Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the PTCH1 Gene

This information explains how having a mutation in the *PTCH1* 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 *PTCH1* 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 *PTCH1* mutation?

If you have a mutation in the *PTCH1* gene, this means you have a condition called Gorlin Syndrome or Nevoid Basal Cell Carcinoma Syndrome (NBCCS).

PTCH1 mutations increase your risk for certain types of cancers, growths, or abnormalities, including:

- Basal cell carcinoma (skin cancer).
- Jaw keratocysts (typically benign cysts of the jaw).
- Skeletal abnormalities.

PTCH1 mutations may also increase your risk for other cancers, but this is less common. Some examples of less common cancers linked to *PTCH1* are:

- Medulloblastoma (MED-yoo-loh-blas-TOH-muh). This is a type of cancer that forms in the cerebellum (the lower, back part of the brain).
- Cardiac or ovarian fibromas (typically benign tumors).

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.

What can I do about my cancer risk if I have a *PTCH1* 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.

Some examples of these cancer screenings include:

- Total body skin exams.
- X-ray of your lower jaw to check for cysts.
- Ultrasound of your ovaries to check for tumors.
- Possible echocardiogram to check for cardiac tumors (growths that can form in the heart muscle, valves, chambers, or around the heart). An echocardiogram is an imaging test that uses ultrasound (sound waves) to take pictures of your heart.

Some of these screenings will happen once a year. Others may happen more or less often. Your genetic counselor will talk with you about screening recommendations and how often you should get them. They will also talk with you about whether there are any other screening or prevention options that may be right for you.

What does a *PTCH1* 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 *PTCH1* 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 with your reproductive partner and a 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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