

LCT gene

lactase

Normal Function

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.

Lactase is produced by some of the cells that line the walls of the small intestine. These cells, called intestinal epithelial cells, have small, finger-like projections called microvilli. As food passes through the intestine, the microvilli collect nutrients so they can be absorbed into the bloodstream. Groups of these microvilli are known collectively as the brush border. Lactase functions at the brush border to break down lactose into smaller sugars called glucose and galactose for absorption.

Health Conditions Related to Genetic Changes

Lactose intolerance

Rarely, *LCT* gene variants (also called mutations) cause congenital lactase deficiency. In this disorder (also known as congenital alactasia), infants are unable to break down lactose in breast milk or formula. Infants who are unable to process lactose are considered lactose intolerant.

Variants in the *LCT* gene can change single protein building blocks (amino acids) in the lactase enzyme. In some cases, variants cause cells to create lactase enzymes that are abnormally short. The variants are believed to interfere with the processing and function of the lactase enzyme, leading to undigested lactose in the small intestine and causing severe diarrhea.

Lactose intolerance in adulthood can also be caused by the gradually decreasing activity (expression) of the *LCT* gene after infancy. This condition is known as lactase nonpersistence, and it occurs in most humans. It can also have non-genetic causes.

Other Names for This Gene

- LAC
- lactase-glycosylceramidase
- lactase-phlorizin hydrolase

- lactase-phlorizin hydrolase preprotein
- lactase-phlorizin hydrolase-1
- LPH
- LPH1
- LPH_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of LCT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3938\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3938[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28LCT%5BTIAB%5D%29+OR+%28lactase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- LACTASE; LCT (<https://omim.org/entry/603202>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3938>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=LCT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=LCT[gene]))

References

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Genomic Location

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

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