

## GEPHE SUMMARY

PMEL17 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~PMEL17~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~PMEL17~#gephebase-summary-title</a> )	Gephebase Gene	GP00001365	GepheID
Published	Entry Status	Prigent	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title</a> )	Trait Category		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title</a> )	Trait		
domestic yak ; wild type black	Trait State in Taxon A		
domestic yak ; brown	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Bos grunniens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+grunniens~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+grunniens~#gephebase-summary-title</a> )	Latin Name	Bos grunniens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+grunniens~#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+grunniens~#gephebase-summary-title</a> )	Latin Name
domestic yak	Common Name	domestic yak	Common Name
Bos mutus grunniens; Poephagus grunniens; domestic yak; yak	Synonyms	Bos mutus grunniens; Poephagus grunniens; domestic yak; yak	Synonyms
species	Rank	species	Rank
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	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	Lineage
Bos (oxen, cattle) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a> )	Parent	Bos (oxen, cattle) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a> )	Parent
30521 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521</a> )	NCBI Taxonomy ID	30521 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

PMEL	Generic Gene Name	P40967 ( <a href="http://www.uniprot.org/uniprot/P40967">http://www.uniprot.org/uniprot/P40967</a> )	UniProtKB Homo sapiens
SILV; PMEL17; P1; S1; SIL; ME20; P100; ME20M; gp100; ME20-M; D12S53E	Synonyms	()	GenebankID or UniProtKB
9606.ENSPO0000402758 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000402758">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000402758</a> )	String		
Belongs to the PMEL/NMB family.	Sequence Similarities		
GO:0042802 : identical protein binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042802">https://www.ebi.ac.uk/QuickGO/term/GO:0042802</a> )	GO - Molecular Function		
GO:0042438 : melanin biosynthetic process ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042438">https://www.ebi.ac.uk/QuickGO/term/GO:0042438</a> )	GO - Biological Process		
GO:0032438 : melanosome organization ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0032438">https://www.ebi.ac.uk/QuickGO/term/GO:0032438</a> )			

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)  
 GO:0005887 : integral component of plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)  
 GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)  
 GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)  
 GO:0005789 : endoplasmic reticulum membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>)  
 GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)  
 GO:0032585 : multivesicular body membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0032585>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>) Presumptive Null

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title>) Molecular Type

Deletion (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title>) Aberration Type

1-9 bp Deletion Size

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

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title>) Experimental Evidence

The genetics of brown coat color and white spotting in domestic yaks (*Bos grunniens*). (2014) (<https://pubmed.ncbi.nlm.nih.gov/24989079>) Main Reference

Zhang MQ; Xu X; Luo SJ Authors

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.

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Additional References

RELATED GEPHE

2 (Kit (type III receptor protein-tyrosine kinase) [pseudoreplicate with two *Bos taurus* KIT entries], MC1R) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^30521^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>) Related Genes

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^30521^/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^30521^#gephebase-summary-title>) Related Haplotypes

EXTERNAL LINKS

COMMENTS

same mutation reported in cattle