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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 *EPCAM* GENE

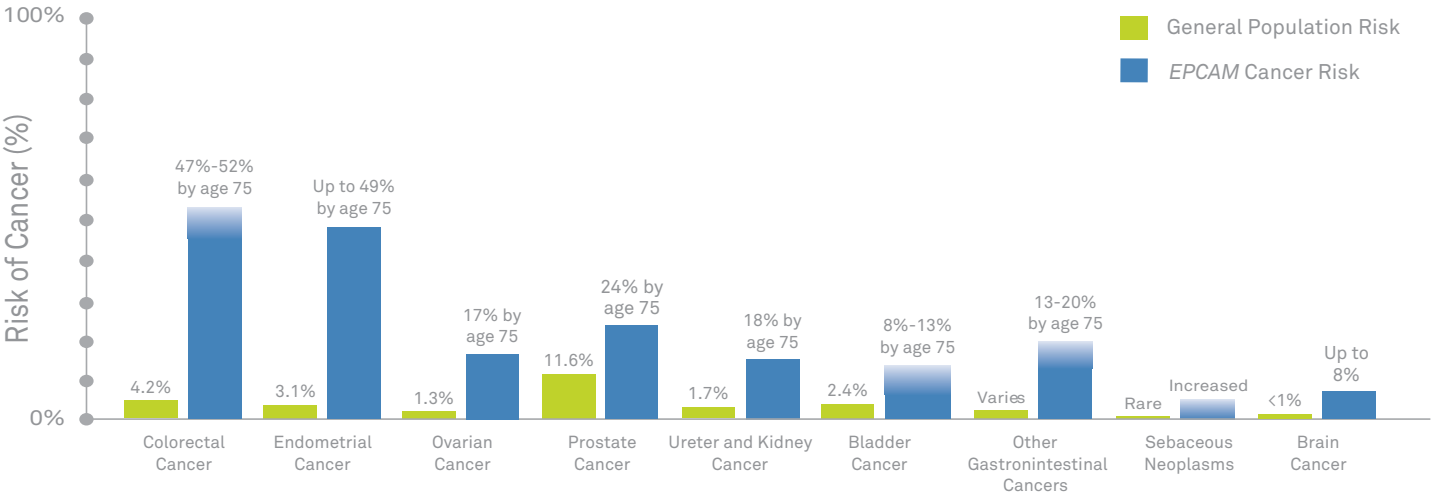
What this result means

Individuals who have a pathogenic or likely pathogenic deletion in the *EPCAM* gene have Lynch syndrome, previously called hereditary nonpolyposis colorectal cancer (HNPCC). Individuals with Lynch syndrome have a higher-than-average chance to develop the following cancers: colorectal, endometrial, ovarian, prostate, ureter, kidney, urinary bladder, stomach, small bowel, bile duct, gallbladder, pancreatic, sebaceous neoplasms, and brain cancer.

Cancer risk

The graph below compares general population cancer risks to the potential cancer risks associated with pathogenic variants in the *EPCAM* 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 *EPCAM* 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 pathogenic deletion in *EPCAM*. 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
Colorectal Cancer	Colonoscopy	20-25 years or 2-5 years prior to earliest diagnosis of colon cancer in the family, if diagnosed under 25 years	Every 1-2 years
	Aspirin	Discuss with healthcare provider	—
Endometrial Cancer	Hysterectomy	Individualized; dependent on family history, childbearing, gene-specific risk, and other medical condition(s)	—
	Endometrial biopsy can be considered	30-35 years; Discuss with healthcare provider	Every 1-2 years
	Transvaginal ultrasound	Insufficient evidence; Discuss with healthcare provider	—
	Risk-reducing agents (eg, birth control pills)	Discuss with healthcare provider	—
Ovarian Cancer	Surgical removal of ovaries and fallopian tubes	Individualized; dependent on family history, childbearing, gene-specific risk, menopausal status, and other medical condition(s)	—
	Transvaginal ultrasound and/or serum CA-125	Insufficient evidence; discuss with healthcare provider	—
	Risk-reducing agents (eg, birth control pills)	Discuss with healthcare provider	—
Prostate Cancer	Prostate cancer screening	Screening should be personalized based on family history, consider beginning PSA screening starting at age 40	Annual
Pancreatic Cancer	Consider pancreatic cancer screening in individuals with both a pathogenic or likely pathogenic variant and a first- or second-degree relative with pancreatic cancer	50 years or 10 years prior to the earliest diagnosis of pancreatic cancer in the family	—
Central Nervous System (CNS) Cancer	Patient education on signs of symptoms of CNS cancer and the importance of discussing any changes with their providers		
Skin Manifestations	Skin exam	Individualized	Every 1-2 years
Other Cancers	Individualized; discuss with healthcare provider		

Source: National Comprehensive Cancer Network. NCCN Clinical Practice Guidelines in Oncology. Genetic/Familial High-Risk Assessment: Colorectal. V1.2021, and Breast, Ovarian, and Pancreatic. V1.2022 **www.NCCN.org**

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What this result means for family members

Family members may have the same *EPCAM* deletion 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 *EPCAM* deletion, while other relatives may need a more comprehensive test with multiple genes. Children of parents who both have an *EPCAM* deletion (or pathogenic variant in the *MSH2* gene) are at risk for constitutional mismatch repair deficiency (CMMRD) syndrome. 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

Hereditary Colon Cancer Takes Guts hcctakesguts.org

Colorectal Cancer Alliance ccalliance.org

National Colorectal Cancer Roundtable nccrt.org

Quest Hereditary Cancer Testing Solutions QuestHereditaryCancer.com

Genetic Information Nondiscrimination Act (GINA) GINAhelp.org

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**.