



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 family members related to you by blood. They are not related to you through marriage or adoption.

Your *POLD1* gene normally helps prevent cancers. A mutation in this gene makes it stop working like it should. This increases your risk for certain types of cancers.

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.

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.

For more information, 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?

If you have a mutation, your genetic counselor will review your results and 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, have them more often than most people, or 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 happens if I don't have a *POLD1* mutation?

If you don't have a mutation, your genetic counselor will review your personal and family history and talk with you about the general cancer screening guidelines you should follow.

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. This means there's an equal chance they will or won't have the 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.

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 9:00 AM to 5:00 PM at 646-888-4050.

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

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