

## GEPHE SUMMARY

<p>lactase (LCT) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=*lactase+(LCT)*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=*lactase+(LCT)*#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000530</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
---	---	---------------------------------	------------------------------------

## PHENOTYPIC CHANGE

<p>Physiology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=*Physiology*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=*Physiology*#gephebase-summary-title</a>)</p> <p>Lactose tolerance (adult) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=*Lactose+tolerance+(adult)*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=*Lactose+tolerance+(adult)*#gephebase-summary-title</a>)</p> <p>Homo sapiens</p> <p>Homo sapiens</p> <p>Data not curated</p> <p>Intraspecific (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=*Intraspecific*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=*Intraspecific*#gephebase-summary-title</a>)</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 (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title</a>)</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) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title</a>)</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) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>NCBI Taxonomy ID</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
---	---	--	--

## GENOTYPIC CHANGE

<p>LCT</p> <p>LAC; LPH; LPH1</p> <p>9606.ENSPO0000264162 (<a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000264162">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000264162</a>)</p> <p>Belongs to the glycosyl hydrolase 1 family.</p> <p>GO:0008422 : beta-glucosidase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008422">https://www.ebi.ac.uk/QuickGO/term/GO:0008422</a>)</p> <p>GO:0017042 : glycosylceramidase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0017042">https://www.ebi.ac.uk/QuickGO/term/GO:0017042</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P09848 (<a href="http://www.uniprot.org/uniprot/P09848">http://www.uniprot.org/uniprot/P09848</a>)</p> <p>M61848 (<a href="https://www.ncbi.nlm.nih.gov/nucleotide/M61848">https://www.ncbi.nlm.nih.gov/nucleotide/M61848</a>)</p> <p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
---	--	--

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 (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>)

Molecular Details of the Mutation

C-13907G

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title>)

Main Reference

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

Authors

Tishkoff SA; Reed FA; Ranciaro A; Voight BF; Babbitt CC; Silverman JS; Powell K; Mortensen HM; Hirbo JB; Osman M; Ibrahim M; Omar SA; Lema G; Nyambo TB; Ghorji J; Bumpstead S; Pritchard JK; Wray GA; Deloukas P

Abstract

A SNP in the gene encoding lactase (LCT) (C/T-13910) is associated with the ability to digest milk as adults (lactase persistence) in Europeans, but the genetic basis of lactase persistence in Africans was previously unknown. We conducted a genotype-phenotype association study in 470 Tanzanians, Kenyans and Sudanese and identified three SNPs (G/C-14010, T/G-13915 and C/G-13907) that are associated with lactase persistence and that have derived alleles that significantly enhance transcription from the LCT promoter in vitro. These SNPs originated on different haplotype backgrounds from the European C/T-13910 SNP and from each other. Genotyping across a 3-Mb region demonstrated haplotype homozygosity extending >2.0 Mb on chromosomes carrying C-14010, consistent with a selective sweep over the past approximately 7,000 years. These data provide a marked example of convergent evolution due to strong selective pressure resulting from shared cultural traits-animal domestication and adult milk consumption.

Additional References

## RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

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

## EXTERNAL LINKS

## COMMENTS