



Trait Profile

MUTYH-Associated Polyposis

Other Names: MYH-Associated Polyposis

MUTYH-associated polyposis is a cancer syndrome characterized by the development of a large number of polyps. It is caused by mutations in the MUTYH gene that makes the MYH glycosylase protein.

Characteristics of MUTYH-Associated Polyposis

MUTYH-Associated Polyposis (MAP) is a genetic condition characterized by the development of many (tens to hundreds) colon polyps. The colon polyps are most often of a specific type, called adenomas. Adenomas are growths that if not removed, have a high chance of becoming cancerous over time. Without surgery, the chance of developing colorectal cancer for a person with MAP can be as high as 100% over their lifetime. On average, colon adenomas are detected by age 50. Another type of polyp (hyperplastic) is common in MAP, but these have a very low cancer risk. Rarely, an individual with MAP may develop colorectal cancer when they have had no or very few polyps.

Individuals with MAP also have an increased chance for polyps to develop in the duodenum (i.e., the beginning of the small intestine). Therefore, the risk for duodenal cancer is also increased (approximately 4% lifetime chance). Individuals with MAP also seem to be at a higher risk for other cancers, including thyroid, ovary, bladder, skin, breast, and uterine. However, the risk for these cancers is less than the risk for colorectal and duodenal cancers.

Diagnosis/Testing

Individuals with MAP have changes or mutations in a gene called MUTYH. The MUTYH gene makes the MYH glycosylase enzyme. This enzyme repairs mistakes that occur when DNA is copied. Mutations in the MUTYH do not allow for these errors to be corrected and as a result, cells may grow uncontrollably. This uncontrolled growth is what can cause the polyps and tumors in affected individuals. An individual must have a mutation in each of their two MUTYH genes to have a diagnosis of MAP.

Management/Surveillance

With appropriate management and surveillance, cancers can be prevented or detected at earlier stages, when there is a greater chance for successful treatment. Individuals with MAP are recommended to have a colonoscopy (i.e., a procedure to examine the inside of the colon and rectum) every 1-2 years starting at the age of 25. If too many polyps occur in the colon and/or they are too difficult to remove, surgical removal of all or part of the colon may be recommended. Upper GI endoscopy (i.e., a procedure to examine the esophagus, stomach, and duodenum) is recommended every 3-5 years starting at the age of 30. Regular physical examinations are recommended, and some healthcare providers also recommend individuals with MAP have an ultrasound of their thyroid. Additional screening recommendations may be made based on the individuals family history. Also, since MAP is a genetic condition that has only been understood within the past ten years, additional screening recommendations may be created after more research is completed.

Mode of inheritance

MAP is inherited in an autosomal recessive pattern. This means that an individual has to inherit to MUTYH

mutations (i.e., one from each parent) to be affected with MAP. If both parents have one MUTYH mutation, together they have a 1 in 4 (25%) chance with each pregnancy of having a child with MAP.

Risk to family members

Parents of a child with MAP have one MUTYH mutation. If a sibling of a child with MAP is unaffected, he/she has a 2 in 3 (66%) chance of having one MUTYH mutation.

Special considerations

Research has shown that individuals with one MUTYH mutation may also have an increased risk for colorectal cancer, although not necessarily at younger ages than the general population. It is important for those with one MUTYH mutation to receive regular colon screening.

Resources

Genetics Home Reference: MUTYH

<http://ghr.nlm.nih.gov/gene/MUTYH>

Cancer.Net: MYH-Associated Polyposis

<http://www.cancer.net/cancer-types/myh-associated-polyposis>

NCHPEG: MYH-associated polyposis fact sheet

[http://www.nchpeg.org/documents/crc/11-0456%20Fact%20sheets%20\(MAP\).pdf](http://www.nchpeg.org/documents/crc/11-0456%20Fact%20sheets%20(MAP).pdf)

References

Brand R, Nielsen M, Lynch H, Infante E. (Updated 4 October 2012). MUTYH-Associated Polyposis. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at <http://www.ncbi.nlm.nih.gov/books/NBK107219/>. Accessed [06/30/2013].

[National Comprehensive Cancer Network](#). NCCN Clinical Practice Guidelines in Oncology: Colorectal Cancer Screening. Version 1.2013. (Available online with registration).

Created: 06/2013

Created by: Maureen Flynn, MS, LCGC, MPH

Updated: mm/yyyy

Edited by: Seema Jamal, MSc, LCGC