GEPHE SUMMARY

Gephebase Gene
lactase (LCT)

GephelD
GP00000531

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Lactose tolerance (adult)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Taxon A

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
LCT

Synonyms
LAC; LPH; LPH1

String
9606.ENSP00000264162

Sequence Similarities
Belongs to the glycosyl hydrolase 1 family.

GO - Molecular Function
GO:0008422 : beta-glucosidase activity
GO:0017042 : glycosylceramidase activity
GO:0000016 : lactase activity

GO - Biological Process
GO:0005975 : carbohydrate metabolic process
GO:0044245 : polysaccharide digestion

GO - Cellular Component

UniProtKB Homo sapiens
P09848

GenebankID or UniProtKB
M61848

GO:0005886 : plasma membrane
GO:0016324 : apical plasma membrane
GO:0016020 : membrane
GO:0005887 : integral component of plasma membrane

Presumptive Null
No

Molecular Type
Cis-regulatory

Aberration Type
SNP

Molecular Details of the Mutation
T-13915G

Experimental Evidence
Association Mapping

Main Reference
Convergent adaptation of human lactase persistence in Africa and Europe. (2007)

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

RELATED GEPHE

Related Genes
No matches found.
Related Haplotypes
4

COMMENTS