Understanding Your Positive CDKN2A Genetic Test Result
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 THINGS TO KNOW

1. **CDKN2A mutation**: Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the CDKN2A gene.

2. **Familial atypical multiple mole melanoma syndrome**: People with CDKN2A mutations have familial atypical multiple mole melanoma (FAMMM) syndrome.

3. **Cancer risks**: You have an increased chance to develop melanoma (skin cancer) and pancreatic cancer.

4. **What you can do**: There are risk management options to detect cancer early or lower your risk to develop cancer. It is important to discuss these options with your doctor, and decide on a plan that best manages your cancer risks.

5. **Family**: Family members may also be at risk – they can be tested for the CDKN2A mutation that was identified in you.

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**CDKN2A LIFETIME CANCER RISKS (%)**

- **Melanoma**: 28-67%  
  - General Population
  - CDKN2A Mutation Carrier

- **Pancreatic Cancer**: 2%  
  - 17-25%  
  - General Population
  - CDKN2A Mutation Carrier

**CDKN2A MUTATIONS IN THE FAMILY**

There is a 50/50 random chance to pass on a genetic mutation in CDKN2A to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations.

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*The above cancer risks represent the typical range for individuals with a mutation in this gene. If available, cancer risks specific to the mutation found in you will be provided in your results report.

*One recent study suggests that the lifetime risk for pancreatic cancer may be as high as 58%.
# Understanding Your Positive CDKN2A Genetic Test Result

**INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC**

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Your testing shows that you have a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or a variant that is likely pathogenic in the CDKN2A gene. Both of these results should be considered positive.

Everyone has two copies of the CDKN2A gene, which we randomly inherit from each of our parents. Mutations in one copy of the CDKN2A gene can increase the chance for you to develop certain types of cancer in your lifetime.

People with a CDKN2A mutation have familial atypical multiple mole melanoma (FAMMM) syndrome.

You have an increased chance to develop non-cancerous moles (usually <50 moles, including some atypical moles), cutaneous (skin) melanoma, pancreatic cancer, and astrocytoma (a cancer of the brain).

Options for screening and early detection may include a total body skin examination and screening of the pancreas. Talk to your doctor about which options may be right for you.

Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than in the general population, and is often more frequently done. It is important to discuss these options with your doctor.

Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the CDKN2A mutation that you carry, and other family members (like your aunts, uncles, cousins) may also inherit it. Your relatives can be tested for this same mutation. Depending on the family history, those who DO NOT have it may not have an increased lifetime chance (above the general population) to develop cancer.

It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

### REACH OUT

- Aim at Melanoma Foundation [aimatmelanoma.org](http://aimatmelanoma.org)
- American Cancer Society [cancer.org](http://cancer.org)
- Genetic Information Nondiscrimination Act (GINA) [ginahelp.org](http://ginahelp.org)
- National Society of Genetic Counselors [nsgc.org](http://nsgc.org)
- Canadian Association of Genetic Counsellors [cagc-accg.ca](http://cagc-accg.ca)

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your CDKN2A result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.