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Clinician Management Resource for *CTNNA1*

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic variant in the *CTNNA1* gene. 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]	AGE TO START	FREQUENCY
Gastric Cancer*		
The following recommendations only apply to individuals with a truncating/loss-of-function P/LP variant in *CTNNA1*.		
Gastrectomy is recommended for patients meeting any of the following criteria: ▪ Established stage pT1b or higher signet ring cell carcinoma (SRCC). ▪ Persistent signs and symptoms that may be associated with more advanced-stage SRCC that are unexplained by other medical conditions. ▪ Endoscopic findings that may suggest presence of more advanced SRCC. Patients without any of the above criteria should have the opportunity to engage in shared decision-making, taking into account the pros and cons of risk-reducing gastrectomy versus endoscopic surveillance.	Individualized	N/A
For patients electing endoscopic surveillance, the following strategies are recommended: ▪ Upper endoscopy surveillance should be performed at centers with expertise in *CTNNA1* gastric cancer. ▪ History of *CTNNA1* should be clearly indicated on pathology requisition and multi disciplinary discussion of any abnormal findings is encouraged. ▪ Endoscopic exams should be high quality.**	Individualized	For patients who do not meet criteria for recommended gastrectomy after the surveillance exam: repeat in 6-12 months For patients who meet criteria for gastrectomy after the surveillance exam but decline gastrectomy: repeat in 6 months

* Increased gastric cancer risk has only been associated with truncating/loss-of-function *CTNNA1* variants, whereas pathogenic missense variants are associated with macular dystrophy and have not been shown to confer increased gastric cancer risk. Given the still limited understanding and rarity of this syndrome, it is recommended for patients with pathogenic/likely pathogenic *CTNNA1* variants to be referred to institutions with expertise in managing risks for cancer associated with *CTNNA1*.

** Refer to HGAST-B, page 4 of 5 of the NCCN Guidelines for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, Esophageal, and Gastric for a definition of high-quality endoscopic surveillance exams.

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 *CTNNA1* Genetic Test Result

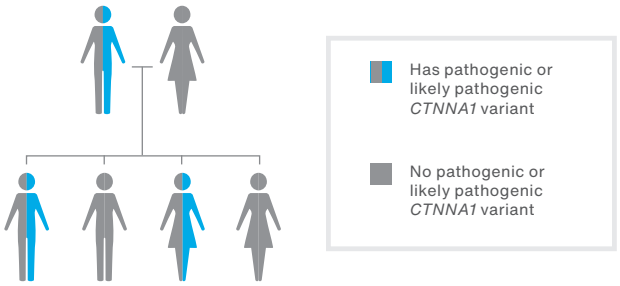
INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

4 Things To Know

1	**Result**	Your testing shows that you have a pathogenic or likely pathogenic variant in the *CTNNA1* gene.
2	**Cancer risks**	People with pathogenic or likely pathogenic *CTNNA1* variants have an increased chance to develop gastric cancer. Though the exact lifetime risk is not yet known, if there is no family history of gastric cancer, then the chance to develop gastric cancer is likely to be less than 5%. Your risk may be higher if you have a family history of gastric cancer.
3	**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.
4	**Family**	Family members may also be at risk – they can be tested for the pathogenic or likely pathogenic *CTNNA1* 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.

CTNNA1 in the Family

There is a 50/50 random chance to pass on the pathogenic or likely pathogenic *CTNNA1* variant to each of your children.



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 *CTNNA1* 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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MKT-ONCO-FLYR-10183-EN v3 08.04.26