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 135,787,850 to 135,837,195 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (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%29+OR+%28lactase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29%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
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3938
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P09848

Sources for This Summary

- OMIM: LACTASE
  http://omim.org/entry/603202

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