

GEPHE SUMMARY

<p>lactase (LCT) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=*lactase+(LCT)*#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000529</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=*Physiology*#gephebase-summary-title)</p> <p>Lactose tolerance (adult) (https://www.gephebase.org/search-criteria?/and+Trait=*Lactose+tolerance+(adult)*#gephebase-summary-title)</p> <p>Homo sapiens</p> <p>Homo sapiens European with lactase persistence</p> <p>Data not curated</p> <p>Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=*Intraspecific*#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>LCT</p> <p>LAC; LPH; LPH1</p> <p>9606.ENSPO0000264162 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000264162)</p> <p>Belongs to the glycosyl hydrolase 1 family.</p> <p>GO:0008422 : beta-glucosidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008422)</p> <p>GO:0017042 : glycosylceramidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0017042)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P09848 (http://www.uniprot.org/uniprot/P09848)</p> <p>M618636 (https://www.ncbi.nlm.nih.gov/nuccore/M618636)</p> <p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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GO:0000016 : lactase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0000016>)
GO - Biological Process

GO:0005975 : carbohydrate metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005975>)

GO:0044245 : polysaccharide digestion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044245>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

GO:0016324 : apical plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016324>)

GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)

GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

Presumptive Null

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

Cis-regulatory ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type+Cis-regulatory))

Aberration Type

SNP ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type+SNP))

Molecular Details of the Mutation

C-13910T

Experimental Evidence

Linkage Mapping ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence+Linkage+Mapping))

Main Reference

Identification of a variant associated with adult-type hypolactasia. (2002) (<https://pubmed.ncbi.nlm.nih.gov/11788828>)

Authors

Enattah NS; Sahi T; Savilahti E; Terwilliger JD; Peltonen L; Järvelin I

Abstract

Adult-type hypolactasia, also known as lactase non-persistence (lactose intolerance), is a common autosomal recessive condition resulting from the physiological decline in activity of the lactase-pherorizin hydrolase (LPH) in intestinal cells after weaning. LPH hydrolyzes lactose into glucose and galactose. Sequence analyses of the coding and promoter regions of LCT, the gene encoding LPH, has revealed no DNA variations correlating with lactase non-persistence. An associated haplotype spanning LCT, as well as a distinct difference in the transcript levels of 'non-persistence' and 'persistence' alleles in heterozygotes, suggest that a cis-acting element contributes to the lactase non-persistence phenotype. Using linkage disequilibrium (LD) and haplotype analysis of nine extended Finnish families, we restricted the locus to a 47-kb interval on 2q21. Sequence analysis of the complete region and subsequent association analyses revealed that a DNA variant, C/T-13910, roughly 14 kb upstream from the LCT locus, completely associates with biochemically verified lactase non-persistence in Finnish families and a sample set of 236 individuals from four different populations. A second variant, G/A-22018, 8 kb telomeric to C/T-13910, is also associated with the trait in 229 of 236 cases. Prevalence of the C/T-13910 variant in 1,047 DNA samples is consistent with the reported prevalence of adult-type hypolactasia in four different populations. That the variant (C/T-13910) occurs in distantly related populations indicates that it is very old.

Additional References

Convergent adaptation of human lactase persistence in Africa and Europe. (2007) (<https://pubmed.ncbi.nlm.nih.gov/17159977>)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

4 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase+lactase+(LCT)/and+Taxon+ID+9606/or+Gene+Gephebase+lactase+(LCT)/and+Taxon+ID+9606))

EXTERNAL LINKS

COMMENTS