

Individuals with MUTYH-Associated Polyposis syndrome (MAP) have a high lifetime risk for colon polyps and [colon cancer](#). There are usually tens to hundreds of polyps found in the large intestine, but some people may develop colon cancer without polyps.

Pathogenic (or harmful) variants in the MUTYH gene cause MAP. MAP is inherited in an [autosomal recessive](#) pattern, meaning that each of someone's parents carry a pathogenic variant in the MUTYH gene, and both parents pass that pathogenic variant down to a child. Two people who carry pathogenic variants in the MUTYH gene have a 25% chance for each of their children to have MAP. Notably, women and men both have the MUTYH gene and have the same chances to inherit and pass down variants in this gene. Therefore, both sides of the family are important when assessing inherited risk.

Cancer Type	General Population Risk	Risk with MAP
Colon	5.5%	80%
Duodenal (small intestine)	<1%	4%
Stomach	<1%	1%

Individuals with MAP are also at increased risk of:

- Polyps in the stomach and small intestine
- Spots, like freckles, on the inside of your eye called CHRPE
- Cysts in your jaw bone, liver, or kidney
- Fatty tumors, called subcutaneous lipomas
- Other tumors of the skin that can start in the glands or different skin layers, called sebaceous gland adenomas or epitheliomas

GENETIC TESTING FOR MUTYH

Genetic testing is available for this condition. This can be done on a blood or saliva sample. The results take about 3 weeks to return. There are different ways to complete this testing. This can include:

- [Single site analysis](#): Testing specific to a known pathogenic variant in the family
- [Full gene sequencing](#) and [rearrangement analysis](#): Comprehensive testing to search for all currently detectable variants in the gene
- [Gene panels](#): Newer, more broadly based gene tests that would include MUTYH gene, but other genes known or suspected to be associated with colon polyps and increased cancer risks

Those who undergo testing for the MUTYH gene due to a personal history of many colon polyps should also be offered testing for another genetic predisposition to colon polyps called [Familial Adenomatous Polyposis \(FAP\)](#). These two conditions are caused by pathogenic variants in two separate genes but it is often difficult to distinguish between the two conditions without genetic testing.

WHO SHOULD BE OFFERED TESTING FOR MUTYH?

The [National Comprehensive Cancer Network \(NCCN\)](#) is a group of medical professionals that regularly meet to look over any updates in research studies and determine recommendations for who should be considered at a higher risk for one of these gene variants, and thus should be offered genetic testing:

- Personal history of at least 20 adenomatous colon polyps. Testing can be considered if 10-20 polyps found.
- Family history of a known MUTYH pathogenic variant, or a family member has a positive test result.
- Those with serrated polyposis syndrome and have at least five adenomatous polyps. There is no clear genetic cause for serrated polyposis syndrome, but testing for MUTYH can be done, especially with a personal history of adenomas along with the serrated polyps.

CANCER SCREENING AND RISK MANAGEMENT FOR MUTYH ASSOCIATED POLYPOSIS (MAP)

If you are tested and found to have MUTYH/MAP, it is recommended to discuss your management plan with your healthcare team, and if available, to seek consultation through a specialized high-risk clinic. General recommendations are included here based on the updated guidelines of the [NCCN](#), but may be tailored to your specific medical and family history.

For individuals with MAP who have not had polyps; or for siblings of people with MAP who have not had testing themselves:

- Colonoscopy starting at age 25-30, continuing every 2-3 years if no symptoms. If polyps discovered, go to plan below.
- Annual physical exam.
- Baseline endoscopy at 30-35.

For individuals with MAP who have already had polyps:

- Younger than 21: colonoscopy every 1-2 years. Surgery can be considered if too many polyps.
- 21 or older, with manageable polyp burden: colonoscopy every 1-2 years. Consider surgical evaluation and colectomy as appropriate
- Too many polyps to handle endoscopically: consider surgery. Exact type should be discussed with physician.
- Annual physical exam.
- Baseline endoscopy at 30-35.

PEOPLE WHO HAVE ONE PATHOGENIC VARIANT IN MUTYH (MAP CARRIERS)

Individuals who carry one pathogenic variant in the MUTYH gene are considered carriers for MAP. One to two percent of the general population are thought to be carriers for MAP. Some studies have shown that carriers for MAP may have a slightly increased risk for [colon cancer](#), but specific numbers are not currently available. The [National Comprehensive Cancer Network](#) recommends that if someone is a carrier for MAP and they have a first-degree relative (parent, sibling, child) with [colorectal cancer](#), they should start colonoscopy screening at age 40 (or 10 years prior to the age of the first-degree relative's age at diagnosis, whichever is earlier). If the colonoscopy is normal, it should be repeated every 5 years.

There are currently no specific data available to determine whether specialized screening is needed for individuals who are carriers for MAP who do not have a first-degree relative with [colorectal cancer](#), and the plan for screening should be discussed with a primary care provider. In the general population, [colon cancer](#) screening is recommended to begin at age 50 (with no genetic risk factors or family history of [colorectal cancer](#)).

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.