Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the MUTYH Gene

This information explains how having a mutation in the *MUTYH* gene may affect you and your family.

In this resource, the word “family” means people related to you by blood. They’re not related to you through marriage or adoption. We also call these family members your blood relatives.

Your *MUTYH* gene normally helps prevent cancers. A mutation in this gene causes it to stop working like it should. This increases your risk for certain types of cancers. Your risk for cancer depends on whether you have 1 or 2 *MUTYH* mutations.

It is important to understand that having a mutation in this gene does not mean you will definitely develop cancer. It means that you have an increased risk of developing certain types of cancers. The type of cancer can vary among people who have mutations in this gene, even within the same family.

What is my cancer risk if I have 1 *MUTYH* mutation?

Most people with a *MUTYH* mutation inherit only 1 mutation from a parent.

If you have 1 mutation in the *MUTYH* gene, you may have a slightly increased risk for colorectal (colon and rectal) cancer. This is more likely if you have family history of colorectal cancer.

What is my cancer risk if I have 2 *MUTYH* mutations?

A person may inherit 2 mutations in the *MUTYH* gene, one from each parent, but this is less common.

If you have 2 *MUTYH* mutations, this means you have a condition called MUTYH-Associated Polyposis (MAP). This increases your risk for certain types of cancers, including:

- Colorectal cancer and polyps (growths of tissue) in your colon or rectum.
- Small intestine (small bowel) cancer and polyps in your intestine.

MAP may also increase your risk for other cancers, but this is less common. Examples of less common cancers linked to MAP are:

- Thyroid cancer
- Ovarian cancer
- Bladder cancer

As we learn more about these mutations, we may learn they increase the risk for other types of cancers. Your genetic counselor will give you more information about your cancer risk if you have a mutation.

To learn more, read
Hereditary Colon Cancer and Polyposis:
www.mskcc.org/genetics/colon-cancer-polyposis

What can I do about my cancer risk if I have 1 or 2 *MUTYH* mutations?

Your genetic counselor will review your results. They will talk with you about what cancer your mutation is linked to. As we research these mutations, we may learn they raise the risk for other types of cancers.

Your genetic counselor will also review your personal and family history of

cancer and give you cancer screening recommendations. They may recommend you start having cancer screenings at a younger age or have them more often than most people. They may also suggest you get specialized screenings to help find cancer as early as possible.

Their recommendations will depend on whether you have 1 or 2 *MUTYH* mutations. Some examples of these cancer screenings include:

- Colonoscopies starting at an earlier age and more often than most people.
- An upper endoscopy (a procedure that les your doctor see inside your stomach and small intestine).
- Having a urinalysis (a test to look for blood in your urine).

If you have 2 *MUTYH* mutations and are having surgery for colon cancer or polyps, your genetic counselor may recommend you have extra colon tissue removed to prevent colon cancer.

Your genetic counselor will also talk with you about whether there are any other screening or prevention options that may be right for you.

What does a *MUTYH* mutation mean for my blood relatives?

If you have a mutation in 1 of your *MUTYH* genes, your biological parents, siblings, and children each have a 50% chance of having the same mutation. Your distant family members may also be at risk for having the same mutation.

If you have 2 mutations in your MUTYH genes, your biological parents and children may have 1 or 2 MUTYH mutations. Your siblings may have 1 or 2 MUTYH mutations, or they may not have inherited any MUTYH mutations.

Males and females have an equal chance of passing down this mutation in their family. Your genetic counselor will review your family history and talk with you about whether they recommend genetic testing for your blood relatives.

What does this mean for family planning?

If you have 1 or 2 *MUTYH* mutations and plan to have children, there are options to prevent your children from inheriting this mutation. You may want to discuss these options especially if both you and your partner have *MUTYH* mutations.

If you both have *MUTYH* mutations, which is rare, there's a chance your child could be born with 2 *MUTYH* mutations and have MAP. For more information about genetic testing and family planning, talk with your genetic counselor.

What happens if I do not have a mutation?

If you do not have a mutation or if we find a variant of uncertain significance (VUS), your genetic counselor will review your personal and family history of cancer. They'll talk with you about the general cancer screening guidelines you should follow.

A VUS is a change in a gene, but we don't yet know if it is linked with a higher risk for cancer. Most VUS are eventually found to be normal changes that do not affect your health.

Contact information

If you have any questions or concerns, talk with a genetic counselor in the Clinical Genetics Service. You can reach them Monday through Friday from 9 a.m. to 5 p.m. at 646-888-4050.

For more resources, visit www.mskcc.org/pe to search our virtual library.

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