

## TRIP13 gene

thyroid hormone receptor interactor 13

### Normal Function

The *TRIP13* gene provides instructions for making a protein that has several roles in cell division. One important role is to help ensure proper chromosome separation when cells divide. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids, which are attached to one another during the early stages of cell division. The sets of chromosomes align within the cell, with each chromatid attached to a structure called a spindle microtubule; when all chromatids are correctly attached, the spindle microtubule pulls the two halves of the chromatid pair to opposite sides of the cell. The cell then divides in two such that each new cell contains one complete set of chromosomes.

Cells have a mechanism, called the spindle assembly checkpoint, that delays cell division until each sister chromatid is attached to a spindle microtubule. The TRIP13 protein appears to regulate this checkpoint, although the exact mechanism is unclear.

### Health Conditions Related to Genetic Changes

#### Mosaic variegated aneuploidy syndrome

At least two *TRIP13* gene mutations have been found to cause mosaic variegated aneuploidy (MVA) syndrome. This condition is typically characterized by cells with abnormal numbers of chromosomes, a situation known as aneuploidy. Individuals with MVA syndrome caused by *TRIP13* gene mutations have a high risk of developing a form of kidney cancer called Wilms tumor in childhood. They may also grow slowly and have an unusually small head size (microcephaly).

*TRIP13* gene mutations involved in MVA syndrome lead to production of an abnormally short protein that is quickly broken down. As a result, cells lack TRIP13 protein. A shortage of this protein impairs the spindle assembly checkpoint, and cell division proceeds, even if not all the chromatids are attached to spindle microtubules. Unattached chromatids are not positioned correctly for separation, and the resulting cells often have abnormal numbers of chromosomes. Some people with *TRIP13* gene mutations have chromosome abnormalities that indicate problems with chromatid separation, although they do not have abnormal numbers of chromosomes in their cells.

Research suggests that problems with the spindle assembly checkpoint underlie the development of cancer in MVA syndrome, although the mechanism is not completely understood. It is also unclear how *TRIP13* gene mutations lead to other features of MVA syndrome. Researchers speculate that the abnormal cells undergo self-destruction (apoptosis). The signs and symptoms of MVA syndrome may be due to the loss of cells from various tissues during early development.

- 16E1-BP
- 16E1BP
- HPV16 E1 protein binding protein
- human papillomavirus type 16 E1 protein-binding protein
- pachytene checkpoint protein 2 homolog isoform 1
- pachytene checkpoint protein 2 homolog isoform 2
- thyroid receptor-interacting protein 13
- TR-interacting protein 13
- TRIP-13

### Tests Listed in the Genetic Testing Registry

- ## Scientific Articles on PubMed

- ## Catalog of Genes and Diseases from OMIM

- Reprinted from MedlinePlus Genetics (<https://medlineplus.gov/genetics/>)

try/604507)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9319>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=TRIP13\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=TRIP13[gene]))

### **References**

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### **Genomic Location**

The *TRIP13* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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