Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the PMS2 Gene

This information explains how having a mutation in the *PMS2* 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 *PMS2* gene normally helps prevent cancers. A mutation in this gene causes it to stop working like it should.

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 a *PMS2* mutation?

If you have a mutation in the PMS2 gene, this means you have a condition called Lynch syndrome.

Lynch syndrome increases your risk for certain types of cancers, including:

- Colorectal (colon and rectal) cancer
- Uterine (endometrial) cancer

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

- Ovarian cancer
- Stomach cancer
- Small intestine (small bowel) cancer
- Urinary tract cancer
- Pancreatic cancer
- Hepatobiliary tract cancer (cancer in the cells of the liver, bile ducts, and gallbladder)
- Brain cancer
- Sebaceous carcinoma (cancer in the glands in your skin that make oil)

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 *Lynch Syndrome & Genetic Testing* (www.msk.org/genetics/lynch-syndrome).

Some data suggests that people who have an *MSH6* gene mutation may have lower overall cancer risks when compared to people who have mutations in one of the other Lynch syndrome genes (such as *MLH1*, *MSH2*, and *EPCAM*). It is important to stay in contact with your genetic counselor and care team as information about these risks may change over time.

What can I do about my cancer risk if I have a *PMS2* mutation?

Your genetic counselor will review your results. They will talk with you about what cancer your mutation is linked to.

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.

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 lets your doctor see inside your stomach and small intestine).
- A urinalysis (a test to look for blood in your urine).

Your genetic counselor may also talk with you about having surgery to try to keep cancer from developing, such as:

- Surgery to remove your uterus to prevent endometrial cancer.
- Surgery to remove your ovaries to prevent ovarian cancer.

If you're having surgery for colon cancer or polyps, your genetic counselor may recommend you have extra colon tissue removed to prevent colon cancer.

If you decide to have surgery, talk with your genetic counselor about the right time to have it. Surgery to remove the uterus and ovaries affects fertility (your ability to have biological children). If you plan to have biological children, your genetic counselor can talk with you about your options.

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 *PMS2* mutation mean for my blood relatives?

If you have a mutation, 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.

Males and females have an equal chance of passing down a mutation in their family. You only need to inherit a mutation from one parent to have an increased risk for cancer.

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 a *PMS2* mutation and plan to have children, there are options to prevent your children from inheriting the mutation. You may want to consider discussing these options especially if both you and your partner have a *PMS2* mutation.

If you both have a mutation in the *PMS2* gene, there's a chance your child could be born with a rare but serious condition called Constitutional Mismatch Repair Deficiency (CMMR-D) syndrome. CMMR-D syndrome causes a higher risk for childhood cancers. If you already have children, it's unlikely they have CMMR-D since this is usually diagnosed early in life. 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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