Ambry Genetics®

Clinician Management Resource for *BMPR1A* (Juvenile polyposis syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a *BMPR1A* gene mutation. 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	AGE TO START	FREQUENCY^
Colorectal Cancer		
Colonoscopy with polypectomy** for pediatric patients	12-15 years old Colonoscopy should be initiated at an earlier age or repeated more frequently if signs/symptoms of GI blood loss.	Every 2-3 years if polyps are found, or shorter intervals based on polyp size, number, and pathology. If no polyps, resume at 18 years old.
Colonoscopy with polypectomy** for adult patients	18 years old	Every 1-3 years. Intervals should be based on polyp size, number, and pathology.
Stomach Cancer		
Upper endoscopy with polypectomy** for pediatric patients	12-15 years old Endoscopy should be initiated at an earlier age or repeated more frequently if signs/symptoms of GI blood loss.	Every 2-3 years if polyps are found, or shorter intervals based on polyp size, number, and pathology. If no polyps, resume at 18 years old.
Upper endoscopy with polypectomy** for adult patients	18 years old	Every 1-3 years. Intervals should be based on polyp size, number, and pathology.

* Due to the rarity of the syndrome and complexities of diagnosing and managing individuals with juvenile polyposis syndrome, referral to a specialized team is recommended.

** Gastrectomy and/or colectomy should be considered if polyp burden or polyp-related symptoms (i.e., anemia) cannot be controlled endoscopically or prevent optimal surveillance for cancer.

^ Any new signs/symptoms of GI disease should receive timely workup in both the pediatric and adult populations regardless of surveillance interval.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal. V2.2022. © National Comprehensive Cancer Network, Inc. 2022. All rights reserved. Accessed December 20, 2022. 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.

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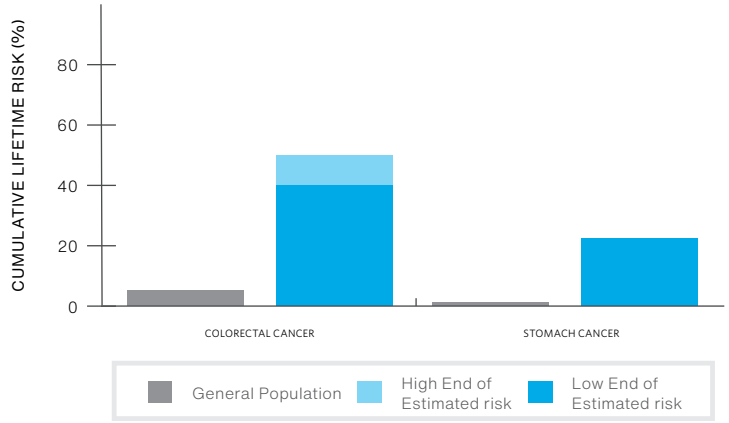
Understanding Your Positive *BMPR1A* Genetic Test Result

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

5 Things to know

1	*BMPR1A* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *BMPR1A* gene.
2	Juvenile polyposis syndrome	People with *BMPR1A* mutations have juvenile polyposis syndrome (JPS).
3	Cancer risks and other medical concerns	You have an increased chance to develop non-cancerous gastrointestinal polyps, as well as colorectal and possibly stomach 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 doctor and decide on a plan that works for you.
5	Family	Family members may also be at risk – they can be tested for the *BMPR1A* mutation that was found 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.

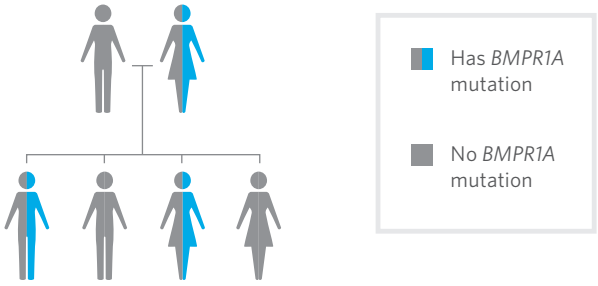
BMPR1A Mutation Lifetime Cancer Risks (%)*



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

BMPR1A Mutations in the Family

There is a 50/50 random chance to pass on a *BMPR1A* mutation to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations.



Reach Out

RESOURCES

- Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer
- Hereditary Colon Cancer Foundation hcctakesguts.org
- Genetic Information Nondiscrimination Act (GINA) ginahelp.org
- 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 *BMPR1A* 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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