

About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which gives instructions to cells about how to grow and work together. We have two copies of each gene in each cell—one from our mother and one from our father. When genes work right, they help stop cancer cells from developing. If one copy of a gene has a mutation, it cannot function as it should. This increases the risk for certain cancers.

The *SDH* genes have instructions for turning food into energy and helping fix mistakes in DNA. The four genes involved are *SDHA*, *SDHB*, *SDHC*, and *SDHD*. If there is a mutation in one of these genes, it can cause cells to grow and divide too much. This can lead to tumors called *parangliomas* and *pheochromocytomas*. These tumors are often benign (non-cancerous) but can be cancer and spread in some cases. This condition is called *hereditary paraganglioma/pheochromocytoma syndrome*.

Parangliomas and Pheochromocytomas

Parangliomas (PGLs) are slow-growing tumors that develop along nerves or blood vessels. They are usually found in the head and neck, but can also grow in the chest, abdomen, and other parts of the body. Signs of PGLs include difficulty swallowing, hoarseness, pain, cough, ringing in the ears, and hearing loss. Some PGLs, especially ones in the head and neck, can cause problems by putting pressure on nerves or blood vessels.

Pheochromocytomas (pheos or PCCs) are a type of paraganglioma that occurs in the adrenal glands, which sit on top of the kidneys. These tumors are usually benign, but they often make chemicals that can affect blood pressure and cause headaches, abnormal heart beat, anxiety, nausea, and weight loss.

SDHB Mutations and Tumor Risk

About 7 in 10 people with *SDHB* mutations will develop PGLs or PCCs in their lifetime. These can happen at young ages. About 3 in 10 will have a tumor by age 30. These tumors grow in the abdomen and pelvis most of the time, but they can also happen in the head and neck. PGLs don't usually spread to other organs for the general population, but they can in people with the *SDHB* mutation.

Kidney cancer, thyroid cancer, and a type of stomach tumor (called a *stromal tumor*) can also happen in people with *SDHB* gene mutations. The risk for these other tumors is low.

Recommendations

Knowing if you have an *SDHB* mutation can help you manage your medical care.

MEN AND WOMEN

If you know you have an *SDHB* mutation, it is important to find tumors early. It is also important to tell your doctor that you have this mutation before any medical procedures. Recommendations may vary according to your age.

Ages 8-18:

- MRI of the whole body every 2-3 years
- Neck MRI every 2-3 years

Adults:

- PET/CT scan as your doctor recommends

All ages:

- Physical exam every year with blood pressure check
- Blood test every year
- Blood tests before pregnancy or surgical procedures

Your doctor may recommend more screening depending on your personal or family history.

KIDS AND SIBLINGS

Children and siblings of people with an *SDHB* mutation have a 1 in 2 chance of also having the mutation and should get tested by age 8. If they have it, they have a higher risk of PGLs and PCCs and should follow the screening plan above. If they do not have the mutation, they do not have a higher risk and should follow the general cancer screening guidelines.

It is important to determine which side of the family carries the *SDHB* mutation to know which family members are at risk for tumors. A genetic counselor can help you know who in your family should be tested.

Do you have questions about your risk for cancer?
Our doctors and genetic counselors can help
find the cancer screening plan you need.

**Call Huntsman Cancer Institute's Family
Cancer Assessment Clinic to learn more:
801-587-9555.**