

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>GP00000532</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>Human San hunter gatherer & Bantu speaking farmer groups</p> <p>East Africa allele 1 with lactase persistence ; including Human Nama Khoe pastoralist group</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>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p>
<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title)</p> <p>human</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>species</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>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>No</p>	<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=*Homo+sapiens*#gephebase-summary-title)</p> <p>human</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>species</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>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>No</p>		

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>M61848 (https://www.ncbi.nlm.nih.gov/nucleotide/M61848)</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 ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

G14010C ; In Khoe Pastoralist group lactase persistence (LP)-regulatory region 2 SNPs with greatest frequencies 13910C>T and 14010G>C

Experimental Evidence

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

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

Lactase persistence alleles reveal partial East African ancestry of southern African Khoe pastoralists. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24704072>)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

4 ([#gpepbase-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

@Introgression the LP variants in Khoe result from an introgression from an East African pastoralist population that migrated south prior 1.3 thousands years ago