 bases upstream of the translation start, c.-71_-1, seq only).

** Other genes not analyzed with this test may also be associated with cancer.

Indication for Testing: It is our understanding that this individual was identified for testing due to a personal or family history suggestive of a hereditary predisposition for cancer.

Patient Information: Sex assigned at birth is a label given to an individual at birth, typically "male" or "female". In this report, the terms "male", "female", "he", "she", "woman", and "man" refer to sex assigned at birth.

Associated Cancer Risks and Clinical Management: The "MyRisk Management Tool" associated with this report provides a summary of cancer risk and professional society medical management guidelines that may be useful in developing a plan for this patient based on any clinically significant test results and/or reported personal/family history. In some cases, a MyRisk Management Tool cannot be provided, such as when the result has a special interpretation or includes a mutation with unusual characteristics.

Analysis Description: The Technical Specifications summary (myriad.com/technical-specifications) describes the analysis, method, performance, nomenclature, and interpretive criteria of this test. Current testing technologies are unable to definitively determine whether a variant is germline or somatic in origin, which may significantly impact risk estimates and medical management; therefore, these results should be correlated with this patient's personal and family history. The interpretation of this test may also be impacted if the patient has a hematologic malignancy or an allogeneic bone marrow transplant.

CLASSIFICATION DISCLAIMER

THE CLASSIFICATION AND INTERPRETATION OF ALL VARIANTS IDENTIFIED IN THIS ASSAY REFLECTS THE CURRENT STATE OF MYRIAD'S SCIENTIFIC UNDERSTANDING AT THE TIME THIS REPORT WAS ISSUED. VARIANT CLASSIFICATION AND INTERPRETATION MAY CHANGE FOR A VARIETY OF REASONS, INCLUDING BUT NOT LIMITED TO, IMPROVEMENTS TO CLASSIFICATION TECHNIQUES, AVAILABILITY OF ADDITIONAL SCIENTIFIC INFORMATION, AND OBSERVATION OF A VARIANT IN MORE PATIENTS.

Please contact Myriad Medical Services at 1-800-469-7423 X 3850 to discuss any questions regarding this result.

These test results should only be used in conjunction with the patient's clinical history and any previous analysis of appropriate family members. The patient's clinical history and test results should not be disclosed to a third party, unless related to treatment or payment for treatment, without the patient's express written authorization. It is strongly recommended that these results be communicated to the patient in a setting that includes appropriate genetic consultation. This test was developed and its performance characteristics determined by Myriad Genetic Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that clearance or approval for laboratory-developed tests is not required.

This **Authorized Signature** pertains to this laboratory report:

Benjamin B. Roa, PhD
Diplomate ABMGG
Laboratory Director

Genetic testing was completed by CLIA and CAP accredited laboratories in the United States located at: 320 Wakara Way Salt Lake City, UT 84108 CLIA IDs: 46D0880690
The following personnel codes and laboratory director signature may reflect remote review of digital data: 2987, 4104, 4476

Myriad genetics

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Clinical & Cancer Family History Information

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PERSONAL / FAMILY CANCER HISTORY SUMMARY

FAMILY MEMBER	CANCER / CLINICAL DIAGNOSIS	AGE AT DIAGNOSIS
Patient	None	
Mother	Breast, Invasive	47
Aunt Maternal	Breast, Invasive	40
Grandmother Paternal	Breast, Invasive	60

PATIENT CLINICAL HISTORY SUMMARY

Patient's age	35	Hormone Replacement Therapy (HRT)	No
Ancestry	White/Non-Hispanic	- HRT: Treatment Type	N/A
Height	5 ft 7 in	- HRT: Current user	N/A
Weight	175 lbs	- Number of years ago started	N/A
Age of menarche	13	- Additional years of intended use	N/A
Patient's menopausal status	Pre-menopausal	- HRT: Past user	N/A
- Age of onset	N/A	- Number of years ago ended	N/A
Age of first live birth	27	Breast biopsy	Not Specified
NUMBER OF PATIENT'S FEMALE RELATIVES		**MAMMOGRAPHIC DENSITY**	
Daughters	1	Breast Density Assessed	Not Specified
Sisters	2	Volpara® Volumetric Density	Not Specified
Maternal Aunts	2	Vas Percentage Density	Not Specified
Paternal Aunts	2	BI-RADS® ATLAS Density	Not Specified

Myriad genetics

Clinical & Cancer Family History Information

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The clinical information displayed here was provided by a qualified healthcare provider on the Test Request Form and other documents, and was not verified by Myriad. Female relatives refers to sex assigned at birth, which is a label given to an individual at birth, typically "male" or "female". Family members listed as "other" are not included in a Tyrer-Cuzick breast cancer risk estimate or other personal/family history assessments. For more information see the Specifications for Personal/Family History Analysis at http://myriad.com/technical-specifications.

The accuracy and completeness of the information provided in the Clinical and Cancer Family History Information section of the report (e.g. height and weight, age of menarche) may significantly affect the accuracy of breast cancer risk estimates provided based on either Tyrer-Cuzick or RiskScore.

RiskScore is not valid, and may significantly over- or under-estimate breast cancer risk for individuals who do not meet the eligibility criteria in effect when the testing was performed. The current criteria are: 1) sex assigned at birth is female 2) age is 18 to 84 years, 3) no personal history of breast cancer, LCIS, hyperplasia (with or without atypia), or a breast biopsy with unknown results, 4) there is no mutation detected in a breast cancer risk gene (other than a monoallelic *CHEK2* mutation in a White/Non-Hispanic or Ashkenazi Jewish individual), 5) the individual's relatives have not been found to have a mutation in a high-penetrance breast cancer risk gene and 6) the sample was submitted with a current Test Request Form and the ordering healthcare provider has not determined that RiskScore is inappropriate for the patient. If this is an amended report for a patient tested in the past, please refer to the MyRisk Technical Specifications at http://myriad.com/technical-specifications for the eligibility criteria in effect at the time of the original testing.

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CLINICAL HISTORY ANALYSIS: NO ADDITIONAL MANAGEMENT GUIDELINES IDENTIFIED BASED ON THE CLINICAL HISTORY PROVIDED

Other clinical factors may influence individualized management. This analysis may be incomplete if details about cancer diagnoses, ages, family relationships or other factors were omitted or ambiguous. If this patient also has a clinically significant mutation, the recommendations based on the clinical history analysis should be considered in light of the possibility that this mutation explains all or some of the cancer history in the family.

GENE	MUTATION	THIS GENETIC TEST RESULT IS ASSOCIATED WITH THE FOLLOWING CANCER RISKS:
BRCA2	c.32_33delinsA (p.Phe11Tyrfs*14) Heterozygous	HIGH RISK: Breast, Ovarian, Pancreatic
		ELEVATED RISK: Skin

Please see the Genetic Test Result for more details on any variant(s) detected in this patient, including variant classification information.

The terms "male", "female", "he", "she", "women", and "men" refer to sex assigned at birth.

ADDITIONAL FINDINGS: NO VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) IDENTIFIED

TYRER-CUZICK BREAST CANCER RISK CALCULATION

REMAINING LIFETIME BREAST CANCER RISK: Not Calculated	5-YEAR BREAST CANCER RISK: Not Calculated

The Tyrer-Cuzick breast cancer risk estimate is only calculated for individuals who meet the following criteria: 1) sex assigned at birth is female 2) age is younger than 85 years, 3) no known mutation or inconclusive result has been found in the individual or any of their relatives, and 4) the sample was submitted with a current Test Request Form that includes all of the fields required to collect the information used in the calculation, and the provider has not indicated on the Test Request Form that the Tyrer-Cuzick calculation is not appropriate for the patient. Version 8.0 of the Tyrer-Cuzick model was used for this risk estimate. Tyrer-Cuzick model is available for download at the EMS-Trials website, http://www.ems-trials.org/riskevaluator.

CLINICAL OVERVIEW OF GENETIC FINDINGS

BRCA2-associated hereditary breast and ovarian cancer syndrome

- This patient has been found to have a mutation in the *BRCA2* gene. Individuals with mutations in *BRCA2* have *BRCA2*-associated hereditary breast and ovarian cancer syndrome.
- Women with *BRCA2* mutations have a risk for breast cancer that is greatly increased over the 12.5% lifetime risk for women in the general population of the United States.

- Women with *BRCA2* mutations also have high risks for ovarian, fallopian tube, and primary peritoneal cancer.
- Men with *BRCA2* mutations have a high risk for breast cancer and prostate cancer. The increase in prostate cancer risk is most significant at younger ages. Additionally, men with a *BRCA2* mutation have a higher risk for an aggressive prostate cancer.
- Male and female patients with *BRCA2* mutations also have a high risk for exocrine pancreatic cancer. These are cancers developing in the enzyme-secreting cells of the pancreas.
- Male and female patients with *BRCA2* mutations also have an elevated risk for melanomas of both the skin and eyes.
- Although there are high cancer risks for patients with *BRCA2* mutations, there are interventions that have been shown to be effective at reducing many of these risks. Guidelines from the National Comprehensive Cancer Network (NCCN) for the medical management of patients with *BRCA2* mutations are listed below. It is recommended that patients with *BRCA2* mutations be managed by a multidisciplinary team with experience in the prevention and treatment of the cancers associated with *BRCA2* mutations.

WHAT ARE THE PATIENT'S CANCER RISKS?

These risk tables show the clinically significant cancer risks identified as part of this patient's testing. Testing for some patients does not include some of the analyses listed:

- GENETIC RESULT: Mutations detected in any of the hereditary cancer genes included on the MyRisk panel.
- BREAST CANCER RISKSCORE: RiskScore estimate of remaining lifetime breast cancer risk if 20% or greater
- CLINICAL HISTORY ANALYSIS for breast cancer risk: Tyrer-Cuzick model estimate of remaining lifetime breast cancer risk of 20% or greater
- CLINICAL HISTORY ANALYSIS for breast, colorectal, pancreatic, prostate and melanoma cancer: Analysis of the patient's personal and family history.

The risks for each of these results are provided separately. If the risk for any individual cancer is affected by more than one of these results, the risk associated with each finding is listed in a separate table. At this time, there is not enough information to estimate risks for cancers affected by more than one gene mutation, or risks based on both gene mutations and personal/family history.

Risks Due to *BRCA2*-associated hereditary breast and ovarian cancer syndrome

CANCER TYPE	CANCER RISK	RISK FOR GENERAL POPULATION	RELATED TO
FEMALE BREAST			
To age 50	17%-35%	2.1%	*BRCA2*
To age 70	38%-84%	7.5%	*BRCA2*
Second primary within 5 years of first breast cancer diagnosis	4%-9%	1.6%	*BRCA2*
OVARIAN			
To age 50	0.4%-4%	0.2%	*BRCA2*
To age 70	15%-27%	0.6%	*BRCA2*
Ovarian cancer within 10 years of a breast cancer diagnosis	6.8%	<1.0%	*BRCA2*
PANCREATIC			
To age 80	7%, or higher if there is a family history of pancreatic cancer.	1.1%	*BRCA2*

MyRisk Management Tool

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Risks Due to *BRCA2*-associated hereditary breast and ovarian cancer syndrome

CANCER TYPE	CANCER RISK	RISK FOR GENERAL POPULATION	RELATED TO
MELANOMA			
To age 80	Elevated risk for melanomas of both the skin and eye	1.6%	*BRCA2*

WHAT MANAGEMENT FOR CANCER RISKS SHOULD BE CONSIDERED?

This overview of clinical management guidelines is based on the patient's genetic test results and the Clinical History Analysis. Medical management guidelines are summarized from established medical societies, primarily the National Comprehensive Cancer Network (NCCN). If there are overlapping management guidelines for any individual cancer due to more than one of these results, the guidelines associated with each finding are listed in separate tables, even if they are the same. At this time, there are no medical society guidelines for how to adjust management when there are multiple sources of risk, such as from more than one gene mutation, or a mutation and a personal/family history of cancer. In these cases, it may be appropriate to use the most aggressive of the management options provided.

The overview provided below should not be used as the sole source of information to determine medical management. The references cited should always be consulted for more details and updates to the recommendations.

No information is provided related to treatment of a previous or existing cancer or polyps. The recommendation summaries below may require modification due to the patient's personal medical history, past surgeries and other treatments. Patients with a past history of cancer, benign tumors, or pre-cancerous findings may be candidates for long term surveillance and risk-reduction strategies beyond what is necessary for the treatment of their initial diagnosis. Any discussion of medical management options is for general information purposes only and does not constitute a recommendation. While genetic testing and medical society recommendations provide important and useful information, medical management decisions should be made in consultation between each patient and his or her healthcare provider.

Management Options for *BRCA2*-associated hereditary breast and ovarian cancer syndrome

PROCEDURE	AGE TO BEGIN	FREQUENCY Unless otherwise indicated by findings	RELATED TO
FEMALE BREAST			
Breast awareness - Women should be familiar with their breasts and promptly report changes to their healthcare provider. Periodic, consistent breast self-examination (BSE) may facilitate breast awareness. [2]	18 years	NA	*BRCA2*
Clinical breast examination [2]	25 years	Every 6 to 12 months	*BRCA2*
Breast MRI with contrast [2]	25 years, or individualized to a younger age if a relative has been diagnosed younger than age 30.	Annually	*BRCA2*
Mammography [2]	30 years. If MRI unavailable, start at 25 years, or individualized to a younger age if a relative has been diagnosed younger than age 30.	Annually	*BRCA2*
Consider risk-reducing mastectomy. [2]	Individualized	NA	*BRCA2*
Consider options for breast cancer risk-reduction agents (i.e. tamoxifen). [2]	Individualized	NA	*BRCA2*

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Management Options for *BRCA2*-associated hereditary breast and ovarian cancer syndrome

PROCEDURE	AGE TO BEGIN	FREQUENCY Unless otherwise indicated by findings	RELATED TO
OVARIAN			
Bilateral salpingo-oophorectomy [2]	35 to 45 years, upon completion of childbearing	NA	*BRCA2*
Consider options for ovarian cancer risk-reduction agents (i.e. oral contraceptives). [2,5]	Individualized	NA	*BRCA2*
PANCREATIC			
For patients with a family history of pancreatic cancer, consider available options for pancreatic cancer screening, including the possibility of endoscopic ultrasonography (EUS) and MRI/magnetic resonance cholangiopancreatography (MRCP). It is recommended that patients who are candidates for pancreatic cancer screening be managed by a multidisciplinary team with experience in screening for pancreatic cancer, preferably within research protocols. [3]	Age 50, or 10 years younger than the earliest age of pancreatic cancer diagnosis in the family	Annually	*BRCA2*
Provide education about ways to reduce pancreatic cancer risk, such as not smoking and losing weight. [8]	Individualized	Individualized	*BRCA2*
MELANOMA			
Whole-body skin and eye examinations, and education about minimizing exposure to UV radiation. [2]	Individualized	Annually	*BRCA2*
FOR PATIENTS WITH A CANCER DIAGNOSIS			
For patients with a gene mutation and a diagnosis of cancer, targeted therapies may be available as a treatment option for certain tumor types (e.g., platinum chemotherapy, PARP-inhibitors) [1,4,6,7,8,9,10]	NA	NA	*BRCA2*

1. Armstrong DK, et al. NCCN Clinical Practice Guidelines in Oncology®: Ovarian Cancer Including Fallopian Tube Cancer and Primary Peritoneal Cancer. V 1.2023. Dec 22. Available at https://www.nccn.org.
2. Daly M et al. NCCN Clinical Practice Guidelines in Oncology®: Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic. V 3.2023. Feb 13. Available at https://www.nccn.org.
3. Goggins M, et al. Management of patients with increased risk for familial pancreatic cancer: updated recommendations from the International Cancer of the Pancreas Screening (CAPS) Consortium. Gut. 2020 69:7-17. PMID: 31672839.
4. Gradishar WJ et al. NCCN Clinical Practice Guidelines in Oncology®: Breast Cancer. V 4.2023. Mar 23. Available at https://www.nccn.org.
5. Gupta S, et al. NCCN Clinical Practice Guidelines in Oncology® Genetic/Familial High-Risk Assessment: Colorectal. V 1.2023. May 30. Available at https://www.nccn.org.
6. Schaeffer E, et al. NCCN Clinical Practice Guidelines in Oncology®: Prostate Cancer. V 1.2023. Sep 16. Available at https://www.nccn.org.
7. Tempero MA, et al. NCCN Clinical Practice Guidelines in Oncology®: Ampullary Adenocarcinoma. V 2.2022. Dec 6. Available at https://www.nccn.org.
8. Tempero MA, et al. NCCN Clinical Practice Guidelines in Oncology®: Pancreatic Adenocarcinoma. V 2.2022. Dec 6. Available at https://www.nccn.org.
9. https://www.accessdata.fda.gov/drugsatfda_docs/label/2016/209115s000lbl.pdf
10. https://www.accessdata.fda.gov/drugsatfda_docs/label/2018/208558s002lbl.pdf

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Notes for Personalized Management:

INFORMATION ON HOW CANCER RISKS AND MANAGEMENT ARE DETERMINED

The MyRisk Management Tool provides cancer risk levels based on analysis of genetic test results (see MyRisk Genetic Result) and a summary of medical society management recommendations based on both the genetic test results and a limited analysis of the patient's clinical history related to the risk for breast, colorectal, prostate, melanoma and pancreatic cancers. Here are some important points to understand as you interpret this test report and decide on the best plan for management:

- Comprehensive patient management. The management recommendations presented in this report are a summary of management options recommended by the National Comprehensive Cancer Network (NCCN) and other established medical societies and are general in nature. The patient's actual management should be modified based on personal medical history, surgeries and other treatments. A comprehensive risk assessment and management plan may take into account this report and other aspects of the patient's personal/family medical history (e.g., all known clinical diagnoses), as well as lifestyle, environmental and other factors.
- Risk estimates based on provider-supplied information. Some of the risk estimates and management recommendation summaries provided in this report are based on our interpretation of information supplied by the ordering health care provider on the test request form (see Specifications for Personal/Family History analysis at myriad.com/technical-specifications). The patient's actual risks and appropriate management may be significantly different if details provided for cancer diagnoses, ages, family relationships or other factors were incorrect, omitted, ambiguous or have since changed. Please review the clinical history listed on the Clinical & Family History Information page of this report to make sure that the information used was provided and interpreted correctly.
- Variability in Tyrer-Cuzick risk estimates. Tyrer-Cuzick estimates of breast cancer risk can vary significantly based on the way in which the model is used, and the estimate provided here may be higher or lower than what would be calculated by other users. For complete details of how Myriad calculates Tyrer-Cuzick risk estimates, including how Myriad handles information provided in a format not compatible with the model, please see the Specifications for Personal/Family History analysis at myriad.com/technical-specifications. These Specifications also include information for recalculating the Tyrer-Cuzick breast cancer risk estimate if desired.
- What is meant by "High Risk" and "Elevated Risk"? In the Genetic Test Result Summary, a gene-associated cancer risk is described as "High Risk" for a cancer type if all of the following conditions are met: the absolute risk of cancer is approximately 5% or higher, the increase in risk over the general population is approximately 2 to 3-fold or higher, and there is significant data from multiple studies supporting the cancer risk estimate. A gene is described as "Elevated Risk" for a cancer type if there is sufficient data to support an increase in cancer risk over the general population risk, but not all criteria for "High Risk" are met.

INFORMATION FOR FAMILY MEMBERS

Family members should talk to their healthcare providers about hereditary cancer testing to help define their own risk and assist in the interpretation of this patient's genetic test result.

- This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for individuals who have this/these mutation(s) are provided below.
- **Family members should talk to a healthcare provider about genetic testing.** Close relatives such as parents, children, and siblings have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, parents siblings, and grandparents also have a chance for carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention. More resources for family testing are available at MySupport360.com.

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Additional Information for *BRCA2*-associated hereditary breast and ovarian cancer syndrome

- In rare instances, an individual may inherit mutations in both copies of the *BRCA2* gene, leading to the condition Fanconi anemia, complementation group D1 (FANCD1). This condition is rare and includes physical abnormalities, growth retardation, progressive bone marrow failure and a high risk for cancer. The children of this patient are at risk of inheriting FANCD1 only if the other parent is also a carrier of a *BRCA2* mutation. Screening the other biological parent of any children for *BRCA2* mutations may be appropriate.
- Parents who are concerned about the possibility of passing on a *BRCA2* mutation to a future child may want to discuss options for prenatal testing and assisted reproduction techniques, such as pre-implantation genetic diagnosis (PGD).

CANCER RISK FOR *BRCA2* CLINICALLY SIGNIFICANT MUTATION

CANCER TYPE	CANCER RISK	RISK FOR GENERAL POPULATION
	FEMALES	
FEMALE BREAST		
To age 50	17%-35%	2.1%
To age 70	38%-84%	7.5%
Second primary within 5 years of first breast cancer diagnosis	4%-9%	1.6%
OVARIAN		
To age 50	0.4%-4%	0.2%
To age 70	15%-27%	0.6%
Ovarian cancer within 10 years of a breast cancer diagnosis	6.8%	<1.0%
	MALES	
MALE BREAST		
To age 70	6.8%	0.1%
PROSTATE		
To age 70	20%	6.3%
	FEMALES AND MALES	
PANCREATIC		
To age 80	7%, or higher if there is a family history of pancreatic cancer.	1.1%
MELANOMA		
To age 80	Elevated risk for melanomas of both the skin and eye	1.6%

Please contact Myriad Medical Services at 1-800-469-7423 X 3850 to discuss any questions regarding this result.

END OF MANAGEMENT TOOL