

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 *TSC2* GENE

What this result means

Individuals who have a pathogenic or likely pathogenic variant (sometimes called a mutation) in the *TSC2* gene have tuberous sclerosis complex (TSC). Individuals with tuberous sclerosis complex have a higher-than-average chance of developing tumors/cancers in the brain, heart, lungs, kidneys, and skin. Although many TSC-related tumors are not malignant, they may pose serious health risks.

Cancer risk

The table below lists the risks associated with pathogenic variants in the *TSC2* gene. Individual tumor/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 all of the tumors/cancers associated with *TSC2*.

Information about cancer risks related to pathogenic variants in *TSC2* may change over time, so it is important for the ordering healthcare provider, genetic counselor, and patient to keep in contact regarding this result.

Feature	Lifetime risk
Renal Cell Carcinoma	2%–4%
Nonmalignant tumors (brain, heart, lung, kidneys, and skin)	Increased
Neurological sequelae (seizures, elevated prevalence of autism spectrum disorders, and other TSC-associated neuropsychiatric disorders)	Increased

Options for managing cancer risk

Guidelines for cancer prevention and early detection are evolving. For more information, see the below “Additional resources” section. 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.

What this result means for family members

Family members may have the same *TSC2* 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 *TSC2* 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 preimplantation 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 are 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](https://www.findageneticcounselor.com).





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.
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Research opportunities

Prospective Registry of MultiPlex Testing (PROMPT) [PromptStudy.info](https://www.promptstudy.info)

GenomeConnect: The ClinGen Patient Portal
[GenomeConnect.org](https://www.genomeconnect.org)

Additional resources

National Comprehensive Cancer Network
Clinical Practice Guidelines in Oncology
(NCCN Guidelines): Kidney Cancer [NCCN.org](https://www.nccn.org)

Tuberous Sclerosis Complex International
[tscinternational.org](https://www.tscinternational.org)

Tuberous Sclerosis Alliance
[tsalliance.org](https://www.tsalliance.org)

Quest Hereditary Cancer Testing Solutions
[QuestHereditaryCancer.com](https://www.questhereditarycancer.com)

Genetic Information Nondiscrimination
Act (GINA) [GINAhelp.org](https://www.ginahelp.org)

National Society of Genetic Counselors
[FindAGeneticCounselor.com](https://www.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's 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 866.GENE.INFO (436.3463) to speak to a genetic counselor or laboratory director, or visit [QuestDiagnostics.com/VariantIQ](https://www.questdiagnostics.com/VariantIQ).

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