MUTYH Biallelic gene

Associated Syndrome Name: *MUTYH*-associated polyposis syndrome (MAP)

MUTYH Biallelic Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Colorectal	High Risk
Other	High Risk

MUTYH Biallelic gene Overview

MUTYH-associated polyposis syndrome (MAP) [1]

- Individuals with mutations in both copies of the *MUTYH* gene (biallelic mutations) have *MUTYH*-associated polyposis syndrome (MAP).
- Most patients with MAP have between 10 and 100's of colorectal polyps by a mean age of 50. These polyps are primarily adenomas, leading to a high risk for colorectal cancer. Colorectal cancer is sometimes diagnosed in individuals with no past history of polyps.
- Patients with MAP also have an increased risk for cancer of the small bowel. Although the absolute risk is estimated to be only 5%, between 17% and 25% of patients with MAP will have duodenal polyps and upper endoscopy is recommended. Eleven percent of patients with MAP will have gastric polyps, but there does not appear to be any increased risk for gastric cancer.
- Some studies have described a possible increased risk for a wide range of cancers and non-cancer findings in patients with MAP. However, these studies are not conclusive and the estimated risks are not large. For these reasons, there are no medical management guidelines to address these risks.
- Although the risk for colorectal cancer in patients with MAP is very high, it is possible to reduce this risk with appropriate medical management. Guidelines for the management of patients with MAP have been developed by the National Comprehensive Cancer Network (NCCN). These are listed below. It is recommended that patients with *MUTYH* mutations and a diagnosis of MAP be managed by a multidisciplinary team with expertise in medical genetics and the care of patients with hereditary gastrointestinal cancer syndromes.

MUTYH Biallelic gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Colorectal	To age 80[1, 2]	43%-100%	2.8%
Small Bowel/Periampullary	To age 80[2, 3]	5%	0.2%

MUTYH Biallelic 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[3]	25 to 30 years, or earlier if indicated by family history	Every 1 to 2 years

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
	Colorectal surgical evaluation and counseling.[3]	Based on cancer diagnosis and/or polyp number, size and histology	NA
Small Bowel/Periampullary	Upper endoscopy, with consideration of capsule endoscopy to visualize the entire small bowel[3]	30 to 35 years	Every 3 to 5 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 MUTYH Biallelic 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 this patient has mutations in both copies of the *MUTYH* gene, it is almost certain each of their parents and all of their children carry at least one of these *MUTYH* mutations. Siblings are at very high risk for carrying either one or two *MUTYH* mutations. Since even a single *MUTYH* mutation can lead to an increased risk for cancer, it is especially important that this information be shared within the family and relatives talk with a healthcare provider about testing. The cancer risk tables that follow provide cancer risks for individuals with mutations in either one (monoallelic) or both (biallelic) copies of the *MUTYH* gene.

References

1. Nielsen M, et al. *MUTYH* Polyposis. 2021 May 27. In: Pagon RA, et al., editors. GeneReviews® [Internet]. PMID: 23035301.
2. SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2025 Apr 1]. Available from https://seer.cancer.gov/explorer/.
3. Gupta S, et al. NCCN Clinical Practice Guidelines in Oncology® Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. V 4.2024. Apr 2. Available at https://www.nccn.org.

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