UT Southwestern Harold C. Simmons Comprehensive Cancer Center | NCI Designated Comprehensive Cancer Center

MUTYH Gene Mutations

What You Should Know About _MUTYH_-Associated Polyposis (MAP) Syndrome

Individuals who inherit two *MUTYH* mutations, one from each parent (i.e., biallelic mutations), have a condition called MAP syndrome, which is a form of inherited colorectal cancer. Individuals with MAP syndrome develop numerous polyps (adenomas) in the colon. Individuals with MAP syndrome usually have between 15 and 100 polyps, but some individuals with MAP syndrome have less than 15 polyps or greater than 100 polyps. In addition to colon polyps, individuals with MAP syndrome may develop tumors in the upper gastrointestinal system, CHRPE (multiple areas of pigmentation in the retina of the eye), osteomas (benign bone tumors) of the jaw, dental abnormalities, and benign tumors of the hair follicle.

The Risk for Cancer Associated with MAP

If left untreated, colon polyps will develop into cancer. Individuals with MAP syndrome have up to an 80% risk to develop colon cancer in their lifetime and a 5% risk to develop cancer in the duodenum (small intestine).

Managing the Risk

Recommendations to manage these risks include:

- Colonoscopy every 2-3 years, beginning at age 25-30
- Upper endoscopy every 3-5 years, beginning at age 30-35
- Once severe polyposis or cancer is detected, it is recommended that the colon be removed. After colon surgery, annual surveillance of the rectum is recommended.
- Non-steroidal anti-inflammatory drugs (NSAIDS) have been shown to reduce the number and progression of adenomas.

The Risks to Family Members

MAP syndrome is caused by mutations in the *MUTYH* gene. MAP syndrome is inherited in an autosomal recessive fashion, meaning that a person must inherit a mutation in the *MUTYH* gene from both of their parents to have MAP syndrome. Brothers and sisters of a person with MAP have a 25% (1 in 4) risk to inherit MAP syndrome, a 50% (1 in 2) risk to have one *MUTYH* mutation, and a 25% (1 in 4) chance that they will not have a *MUTYH* mutation. Approximately 1-2% of the population has a *MUTYH* mutation, so it is important to know that individuals with one *MUTYH* mutation have an increased risk for having a child with MAP syndrome, and their spouse should be offered testing to see if they also have a *MUTYH* mutation.

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