

## 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 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.

## *SDHD* Mutations and Tumor Risk

A person whose mother or father has an *SDHD* mutation has a 1 in 2 chance of passing the mutation to each of his or her children. The *SDHD* gene is different than the other *SDH* genes. With *SDHD*, a person has a higher risk of tumors **only if** the mutation comes from their father. If the mother has the mutation, the child can still carry the mutation but does not have a higher risk of tumors.

People with an *SDHD* mutation from their father have a high risk of getting PGLs and PCCs, developing tumors at younger ages, and having more than one tumor. About 7 in 10 people develop a PGL before age 50. The tumors mainly grow in the head and neck but can also happen in the chest and abdomen. Rarely, these tumors spread or become cancer. On average, people find these tumors around age 35.

## Recommendations

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

### MEN AND WOMEN

If you know you have an *SDHD* 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 *SDHD* mutation have a 1 in 2 chance of also having it and should be tested by age 8. If they have the mutation from their father, they have a higher risk of tumors and should follow the screening plan above. If they have the mutation from their mother, they do not need to have tumor screenings, but their kids may need testing. If they did not inherit 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 *SDHD* 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.**