BMPR1A gene

Associated Syndrome Name: Juvenile polyposis syndrome (JPS)

BMPR1A Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Colorectal	High Risk
Gastric	High Risk
Other	Elevated Risk

BMPR1A gene Overview

Juvenile polyposis syndrome (JPS) [1, 2, 3]

- Individuals with *BMPR1A* mutations have juvenile polyposis syndrome (JPS).
- Patients with JPS have a high risk for cancer as a result of hamartomatous polyps in the gastrointestinal system, particularly in the colon, rectum and stomach. The presence of these polyps is associated with a high risk for colorectal cancer, and can cause bleeding leading to anemia.
- Patients with JPS also have an elevated risk for small bowel cancer.
- Although there are high risks for cancer in patients with JPS, these risks can be greatly reduced with appropriate medical management. Guidelines from the National Comprehensive Cancer Network (NCCN) are listed below. It is recommended that patients with *BMPR1A* mutations and a diagnosis of JPS be managed by a multidisciplinary team with expertise in medical genetics and the care of patients with hereditary gastrointestinal cancer syndromes.

BMPR1A gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Colorectal	To age 42[3, 4]	20%-25%	<0.2%
	To age 80[1, 3, 4]	40%-50%	2.8%
Gastric	To age 80[1, 4]	Up to 21%	0.6%
Small Bowel	To age 80[1, 3, 4]	Rare, but elevated risk	0.2%

BMPR1A Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Colorectal	Colonoscopy[1, 5, 6]	12 to 15 years, or earlier if symptoms are present	Every 1 to 3 years, depending on age and findings
	Monitor for rectal bleeding and/or anemia.[2, 5, 6]	15 years, or earlier if symptoms are present	Annually
Gastric	Upper endoscopy[1, 6, 7]	12 to 15 years, or earlier if symptoms are present	Every 1 to 3 years, depending on age and findings

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Small Bowel	Capsule endoscopy[5, 6]	15 years	Every 1 to 2 years

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the *BMPR1A* gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

Since *BMPR1A* mutations carry a risk for complications in children and some screenings are recommended to begin by age 15 or younger, consideration should be given to the possibility of mutation testing in childhood.

References

1. Gupta S, et al. NCCN Clinical Practice Guidelines in Oncology® Genetic/Familial High-Risk Assessment: Colorectal. V 1.2023. May 30. Available at https://www.nccn.org.
2. Larsen Haidle J, et al. Juvenile Polyposis Syndrome. 2022 Feb 3. In: Adam MP, et al., editors. GeneReviews® [Internet]. PMID: 20301642.
3. Howe JR, et al. The risk of gastrointestinal carcinoma in familial juvenile polyposis. Ann Surg Oncol. 1998 5:751-6. PMID: 9869523.
4. SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2023 Mar 24]. Available from https://seer.cancer.gov/explorer/.
5. Achatz MI, et al. Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clin Cancer Res. 2017 23:e107-e114. PMID: 28674119.
6. Boland CR, et al. Diagnosis and Management of Cancer Risk in the Gastrointestinal Hamartomatous Polyposis Syndromes: Recommendations From the US Multi-Society Task Force on Colorectal Cancer. Gastroenterology. 2022 162(7):2063-2085. PMID: 35487791.
7. Ajani JA, et al. NCCN Clinical Practice Guidelines in Oncology®: Gastric Cancer. V 1.2023. Mar 10. Available at https://www.nccn.org.

Last Updated on 31-Jan-2024