



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 other dairy products.

Lactase is produced by cells that line the walls of the small intestine. These cells, called intestinal epithelial cells, have finger-like projections called microvilli that absorb nutrients from food as it passes through the intestine so they can be absorbed into the bloodstream. Based on their appearance, 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

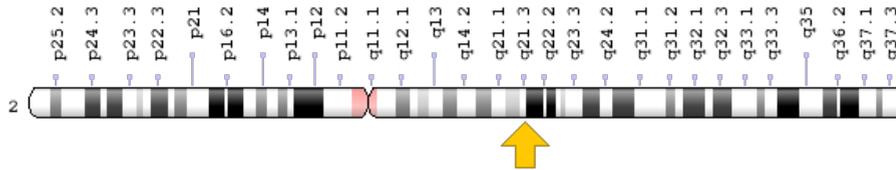
At least nine *LCT* gene mutations cause congenital lactase deficiency, also called congenital alactasia. In this disorder, infants are unable to break down lactose (lactose intolerance) in breast milk or formula. The *LCT* gene mutations change single protein building blocks (amino acids) in the lactase enzyme or result in an enzyme that is abnormally short. The mutations are believed to interfere with the function of the lactase enzyme, leading to undigested lactose in the small intestine and causing severe diarrhea.

Lactose intolerance in adulthood is caused by gradually decreasing activity (expression) of the *LCT* gene after infancy, which occurs in most humans.

Chromosomal Location

Cytogenetic Location: 2q21.3, which is the long (q) arm of chromosome 2 at position 21.3

Molecular Location: base pairs 135,787,840 to 135,837,195 on chromosome 2 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LAC
- lactase-glycosylceramidase
- lactase-phlorizin hydrolase
- lactase-phlorizin hydrolase-1
- lactase-phlorizin hydrolase preproprotein
- LPH
- LPH1
- LPH_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Many Adults are Intolerant of Milk Because They Are Deficient in Lactase
<https://www.ncbi.nlm.nih.gov/books/NBK22593/#A2242>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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
<http://omim.org/entry/603202>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_LCT.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=LCT%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:6530
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:3938>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3938>
- UniProt
<https://www.uniprot.org/uniprot/P09848>

Sources for This Summary

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- Robayo-Torres CC, Nichols BL. Molecular differentiation of congenital lactase deficiency from adult-type hypolactasia. *Nutr Rev.* 2007 Feb;65(2):95-8. Review.
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- Tornaiainen S, Freddara R, Routi T, Gijsbers C, Catassi C, Höglund P, Savilahti E, Järvelä I. Four novel mutations in the lactase gene (LCT) underlying congenital lactase deficiency (CLD). *BMC Gastroenterol.* 2009 Jan 22;9:8. doi: 10.1186/1471-230X-9-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19161632>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2635369/>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/LCT>

Reviewed: May 2010

Published: May 14, 2019

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services