

Risk of Melanoma and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *CDKN2A* gene

What This Result Means

This result means you have change (mutation) in the *CDKN2A* gene. Mutations in this gene can cause familial atypical multiple mole melanoma (FAMMM) syndrome. People with FAMMM may have many unusual moles on their skin. They may also have an increased chance of melanoma, a type of skin cancer. They also have a higher

risk for cancer of the pancreas. The risk for these cancers is shown in the table below.

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.

Type of Cancer	Risk of Cancer	
	With a <i>CDKN2A</i> Mutation	Without a <i>CDKN2A</i> Mutation
Melanoma	28 – 76%	2%
Pancreatic	12 – 58% ^a	1.5%

^a*CDKN2A* p16.

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. For example, you can protect your skin from sun exposure. You can do this by using sunscreen and wearing a hat and other protective clothing. You might also benefit from increased screening (ie, routine skin exams by your doctor). Another thing you can do is to stop smoking and

avoid all tobacco products. This may help reduce your risk for pancreatic cancer. Talk with your healthcare provider to learn more and find out what is best for you.

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.

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.

What This Result Means for Your Family (continued)

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 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

- **GenoMEL the Melanoma Genetics Consortium**
GenoMel.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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