GEPHE SUMMARY

Gephebase Gene
lactase (LCT) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^lactase (LCT)^#gephebase-summary-title)

GepheID
GP00001714

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Lactose tolerance (adult) (https://www.gephebase.org/search-criteria?/and+Trait=^Lactose tolerance (adult)^#gephebase-summary-title)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Taxon A

Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

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

Rank
species

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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

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

Rank
species

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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
LCT

Synonyms
LAC; LPH; LPH1

String
9606.ENSP00000264162 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000264162)

Sequence Similarities
Belongs to the glycosyl hydrolase 1 family.

GO - Molecular Function
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)

UniProtKB Homo sapiens
P09848 (http://www.uniprot.org/uniprot/P09848)

GenebankID or UniProtKB
M61848 (https://www.ncbi.nlm.nih.gov/nuccore/M61848)

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

T14009G (rs869051967) (ss 820486563)

Experimental Evidence

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

Main Reference

Diversity of lactase persistence alleles in Ethiopia: signature of a soft selective sweep. (2013) (https://pubmed.ncbi.nlm.nih.gov/23993196)

Authors

Jones BL; Raga TO; Liebert A; Zmarz P; Bekele E; Danielsen ET; Olsen AK; Bradman N; Troelsen JT; Swallow DM

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(â^—)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(â^—)G, rs41525747; -13915(â^—)G, rs41380347; -14010(â^—)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(â^—)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.

Additional References

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

Related Genes

No matches found.

Related Haplotypes

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)

EXTERNAL LINKS

COMMENTS