

Client Management Resource for individuals with **one** likely pathogenic or pathogenic variant in *MBD4*

This overview of clinical management guidelines is based on this patient's positive test result for one pathogenic or likely pathogenic *MBD4* variant. 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/SURGICAL CONSIDERATIONS ¹	AGE TO START	FREQUENCY
Uveal Melanoma		
Ophthalmology exam	At diagnosis	Annually

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines[®]) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, Esophageal, and Gastric. v1.2026. © National Comprehensive Cancer Network, Inc. 2026. All rights reserved. Accessed June 16, 2026. To view the most recent and complete version of the guideline, go online to [NCCN.org](https://www.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 *MBD4* Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH ONE PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know

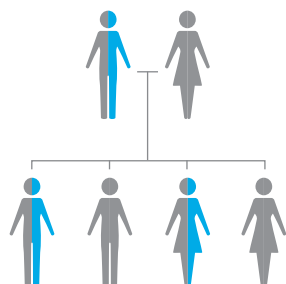
1	Result	Your testing shows that you have one pathogenic or likely pathogenic variant in the <i>MBD4</i> gene.
2	Carrier	People with one pathogenic or likely pathogenic <i>MBD4</i> variant are carriers of <i>MBD4</i> -associated neoplasia syndrome (MANS). People with two pathogenic or likely pathogenic <i>MBD4</i> variants have MANS*. Your result shows you do <u>not</u> have MANS, but your family members may be at risk for it.
3	Cancer risks	There is currently insufficient evidence to suggest an increased cancer risk for carriers (people with only one pathogenic or likely pathogenic <i>MBD4</i> variant) over that of the general population. However, ophthalmological screening for uveal melanoma may be recommended.
4	What you can do	Risk management decisions are very personal. It is important to discuss options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may be at risk- they can be tested for the pathogenic or likely pathogenic <i>MBD4</i> variant that was identified in you, as well as other <i>MBD4</i> variants. It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

* MANS is a disorder characterized by gastrointestinal polyps, colorectal cancer, and acute myelogenous leukemia or myelodysplastic syndrome.

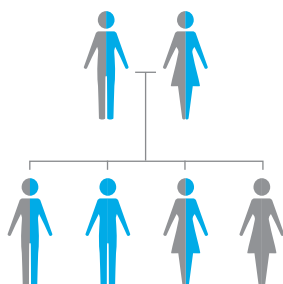
MBD4 in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *MBD4* variant to each of your children. If your partner also happens to carry one pathogenic or likely pathogenic *MBD4* variant, there is a 25% chance that you will both pass on the *MBD4* variant to your child.

One carrier parent, one non-carrier parent



Two carrier parents



- Has two pathogenic or likely pathogenic *MBD4* variants (*MBD4*-associated neoplasia syndrome)
- Has one pathogenic or likely pathogenic *MBD4* variant (carrier)
- No pathogenic or likely pathogenic *MBD4* variants

RESOURCES

- 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 *MBD4* 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.