

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 paragangliomas 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.

Paragangliomas and Pheochromocytomas

Paragangliomas (PGLs) are slow-growing tumors that happen 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 hard time 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 happen in the adrenal glands, which are 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.

SDHC Mutations and Tumor Risk

Researchers are not sure how *SDHC* mutations affect PGL and PCC tumor risk. However, people who have the mutation are more likely to develop these tumors and should get screened more often than the general population. Most of the time these tumors grow in the head and neck area, but they can also happen in the abdomen. In addition to PGLs and PCCs, *SDHC* mutations can lead to Carney-Stratakis syndrome and gastrointestinal stromal tumors (GIST).

Recommendations

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

MEN AND WOMEN

If you know you have an *SDHC* 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 *SDHC* 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, PCCs, and GIST and should follow the screening plan above. If they do not have the mutation, they do not have a higher risk and should follow general cancer screening guidelines.

It is important to find out which side of the family carries the *SDHC* 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.**