

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00001354	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait Category		
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration(coat)^#gephebase-summary-title)	Trait		
domestic yak ; wild type black	Trait State in Taxon A		
domestic yak ; brown	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)	Taxonomic Status		

Taxon A	Latin Name	Taxon B	Latin Name
Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+grunniens^#gephebase-summary-title)	Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+grunniens^#gephebase-summary-title)	Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+grunniens^#gephebase-summary-title)	Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+grunniens^#gephebase-summary-title)
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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	Parent	Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	Parent
30521 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521)	NCBI Taxonomy ID	30521 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=30521)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Mcr1	Generic Gene Name	Q01727 (http://www.uniprot.org/uniprot/Q01727)	UniProtKB Mus musculus
e; Tob; Mcr1; Mshra; Msh-r	Synonyms	0	GenebankID or UniProtKB
10090.ENSMUSP00000095929 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000095929)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)	GO - Molecular Function		
GO:0004980 : melanocyte-stimulating hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004980)			
GO:0031625 : ubiquitin protein ligase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031625)			
GO:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562)			

GO - Biological Process

- GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
- GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
- GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
- GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
- GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)
- GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)
- GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)
- GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
- GO:0010739 : positive regulation of protein kinase A signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010739>)
- GO:0090037 : positive regulation of protein kinase C signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090037>)
- GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)
- GO:2000253 : positive regulation of feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000253>)
- GO:0060259 : regulation of feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060259>)

GO - Cellular Component

- GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
- GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

Yes ([#gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes)) Presumptive Null

Coding ([#gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding)) Molecular Type

SNP ([#gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP)) Aberration Type

Nonsense SNP Coding Change

p.Gln34* and p.Met73Leu and p.Arg142Pro - p.Met73Leu and p.Arg142Pro were only found in heterozygous and their association with phenotype is supposed but not sure Molecular Details of the Mutation

Candidate Gene ([#gpebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene)) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gln	STP	34

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. Abstract

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Additional References

RELATED GEPHE

2 (Kit (type III receptor protein-tyrosine kinase) [pseudoreplicate with two *Bos taurus* KIT entries], PMEL17) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~30521#/and+Trait=Coloration/and+groupHaplotypes=true#gpebase-summary-title>) Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

p.Met73Leu and p.Arg142Pro were only found in heterozygous and their association with phenotype is supposed but not sure