

## MPLKIP gene

M-phase specific PLK1 interacting protein

### Normal Function

The *MPLKIP* gene (formerly known as *C7orf11*) provides instructions for making a protein called M-phase specific PLK1 interacting protein. Based on its interaction with a protein called Plk1, the MPLKIP protein is thought to play a role in cell growth and division. In particular, it may help regulate the cell cycle, which is the cell's way of replicating itself in an organized, step-by-step fashion. The MPLKIP protein also interacts with a protein involved in processing and repairing RNA molecules, which are chemical cousins of DNA.

### Health Conditions Related to Genetic Changes

#### Trichothiodystrophy

Variants (also called mutations) in the *MPLKIP* gene have been identified in people with trichothiodystrophy. This condition affects many parts of the body. *MPLKIP* gene variants cause some cases of the non-photosensitive form of trichothiodystrophy, which is not associated with extreme sensitivity to ultraviolet (UV) rays from sunlight. These gene variants account for less than 20 percent of all cases of non-photosensitive trichothiodystrophy.

All of the known *MPLKIP* gene variants prevent the production of any functional MPLKIP protein. It is unclear how the loss of this protein leads to the characteristic features of trichothiodystrophy, which include slow growth, intellectual disability, and brittle hair.

### Other Names for This Gene

- ABHS
- C7orf11
- chromosome 7 open reading frame 11
- ORF20
- TTD non-photosensitive 1 protein
- TTDN1

- TTDN1\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MPLKIP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=136647\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=136647[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28C7orf11%5BTIAB%5D%29+OR+%28MPLKIP%5BTIAB%5D%29%29+OR+%28TTDN1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- M-PHASE-SPECIFIC PLK1-INTERACTING PROTEIN; MPLKIP (<https://omim.org/entry/609188>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/136647>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MPLKIP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MPLKIP[gene]))

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## Genomic Location

The *MPLKIP* gene is found on chromosome 7 (<https://medlineplus.gov/genetics/chromosome/7/>).

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