

## GEPHE SUMMARY

**Gephebase Gene**  
lactase (LCT)

**Entry Status**  
Published

**GepheID**  
GP00000530

**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**  
Data not curated

**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 Intraspecies?**  
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 Intraspecies?**  
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**

C-13907G

**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**