Clinician Management Resource for *EPCAM* (Lynch syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic variant in the *EPCAM* gene. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1] 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 decisions 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 over time.

SCREENING/SURGICAL CONSIDERATIONS[1]

For individuals with *EPCAM 3'* pathogenic deletions with *MSH2* involvement: Follow the management guidelines outlined in the *MSH2* Clinican Management Resource available at https://www.ambrygen.com/providers/resources/clinical-materials.

For individuals with an *EPCAM 3'* pathogenic deletion without *MSH2* involvement: There are minimal gene-specific data on appropriate cancer surveillance and risk-reducing strategies. NCCN recommends counseling and surveillance based on age, shared decision-making, family history, and extrapolation from other forms of Lynch syndrome with comparable magnitudes of risk.

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. 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.

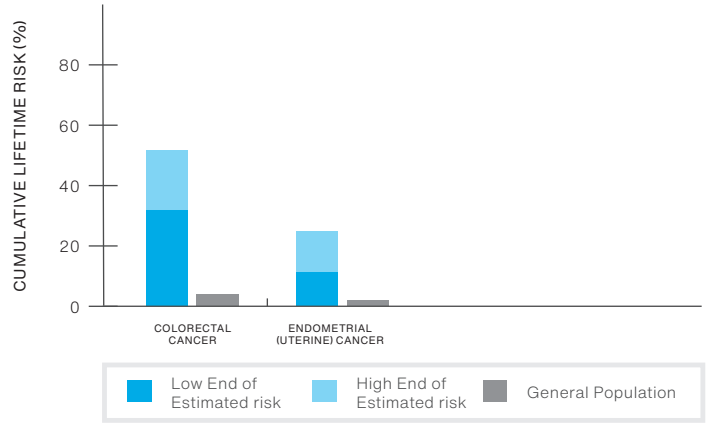
Understanding Your Positive *EPCAM* Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

6 Things To Know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant the *EPCAM* gene.
2	Lynch syndrome	People with deletions of a portion of the *EPCAM* gene have Lynch syndrome have Lynch syndrome, previously known as hereditary non-polyposis colorectal cancer (HNPCC).
3	Cancer risks	You have an increased chance to develop colorectal cancer and endometrial cancer.
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	Other Medical Concerns	Individuals with pathogenic or likely pathogenic *EPCAM* variants may have an increased risk to have a child with constitutional mismatch repair deficiency (CMMRD), but only if their partner also carries a pathogenic or likely pathogenic variant in the *EPCAM* gene. CMMRD is a multisystem disorder characterized by specific physical features and an increased risk for hematologic malignancies, brain tumors, and early-onset Lynch syndrome-associated cancers.
6	Family	Family members may also be at risk – they can be tested for the pathogenic or likely pathogenic *EPCAM* variant that was identified in you. It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

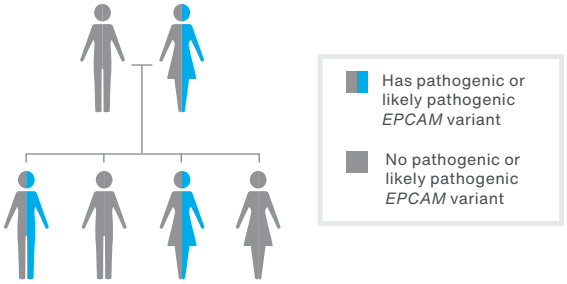
EPCAM Lifetime Cancer Risks*



* Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history. Risk estimates vary depending on MSH2 gene involvement.

EPCAM in the Family

There is a 50/50 random chance to pass on the pathogenic or likely pathogenic *EPCAM* variant to each of your children.



RESOURCES

- AliveAndKickn (Patient Advocacy Group) aliveandkickn.org
- Lynch Syndrome International lynchcancers.com
- National Society of Genetic Counselors nsgc.org
- Canadian Association 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 *EPCAM* 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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