Ambry Genetics®

Clinician Management Resource for *CDKN1B*

This overview of clinical management guidelines is based on this patient's positive test result for *CDKN1B* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network (NCCN) in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries, and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decision but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider and may change.

SCREENING CONSIDERATIONS[1]

Multiple Endocrine Neoplasia type 4 (MEN4)

MEN4 is a newly described endocrine neoplasia. Therefore, penetrance estimates and surveillance guidelines are not available. Given the clinical overlap with MEN1, consideration can be given to following MEN1-related surveillance recommendations in patients with MEN4.*

* Management guidelines for MEN1 are outlined in the *MEN1* Clinician Management Resource, available at www.ambrygen.com/providers/resources/clinical-materials

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines) for Neuroendocrine and Adrenal Tumors v1.2023. National Comprehensive Cancer Network, Inc. 2023. All rights reserved. Accessed December 6, 2023. To view the most recent and complete version of the guideline, go to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.

Understanding Your Positive *CDKN1B* Genetic Test Result

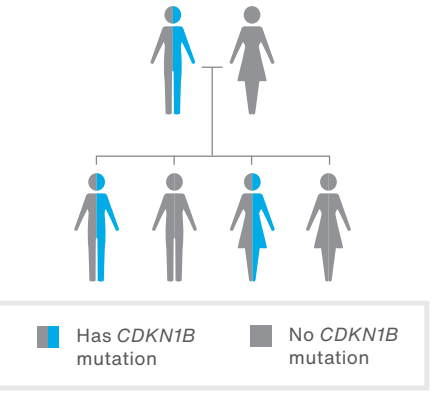
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things To Know

1	*CDKN1B* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *CDKN1B* gene.
2	Multiple endocrine neoplasia type 4	People with *CDKN1B* mutations have multiple endocrine neoplasia type 4 (MEN4).
3	Non-cancerous tumor and cancer risks	You have an increased risk for tumors. Cancerous and non-cancerous tumor types can occur in the following areas: • Parathyroid (four glands in your neck, behind your thyroid) • Pituitary (gland in the brain) • Neuroendocrine system (tumors in cells that produce hormones)
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may also be at risk – they can be tested for the *CDKN1B* mutation that was identified in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

CDKN1B Mutations in the Family

There is a 50/50 random chance to pass on a *CDKN1B* mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth.



RESOURCES

- American Brain Tumor Association abta.org
- American Cancer Society cancer.org
- International Neuroendocrine Cancer Alliance incalliance.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *CDKN1B* 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.

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