

## MCM6 gene

minichromosome maintenance complex component 6

### Normal Function

The *MCM6* gene provides instructions for making a protein that is part of the MCM complex, a group of proteins that functions as a helicase. Helicases attach to particular regions of DNA and temporarily unwind the two spiral strands of these molecules. When a cell prepares to divide to form two cells, helicases unwind the DNA so that it can be copied. The DNA that makes up the chromosomes is duplicated (replicated) so that each new cell will get a complete set of chromosomes. Helicases are also involved in the production of RNA, a chemical cousin of DNA.

### Health Conditions Related to Genetic Changes

#### Lactose intolerance

A specific DNA sequence within the *MCM6* gene called a regulatory element helps control the activity (expression) of a nearby gene called *LCT*. The *LCT* gene provides instructions for making an enzyme called lactase. This enzyme helps to digest lactose, a sugar found in milk and several other dairy products. Lactose intolerance in adulthood is caused by the gradually decreasing expression of the *LCT* gene after infancy, which occurs in most humans.

A few variants (also called mutations) have been identified in the regulatory element of *MCM6* that controls *LCT* gene expression. These variants change single DNA building blocks (nucleotides) in the regulatory element. Each of the variants results in sustained lactase production in the small intestine and the ability to digest lactose throughout life. People without these changes have a reduced ability to digest lactose as they get older, resulting in the signs and symptoms of lactose intolerance.

### Other Names for This Gene

- MCM6\_HUMAN
- minichromosome maintenance deficient 6

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MCM6 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4175\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4175[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28MCM6%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+1800+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- MINICHROMOSOME MAINTENANCE COMPLEX COMPONENT 6; MCM6 (<https://omim.org/entry/601806>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4175>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MCM6\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MCM6[gene]))

## References

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- Liebert A, Lopez S, Jones BL, Montalva N, Gerbault P, Lau W, Thomas MG, Bradman N, Maniatis N, Swallow DM. World-wide distributions of lactase persistence alleles and the complex effects of recombination and selection. Hum Genet. 2017 Nov; 136(11-12):1445-1453. doi: 10.1007/s00439-017-1847-y. Epub 2017 Oct 23. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/29063188>)
- Shastri VM, Subramanian V, Schmidt KH. A novel cell-cycle-regulated interaction of the Bloom syndrome helicase BLM with MCM6 controls replication-linked processes. Nucleic Acids Res. 2021 Sep 7; 49(15):8699-8713. doi: 10.1093/nar/gkab663. Citation on PubMed (<https://www.ncbi.nlm.nih.gov/pubmed/34370039>)

## **Genomic Location**

The *MCM6* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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