

Risk of Bone and Soft Tissue Sarcomas, Brain, and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *TP53* gene

What This Result Means

This result means you have a change (mutation) in the *TP53* gene. Mutations in this gene can cause Li-Fraumeni syndrome (LFS). People with LFS have an increased chance of certain cancers. The most frequent ones are breast cancer, osteosarcoma, and soft tissue sarcomas. Brain cancer, leukemia, adrenocortical carcinoma, and other types of cancer may also occur.

This result doesn't mean you will get cancer—not everyone with a mutation develops cancer. But you are at higher risk than you would be if you didn't have the mutation. The overall risk of cancer for men is 69%-95%. The overall risk for women is 83%-95%. Cancer can occur at any age, even in infancy.

Options for Managing Your Cancer Risk

Your test result shows that you have an increased risk for developing cancer. There are steps you can take to lower that risk. The options are explained briefly below. Talk with your healthcare provider to learn more and find out which is best for you.

- Annual skin exam
- Annual neurologic exam
- Annual whole body MRI

There may be other screening options. These would depend on your personal and family history.

Increased screening cannot prevent you from getting cancer. But it could detect cancer sooner. And, as you know, early detection improves your chances of survival.

Increased Cancer Screening

For Women

- Monthly breast self-exam
- Clinical breast exam every 6–12 months
- Annual breast MRI and/or mammogram

For Women and Men

- Annual comprehensive physical exam
- Colonoscopy every 2–5 years

Surgery

Preventive mastectomy (removal of the breasts) may be an option for women. It may lower the risk of cancer, but surgery is not for everyone.

What This Result Means for Your Family

You share genes in common with your blood relatives, so some of them may have the same mutation that you have. Your parents, brothers, sisters, and children each have a 50% chance of having it. Other blood relatives also have an increased risk.

Think about sharing your test results with your relatives. They might want to find out if they have the mutation too. If so, they should talk with their doctor and/or genetic counselor about testing. They could be tested just for the mutation that you have. Testing for a single mutation

INFORMATION ABOUT YOUR GENETIC TEST RESULT

What This Result Means for Your Family (continued)

may be the best option, as it may cost less. It will still let them know if they have the same mutation you have. In some cases, testing more genes might be suggested. Either way, testing will help them know more about their risk for cancer.

Relative	Risk of Having the Same Mutation
Identical twin	100%
Fraternal twin	50%
Parent, brother, sister, child	50%
Grandparent, uncle, aunt, niece, nephew	25%
First cousin	12.5%

Your Next Steps

- Get a copy of your test results.
- Talk with your doctor or genetic counselor about things you can do to manage your risk.
- Once you and your doctor have made a plan, set up appointments to start managing your risk.
- Think about sharing copies of your test results with your family members. That way, their doctor will know which test to order if they want to be tested.

- Talk with your doctor or genetic counselor regularly. They can keep you up to date about genetic testing and risk management options.

You can help researchers learn even more about this gene and its link to cancer risk. You can do this by participating in a research registry called PROMPT. If you would like to do this, please visit PROMPTStudy.org to learn more.

Additional Resources

- **Li-Fraumeni Syndrome Association**
LFSAssociation.org
- **Facing Our Risk of Cancer Empowered (FORCE)**
FacingOurRisk.org
- **Young Survival Coalition**
YoungSurvival.org
- **Bright Pink**
BrightPink.org
- **Sharsheret®**
Sharsheret.org
- **National Society of Genetic Counselors**
Find a Genetic Counselor
NSGC.org/p/cm/ld/fid=164

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 your doctor. Always talk with your doctor about the meaning of your test results and before you stop, start, or change 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 the accompanying 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 health care 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.

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