Memorial Sloan Kettering Cancer Center

PATIENT & CAREGIVER EDUCATION

About Mutations in the TSC2 Gene

This information explains how having a mutation in the *TSC2* 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 *TSC2* 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 *TSC2* mutation?

If you have a mutation in the *TSC2* gene, this means you have a condition called Tuberous Sclerosis Complex (TSC).

TSC2 increases your risk for certain types of cancers and typically benign tumors including:

- Skin growths (such as angiofibromas of the face, fibrous forehead plaques, ungual fibromas, or shagreen patches) and hypopigmentation of the skin (called ash-leaf spots).
- Tumors of the central nervous system (called subependymal glial nodules, cortical tubers, or giant cell astrocytomas).
- Retina tumors called retinal astrocytic hamartoma. Your retina is the layer of tissue at the back of your eyeball that senses light and helps you see.
- Heart tumors called rhabdomyomas.
- Kidney tumors called renal cysts or angiomyolipoma.
- Lymphangioleiomyomatosis (LAM, lung cysts which are typically in females).

TSC2 mutations may also increase your risk for other cancers or tumors, but this is less common. Examples of less common cancers and tumors linked to *TSC2* are:

- Kidney cancer.
- Oncocytoma (typically benign kidney tumor).

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 *TSC2* 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:

- Eye exam with an ophthalmologist (eye doctor).
- Total body skin exam starting at an earlier age.

- Neurological evaluation and brain imaging.
- Screening for kidney cancer.
- Echocardiogram (ECG) to check for any heart conduction issues.
- Lung function testing and imaging to check for lung cysts.
- Dental exam to look for growths of the gum and pitting of the enamel.

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 *TSC2* 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 *TSC2* 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.

If you have questions or concerns, contact your healthcare provider. A member of your care team will answer Monday through Friday from 9 a.m. to 5 p.m. Outside those hours, you can leave a message or talk with another MSK provider. There is always a doctor or nurse on call. If you're not sure how to reach your healthcare provider, call 212-639-2000.

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

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