About Genes

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

The DICER1 gene controls cell growth. When DICER1 is not working right, cells can grow and divide without stopping. Tumors, and sometimes cancer, can form. Having a mutation in the DICER1 gene can make you more likely than average to get certain cancers and tumors that are not cancer.

DICER1 Mutations, Tumors, and Cancer

At this time, we know little about the likelihood of people with DICER1 mutations getting these types of tumors and cancer. Researchers are still studying DICER1 mutations and their effects.

Most people with a mutation in DICER1 never get tumors or cancer because of it. Tumors and cancer caused by DICER1 are very rare. If they do happen, it is usually before the age of 40. Talk with your doctor if you have any of the signs below.

Here are some tumors and cancers that can happen in people with DICER1 mutations:

**Pleuropulmonary blastoma** (PPB) is a rare type of lung cancer. Lung cysts may also form. PPB usually happens in children younger than age 5.

The signs of PPB resemble common illnesses. Talk with your doctor if you or your child have signs of a cold or flu that last longer than 2 weeks.

**Thyroid tumors.** People with a DICER1 mutation may get tumors on their thyroid. The thyroid is a gland in the neck. It makes hormones that control many important body functions. The tumors usually happen in adults. The first sign is a lump in the neck. Some thyroid tumors are cancerous, and some are not.

**Kidney cancers.** Kidney cancers, such as cystic nephroma, sarcoma, and Wilms tumors can happen in people with DICER1 mutations. The cancers usually happen in children younger than age 4. One of the first signs is a hard mass or swelling in the belly. Belly pain and blood in the urine are other signs.

**Ovary tumors.** Young adult women with DICER1 mutations can get tumors of the ovaries. Some tumors are cancerous, and some are not. These may be signs of ovarian tumors:

- Facial hair
- Deep voice
- Swollen belly
- Pain in lower belly

Ovary tumors can also happen in young girls with DICER1 mutations. Talk to your doctor if these signs of a girl's puberty appear before age 8:

- Breasts starting to grow
- Menstrual periods
- Hair under the arms and near the genitals

**Other rare growths.** Some people with DICER1 mutations can get polyps or tumors in these areas:

- Colon
- Cervix (females only)
- Lower abdomen
- Nose
- Brain
- Eyes

**Recommendations**

Usually, we recommend screening children between birth and age 12. Screening includes x-rays or scans in these areas:

- Lung
- Thyroid
- Abdomen
- Pelvis
- Eyes

For all adults, we recommend screening for thyroid. Women also need screening for gynecological cancers.

**KIDS AND SIBLINGS**

Siblings and children of people with a DICER1 mutation have a 1 in 2 chance of also having the mutation. We recommend genetic testing and counseling for young children because childhood tumors are possible.

People in the family who test negative for the DICER1 mutation are thought to have the same risk for cancer as the general population if they have no other risk factors. They should follow general cancer screening guidelines.

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Resources

If you have a personal or family history of any of these tumor types, you may be eligible for genetic testing and counseling.

If you already know you or a family member has a DICER1 or other gene mutation, our team of doctors and genetic counselors can help find the screening plan you need.

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

Your Notes

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