Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About the Multi-Gene Panel Test for Lynch Syndrome

This information explains the multi-gene panel test for Lynch syndrome. It also explains how your results 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.

This multi-gene panel test is done to check your DNA for gene mutations linked to Lynch syndrome. Lynch syndrome is associated with colorectal, uterine (endometrial), ovarian, and other types of cancer.

Normally, the genes linked to Lynch syndrome help prevent cancer. A mutation in these genes causes them to stop working like they should. This raises your risk for some types of cancers.

It is important to understand that having a Lynch syndrome gene mutation 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 and age at diagnosis can vary among people who have Lynch syndrome, even within the same family.

To learn more, read *Lynch Syndrome & Genetic Testing* (www.msk.org/genetics/lynch-syndrome).

What is a multi-gene panel test?

We can test for mutations 1 gene at a time or many genes at the same time. A multi-gene panel test is when we test many genes at once using the same saliva or blood sample. By testing many genes at once, it may be possible to find a hereditary cause of cancer more quickly.

What genes will be tested on my multi-gene panel?

We'll test the 5 genes linked to Lynch syndrome: *MLH1*, *MSH2*, *MSH6*, *PMS2* and *EPCAM*.

What happens if I have a mutation?

If you have a mutation in 1 of the 5 Lynch syndrome genes, it 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).

What can I do about my cancer risk if I have a Lynch syndrome 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 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.

What does a gene mutation mean for my blood relatives?

The mutations we test for are passed down in a dominant pattern. This means you only need to inherit a mutation from 1 parent to have a higher risk for cancer. Males and females have an equal chance of passing down a mutation in their family.

If you have a mutation in 1 of these genes, it's possible that other blood relatives have it too. There's a 50% chance your biological parents, siblings, and children also have the same mutation.

Your distant family members may also be at risk for having the same mutation. It may be helpful to share this information with them. This includes aunts, uncles, nieces, nephews, and cousins.

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 Lynch syndrome 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 mutation in the same Lynch syndrome gene.

If you both have a mutation in the same Lynch syndrome 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.

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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