GEPHE SUMMARY

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

GepheID
GP00000532

Entry Status
Published

Main curator
Martin

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
Human San hunter gatherer & Bantu speaking farmer groups

Trait State in Taxon B
East Africa allele 1 with lactase persistence ; including Human Nama Khoe pastoralist group

Ancestral State
Data not curated

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

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

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; Ghori 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 (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

@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