

# CDC6 gene

cell division cycle 6

### **Normal Function**

The *CDC6* gene provides instructions for making a protein that is important in the copying of a cell's DNA before the cell divides (a process known as DNA replication). The protein produced from this gene, called cell division cycle 6 or CDC6, is one of a group of proteins known as the pre-replication complex. In a multi-step process, the components of this complex attach (bind) to certain regions of DNA known as origins of replication (or origins), where the process of DNA copying begins. When the pre-replication complex is attached to the origin, replication is able to begin at that location. This tightly controlled process, called replication licensing, helps ensure that DNA replication occurs only once per cell division and is required for cells to divide.

## **Health Conditions Related to Genetic Changes**

### Meier-Gorlin syndrome

At least one mutation in the *CDC6* gene causes Meier-Gorlin syndrome, a condition characterized by short stature, underdeveloped kneecaps, and small ears. This mutation, which is a rare cause of the condition, changes a single protein building block (amino acid) in the CDC6 protein, replacing the amino acid threonine at position 323 with the amino acid arginine (written as Thr323Arg). As a result, assembly of the prereplication complex is impaired, which disrupts replication licensing; however, it is not clear how a reduction in replication licensing leads to Meier-Gorlin syndrome. Researchers speculate that such a reduction delays the cell division process, which slows growth of the bones and other tissues during development. It is not known why development of the kneecaps and ears is particularly affected in Meier-Gorlin syndrome.

#### Other Names for This Gene

- cdc18-related protein
- CDC18L
- CDC6 cell division cycle 6 homolog
- CDC6-related protein
- CDC6\_HUMAN
- cell division control protein 6 homolog

- cell division cycle 6 homolog
- HsCDC18
- HsCDC6
- p62(cdc6)

### **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

Tests of CDC6 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=990[geneid])

#### Scientific Articles on PubMed

 PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CDC6%5BTIAB%5D%29 +OR+%28cell+division+cycle+6%5BTIAB%5D%29%29+AND+%28%28Genes%5B MH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D)

### Catalog of Genes and Diseases from OMIM

CELL DIVISION CYCLE 6; CDC6 (https://omim.org/entry/602627)

#### Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/990)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CDC6[gene])

#### References

- Bicknell LS, Bongers EM, Leitch A, Brown S, Schoots J, Harley ME, Aftimos S, Al-Aama JY, Bober M, Brown PA, van Bokhoven H, Dean J, Edrees AY, Feingold M, Fryer A, Hoefsloot LH, Kau N, Knoers NV, Mackenzie J, Opitz JM, Sarda P, Ross A, Temple IK, Toutain A, Wise CA, Wright M, Jackson AP. Mutations in thepre-replication complex cause Meier-Gorlin syndrome. Nat Genet. 2011 Feb27;43(4): 356-9. doi: 10.1038/ng.775. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21 358632) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/article s/PMC3068194/)
- Fernandez-Cid A, Riera A, Tognetti S, Herrera MC, Samel S, Evrin C, Winkler C, Gardenal E, Uhle S, Speck C. An ORC/Cdc6/MCM2-7 complex is formed in a multistepreaction to serve as a platform for MCM double-hexamer assembly. Mol Cell. 2013May 23;50(4):577-88. doi: 10.1016/j.molcel.2013.03.026. Epub 2013 Apr 18. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23603117)

 Niida H, Kitagawa M. Regulation of DNA replication licensing. Curr DrugTargets. 2012 Dec;13(13):1588-92. doi: 10.2174/138945012803529965. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22998185)

## **Genomic Location**

The *CDC6* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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