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 *STK11* GENE

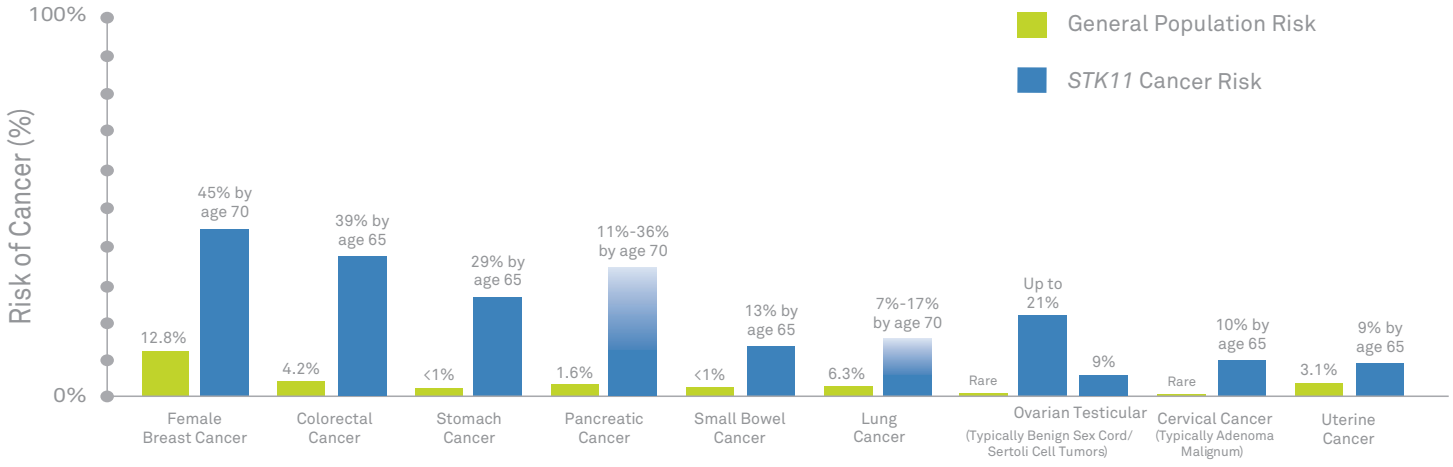
What this result means

Individuals who have a pathogenic or likely pathogenic variant (sometimes called a mutation) in the *STK11* gene have Peutz-Jeghers syndrome (PJS). Individuals with PJS have a higher-than-average chance to develop the following cancers: colorectal, stomach, pancreatic, small bowel, lung, cervical, uterine, and female breast cancer as well as sex cord tumors of the ovaries or testes. Other features may include characteristic freckling and Peutz-Jeghers type harmartomatous polyps of the gastrointestinal tract.

Cancer risk

The graph below compares general population cancer risks to the potential cancer risks associated with pathogenic variants in the *STK11* 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 *STK11* 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. Quest Diagnostics, 2022.

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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 an *STK11* pathogenic variant. These guidelines are evolving and are not specific to any one individual. A referral to an appropriate specialist may be considered. For more information, see the "Research opportunities" and "Additional resources" sections on the last page. 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	Mammogram	30 years	Annual
	Breast MRI with contrast	30 years	Annual
	Clinical Breast Exam	30 years	Every 6 months
	Discuss option of risk-reducing mastectomy		—
Colon Cancer	High quality colonoscopy	8-10 years	Every 2-3 years if polyps are found on initial exam, otherwise resume screening every 2-3 years at 18 or sooner if symptoms are present. Individualize frequency based on findings
Stomach Cancer	Upper endoscopy	8-10 years	Every 2-3 years if polyps are found on initial exam, otherwise resume screening every 2-3 years at 18 or sooner if symptoms are present. Individualize frequency based on findings
Small Bowel Cancer	Small bowel visualization	8-10 years	Based on findings of baseline, then every 2-3 years, and any time symptoms are present
Pancreatic Cancer	Magnetic resonance cholangiopancreatography with contrast or endoscopic ultrasound	30-35 years or consider 10 years younger than the earliest age of onset in the family	Annual
Gynecological Cancer/Tumors	Pelvic examination and Pap smear	18-20 years	Annual
	Physical exam for precocious puberty	8 years	Annual
Male Sex Cord Tumors	Testicular exam	10 years	Annual
Lung Cancer	Education about symptoms, smoking cessation	—	—
Other Management	Refer to multidisciplinary team of specialists familiar with PJS for evaluation and management; consider participation in clinical trials		

Source: National Comprehensive Cancer Network®. NCCN Clinical Practice Guidelines in Oncology. Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V1.2023. Genetic/Familial High-Risk Assessment: Colorectal. V1.2021. **www.NCCN.org**

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What this result means for family members

Family members may have the same *STK11* variant that was identified in this individual. Parents, brothers, sisters, and children 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 *STK11* variant, while other relatives may need a more comprehensive test with multiple genes. 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 (1.866.436.3463). 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 them 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

American College of Gastroenterology gi.org/guidelines

Hereditary Colon Cancer Takes Guts hcctakesguts.org

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