

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
- Swollen belly
- Deep voice
- 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
- Nose
- Cervix (females only)
- Brain
- Lower abdomen
- Eyes

Recommendations

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

- Lung
- Pelvis
- Thyroid
- Eyes
- Abdomen

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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