Quest Advanced™ Oncology

Hereditary Cancer Genetic Test Results

This report is intended to facilitate a discussion between providers and their patients.

INFORMATION FOR INDIVIDUALS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT IN THE *BRCA2* GENE

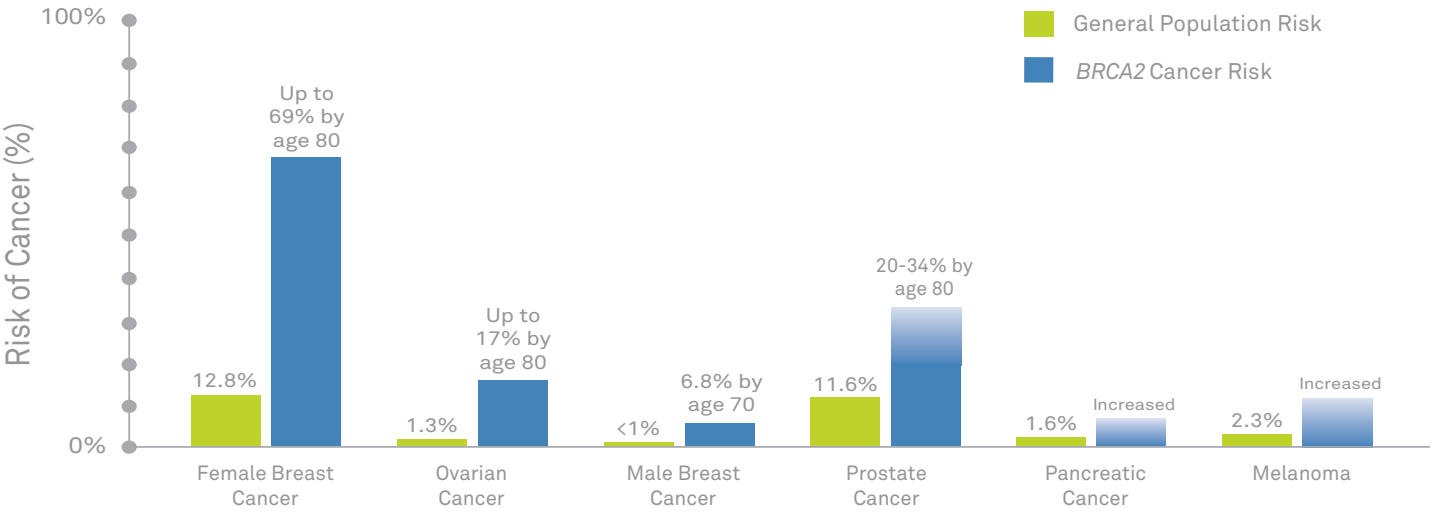
What this result means

Individuals who have a pathogenic or likely pathogenic variant (sometimes called a mutation) in the *BRCA2* gene have BRCA-related breast and/or ovarian cancer syndrome. Individuals with BRCA-related breast and/or ovarian cancer syndrome have a higher-than-average chance to develop the following cancers: female breast, ovarian, male breast, prostate, pancreatic, and melanoma.

Cancer risk

The graph below compares general population cancer risks to the potential cancer risks associated with pathogenic variants in the *BRCA2* gene. Individual cancer risks may be higher or lower depending on the specific variant identified in addition to each individual's gender, age, medical history, and family history. Not everyone with a pathogenic or likely pathogenic variant will develop cancer.

Information about cancer risks related to pathogenic variants in *BRCA2* may change over time, so it is important for the ordering healthcare provider, genetic counselor, and patient to keep in contact regarding this result.



*Data on file.

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Options for managing cancer risk

There are options for cancer prevention and early detection. The following are general guidelines for individuals who have a *BRCA2* pathogenic variant. These guidelines are evolving and are not specific to any one individual. Each individual's gender, age, medical history, family history, quality of life goals, reproductive desires, general health status, and other medical information should be taken into account when developing a medical management plan.

	Considerations for cancer prevention/early detection	Age to begin	Frequency
Female breast cancer	Breast awareness	18 years	Periodic, consistent
	Clinical breast exam	25 years	Every 6–12 months
	Breast MRI with contrast	25 years or modified if there is a family history of breast cancer before age	Annual
	Mammogram with consideration of tomosynthesis	30 years	Annual
	Risk-reducing mastectomy (RRM)	Option to discuss during counseling	—
	Risk-reducing medications	Option to discuss during counseling	—
Ovarian cancer	Risk-reducing salpingo oophorectomy (RRSO)	35–40 years; dependent upon family history and childbearing; may consider delaying until 40–45 years given later age of onset in carriers	—
	Consider transvaginal ultrasound combined with serum CA-125	30–35 years; for women who decide against RRSO, after discussion with physician	—
Male breast cancer	Breast self-exam training and education	35 years	—
	Clinical breast exam	35 years	Every 12 months
	Consider mammogram in men with gynecomastia	50 years or 10 years earlier than the earliest diagnosis of male breast cancer in the family, whichever is first	Annual
Prostate cancer	Prostate cancer screening	40 years	—
Pancreatic cancer	Consider screening for individuals with one or more first-or second-degree relatives with exocrine pancreatic cancer on the same side of the family as the pathogenic variant	50 years or 10 years earlier than the youngest diagnosis of exocrine pancreatic cancer in the family, whichever is first	Annual, or more frequent depending on screening results
	Screening should be performed in experienced high-volume centers, ideally under research conditions		

	Considerations for cancer prevention/early detection	Age to begin	Frequency
Melanoma	General melanoma risk management is appropriate, such as annual full-body skin examination and minimizing UV exposure		
Other Cancers	Education regarding signs and symptoms of cancer(s)		

Source: National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology. Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V2.2021. **www.NCCN.org**

What this result means for family members

Family members could have the same *BRCA2* variant that was identified in this individual. Parents, brothers, sisters, and children may each have a 50% chance of having the same variant. Other blood relatives also have an increased risk for the variant. It is important to share these test results with family members to allow each of them to decide if they want to be tested. Some family members may only need testing for this one *BRCA2* variant, while other relatives may need a more comprehensive test with multiple genes. Children of parents who both have a *BRCA2* variant are at risk for Fanconi anemia type D1. A genetic counselor or other healthcare provider can help determine the most appropriate testing options.

Reproductive information

Individuals interested in family planning should speak to their doctor and/or genetic counselor to discuss reproductive options. This may include discussion of prenatal diagnosis or pre-implantation genetic testing.

Risk assessment and counseling: an important first step

A genetic counselor or other qualified healthcare professional can help explain test results and what they mean for a patient and family members. A team of specialized Quest genetic counselors or clinical geneticists is available to speak with healthcare providers about test results by calling 1.866.GENE.INFO. Patients can access a directory of independent genetic counselors at **FindAGeneticCounselor.com**.

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Creating a plan: a checklist for patients

- ☐ Get a copy of your genetic test results.
- ☐ Talk with your healthcare provider about what this result means and the things you can do to manage your risk.
- ☐ Ask your healthcare provider if additional genetic testing may benefit you.
- ☐ Share your test results with your family members and give them a copy. Their healthcare provider will need this information in order to provide them with the most accurate risk assessment.
- ☐ Talk with your healthcare provider regularly so that you know about any important changes in genetic testing and cancer screening options. Be sure to let him/her know of any changes in your family history, including family members' genetic test results.
- ☐ Consider talking to a genetic counselor about your results.

Research opportunities

Prospective Registry of MultiPlex Testing (PROMPT) PromptStudy.info

GenomeConnect: The ClinGen Patient Portal GenomeConnect.org

Additional resources

Facing Our Risk of Cancer Empowered (FORCE)
FacingOurRisk.org

Bright Pink®
BrightPink.org

Sharsheret®
Sharsheret.org

Foundation for Women's Cancer
FoundationforWomensCancer.org

Quest Hereditary Cancer Testing Solutions
QuestHereditaryCancer.com

National Society of Genetic Counselors
FindAGeneticCounselor.com

This information is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by a healthcare provider. Always talk with a healthcare provider about the meaning of genetic test results and before stopping, starting, or changing any medication or treatment.

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of this report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the healthcare provider's clinical evaluation. Inquiry regarding potential changes to the classification of the variant is strongly recommended prior to making any clinical decision. For questions regarding variant classification updates, please call Quest Diagnostics at 1.866.GENE.INFO (1.866.436.3463) to speak to a genetic counselor or laboratory director, or visit **QuestDiagnostics.com/VariantIQ**.