Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the POLD1 Gene

This information explains how having a mutation in the *POLD1* 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 *POLD1* gene normally helps prevent cancers. A mutation in this gene makes it 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 *POLD1* mutation?

If you have a mutation in the *POLD1* gene, this means you have a condition called Polymerase Proofreading-Associated Polyposis (PPAP) syndrome. PPAP syndrome increases your risk of developing polyps (growths of tissue) in your colon and rectum. These polyps can lead to an increased risk for colorectal (colon and rectal) cancer. PAPP can also cause polyps in your stomach and upper GI tract.

There may also be an increased risk of uterine cancer in females with PPAP, but more research is needed to better understand this risk.

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 a *POLD1* mutation?

Your genetic counselor will review your results. They will talk with you about what cancer(s) 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.

For example, they may recommend getting colonoscopies at an earlier age and more often than most people.

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 *POLD1* 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 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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