

GEPHE SUMMARY

lactase (LCT) (<a +lactase+(lct)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase="^lactase (LCT)^#gephebase-summary-title)	Gephebase Gene	GepheID
	GP00001714	
	Entry Status	Main curator
Published	Courtier	

PHENOTYPIC CHANGE

Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category="^Physiology^#gephebase-summary-title)		Trait Category		
Lactose tolerance (adult) (<a +lactose+tolerance+(adult)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+ Trait="^Lactose tolerance (adult)^#gephebase-summary-title)		Trait		
Homo sapiens		Trait State in Taxon A		
Homo sapiens		Trait State in Taxon B		
Taxon A		Ancestral State		
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status="^Intraspecific^#gephebase-summary-title)		Taxonomic Status		
Taxon A		Taxon B		
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+ Taxon and Synonyms="^Homo sapiens^#gephebase-summary-title)		Latin Name	Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+ Taxon and Synonyms="^Homo sapiens^#gephebase-summary-title)	
human		Common Name	human	
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		Synonyms	human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
species		Rank	species	
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		Lineage	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	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Parent	Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		NCBI Taxonomy ID	9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
No		is Taxon A an Intraspecies?	No	
		is Taxon B an Intraspecies?		

GENOTYPIC CHANGE

LCT	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	P09848 (http://www.uniprot.org/uniprot/P09848)
	String	M61848 (https://www.ncbi.nlm.nih.gov/nuccore/M61848)
9606.ENSPO0000264162 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000264162)		
Belongs to the glycosyl hydrolase 1 family.		
GO:0008422 : beta-glucosidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008422)		
GO:0017042 : glycosylceramidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0017042)		

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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
T14009G (rs869051967) (ss 820486563)	Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping~#gephebase-summary-title)	Experimental Evidence
Diversity of lactase persistence alleles in Ethiopia: signature of a soft selective sweep. (2013) (https://pubmed.ncbi.nlm.nih.gov/23993196)	Main Reference
Jones BL; Raga TO; Liebert A; Zmarz P; Bekele E; Danielsen ET; Olsen AK; Bradman N; Troelsen JT; Swallow DM	Authors
	Abstract

The persistent expression of lactase into adulthood in humans is a recent genetic adaptation that allows the consumption of milk from other mammals after weaning. In Europe, a single allele (-13910(âˆ—)T, rs4988235) in an upstream region that acts as an enhancer to the expression of the lactase gene LCT is responsible for lactase persistence and appears to have been under strong directional selection in the last 5,000 years, evidenced by the widespread occurrence of this allele on an extended haplotype. In Africa and the Middle East, the situation is more complicated and at least three other alleles (-13907(âˆ—)G, rs41525747; -13915(âˆ—)G, rs41380347; -14010(âˆ—)C, rs145946881) in the same LCT enhancer region can cause continued lactase expression. Here we examine the LCT enhancer sequence in a large lactose-tolerance-tested Ethiopian cohort of more than 350 individuals. We show that a further SNP, -14009T>G (ss 820486563), is significantly associated with lactose-digester status, and inÂ vitro functional tests confirm that the -14009(âˆ—)G allele also increases expression of an LCT promoter construct. The derived alleles in the LCT enhancer region are spread through several ethnic groups, and we report a greater genetic diversity in lactose digesters than in nondigesters. By examining flanking markers to control for the effects of mutation and demography, we further describe, from empirical evidence, the signature of a soft selective sweep.

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In Vitro Functional Analyses of Infrequent Nucleotide Variants in the Lactase Enhancer Reveal Different Molecular Routes to Increased Lactase Promoter Activity and Lactase Persistence. (2016) (https://pubmed.ncbi.nlm.nih.gov/27714771)	

RELATED GEPHE

No matches found.	Related Genes
4 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~lactase (LCT)^/and+Taxon ID=~9606~/or+Gene Gephebase=~lactase (LCT)^/and+Taxon ID=~9606~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS