GEPHE SUMMARY

Gephebase Gene
PMEL17

GephelD
GP00001365

Entry Status
Published

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
domestic yak ; wild type black

Trait State in Taxon B
domestic yak ; brown

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Bos grunniens

Common Name
domestic yak

Synonyms
Bos mutus grunniens; Poephagus grunniens; domestic yak; yak

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; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus)

NCBI Taxonomy ID
30521

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Bos grunniens

Common Name
domestic yak

Synonyms
Bos mutus grunniens; Poephagus grunniens; domestic yak; yak

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; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus)

NCBI Taxonomy ID
30521

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
PMEL

Synonyms
SILV; PMEL17; P1; SI; SIL; ME20; P100; ME20M; gp100; ME20-M; D12S53E

String
9606.ENSP00000402758

Sequence Similarities
Belongs to the PMEL/NMB family.

GO - Molecular Function
GO:0042802 : identical protein binding

GO - Biological Process
GO:0042438 : melanin biosynthetic process
GO:0032438 : melanosome organization

GO - Cellular Component
GO:0005886 : plasma membrane
GO:0005887 : integral component of plasma membrane
GO:0005576 : extracellular region
GO:0005794 : Golgi apparatus
GO:0005789 : endoplasmic reticulum membrane

UniProtKB Homo sapiens
P40967

GenebankID or UniProtKB

GO:0042470 : melanosome
GO:0032585 : multivesicular body membrane

Presumptive Null
No

Molecular Type
Coding

Aberration Type
Deletion

Deletion Size
1-9 bp

Molecular Details of the Mutation
c.50_52del p.Leu18del in signal peptide

Experimental Evidence
Candidate Gene

Main Reference
The genetics of brown coat color and white spotting in domestic yaks (Bos grunniens). (2014)

Authors
Zhang MQ; Xu X; Luo SJ

Abstract
Domestic yaks (Bos grunniens) exhibit two major coat color variations: a brown vs. wild-type black pigmentation and a white spotting vs. wild-type solid color pattern. The genetic basis for these variations in color and distribution remains largely unknown and may be complicated by a breeding history involving hybridization between yaks and cattle. Here, we investigated 92 domestic yaks from China using a candidate gene approach. Sequence variations in MC1R, PMEL and TYRP1 were surveyed in brown yaks; TYRP1 was unassociated with the coloration and excluded. Recessive mutations from MC1R, or p.Gln34*, p.Met73Leu and possibly p.Arg142Pro, are reported in bovids for the first time and accounted for approximately 40% of the brown yaks in this study. The remaining 60% of brown individuals correlated with a cattle-derived deletion mutation from PMEL (p.Leu18del) in a dominant manner. Degrees of white spotting found in yaks vary from color sidedness and white face, to completely white. After examining the candidate gene KIT, we suggest that color-sided and all-white yaks are caused by the serial translations of KIT (Cs6 or Cs29) as reported for cattle. The white-faced phenotype in yaks is associated with the KIT haplotype S(wf) . All KIT mutations underlying the serial phenotypes of white spotting in yaks are identical to those in cattle, indicating that cattle are the likely source of white spotting in yaks. Our results reveal the complex genetic origins of domestic yak coat color as either native in yaks through evolution and domestication or as introduced from cattle through interspecific hybridization.

Additional References

RELATED GEPHE

Related Genes
2 (Kit (type III receptor protein-tyrosine kinase) [pseudoreplicate with two Bos taurus KIT entries], MC1R)
Related Haplotypes
1

EXTERNAL LINKS

COMMENTS

same mutation reported in cattle