MUTYH Monoallelic gene

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

MUTYH Monoallelic gene Overview

Carrier Status for *MUTYH*-associated polyposis syndrome (MAP) [1, 2, 3, 4, 5]

- Some studies have shown that individuals with a single *MUTYH* mutation (monoallelic *MUTYH*) may have a small increased risk for colorectal cancer compared to individuals in the general population. However, other studies have found no significantly increased risk for colorectal cancer. Given the conflicting data, there are currently no specialized colorectal cancer screening recommendations based on having a single *MUTYH* mutation (monoallelic *MUTYH*).
- Individuals with mutations in both of their copies of the *MUTYH* gene (biallelic mutations) have a condition known as *MUTYH*-associated polyposis (MAP), which is associated with a high risk for cancer. This patient does not have a diagnosis of MAP, but may have relatives who are at risk for this condition. Please see the Information for Family Members section below for details.

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 Monoallelic 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.

This patient carries a single *MUTYH* mutation (monoallelic). This patient's relatives are at risk for carrying a single *MUTYH* mutation, or mutations in both copies of *MUTYH* (biallelic). Relatives who have inherited mutations in both copies of *MUTYH* are at risk for *MUTYH*-associated polyposis syndrome (MAP), with a 43%-100% risk of colorectal cancer risk to age 80 and a 5% risk for small bowel cancer. Genetic testing may be appropriate for close family members to determine whether they are at an increased risk for colorectal and other cancers.

References

1. Jones N, et al. Increased colorectal cancer incidence in obligate carriers of heterozygous mutations in *MUTYH*. Gastroenterology. 2009 137:489-94 PubMed PMID: 19394335.

2. Win AK, et al. Risk of colorectal cancer for carriers of mutations in *MUTYH*, with and without a family history of cancer. Gastroenterology. 2014 146:1208-11. PMID: 24444654.

3. Nielsen M, et al. *MUTYH* Polyposis. 2021 May 27. In: Pagon RA, et al., editors. GeneReviews® [Internet]. PMID: 23035301.

4. Jenkins MA, et al. Risk of colorectal cancer in monoallelic and biallelic carriers of *MYH* mutations: a population-based case-family study. Cancer Epidemiol Biomarkers Prev. 2006 15:312-4. PMID: 16492921.

5. Thompson AB, et al. Monoallelic *MUTYH* pathogenic variants ascertained via multi-gene hereditary cancer panels are not associated with colorectal, endometrial, or breast cancer. Fam Cancer. 2022 Oct;21(4):415-422. PMID: 34981295.

Last Updated on 15-Sep-2026