

GEPHE SUMMARY

Kit (type III receptor protein-tyrosine kinase) [pseudoreplicate with two Bos taurus KIT entries] ([https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit \(type III receptor protein-tyrosine kinase\) \[pseudoreplicate with two Bos taurus KIT entries\]^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)+[pseudoreplicate+with+two+Bos+taurus+KIT+entries]^#gephebase-summary-title))

Gephebase Gene GP00001342 GepheID

Prigent Main curator

Entry Status

Published

PHENOTYPIC CHANGE

Morphology (<https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title>)

Trait Category

Coloration (coat) ([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))

Trait

domestic yak ; wild type solid black

Trait State in Taxon A

domestic yak ; white spotting color sided and all white

Trait State in Taxon B

Ancestral State

Taxon A

Taxonomic Status

Domesticated (<https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title>)

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	Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos grunniens^#gephebase-summary-title)	Bos grunniens
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 Infrasppecies?	No	is Taxon B an Infrasppecies?

GENOTYPIC CHANGE

Kit

Generic Gene Name P05532 (<http://www.uniprot.org/uniprot/P05532>) UniProtKB Mus musculus

W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; SI

Synonyms () GenebankID or UniProtKB

String

10090.ENSMUSP00000005815
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815)

Sequence Similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.

GO - Molecular Function

GO:0004888 : transmembrane signaling receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004888>)

GO:0005524 : ATP binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005524>)

GO:0042803 : protein homodimerization activity

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)
GO:0002020 : protease binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0002020>)
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004714>)
GO:0004713 : protein tyrosine kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004713>)
GO:0019955 : cytokine binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019955>)
GO:0005020 : stem cell factor receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005020>)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)
GO:0035234 : ectopic germ cell programmed cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035234>)
GO:0035162 : embryonic hemopoiesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035162>)
GO:0008584 : male gonad development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008584>)
GO:0001541 : ovarian follicle development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001541>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:0043406 : positive regulation of MAP kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043406>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0043410 : positive regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)
GO:0007283 : spermatogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0007283>)
GO:0008360 : regulation of cell shape
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008360>)
GO:0048070 : regulation of developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048070>)
GO:0006468 : protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006468>)
GO:0060326 : cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060326>)
GO:0006935 : chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0006935>)
GO:0048565 : digestive tract development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048565>)
GO:0006954 : inflammatory response
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006954>)
GO:0019221 : cytokine-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019221>)
GO:0048863 : stem cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048863>)
GO:0048066 : developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0009968 : negative regulation of signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009968>)
GO:0046777 : protein autophosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)
GO:0030218 : erythrocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030218>)
GO:0018108 : peptidyl-tyrosine phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018108>)
GO:0097067 : cellular response to thyroid hormone stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097067>)
GO:1904349 : positive regulation of small intestine smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904349>)
GO:0000187 : activation of MAPK activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000187>)
GO:0046427 : positive regulation of JAK-STAT cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046427>)
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042531>)
GO:0030335 : positive regulation of cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)
GO:0046686 : response to cadmium ion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046686>)
GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)

GO:0031532 : actin cytoskeleton reorganization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031532>)

GO:0002371 : dendritic cell cytokine production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002371>)

GO:0050910 : detection of mechanical stimulus involved in sensory perception of sound
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050910>)

GO:0050673 : epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050673>)

GO:0038162 : erythropoietin-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038162>)

GO:0038093 : Fc receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0038093>)

GO:0007281 : germ cell development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007281>)

GO:0008354 : germ cell migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0008354>)

GO:0006687 : glycosphingolipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006687>)

GO:0035701 : hematopoietic stem cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035701>)

GO:0030097 : hemopoiesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0030097>)

GO:0002327 : immature B cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002327>)

GO:0038109 : Kit signaling pathway (<https://www.ebi.ac.uk/QuickGO/term/GO:0038109>)

GO:0030032 : lamellipodium assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030032>)

GO:0002320 : lymphoid progenitor cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002320>)

GO:0002551 : mast cell chemotaxis (<https://www.ebi.ac.uk/QuickGO/term/GO:0002551>)

GO:0032762 : mast cell cytokine production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032762>)

GO:0043303 : mast cell degranulation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043303>)

GO:0060374 : mast cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060374>)

GO:0035855 : megakaryocyte development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035855>)

GO:0097326 : melanocyte adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0097326>)

GO:0097324 : melanocyte migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0097324>)

GO:0002573 : myeloid leukocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002573>)

GO:0002318 : myeloid progenitor cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002318>)

GO:0043069 : negative regulation of programmed cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043069>)

GO:1904343 : positive regulation of colon smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904343>)

GO:0051091 : positive regulation of DNA-binding transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051091>)

GO:0048170 : positive regulation of long-term neuronal synaptic plasticity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048170>)

GO:0045747 : positive regulation of Notch signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045747>)

GO:0031274 : positive regulation of pseudopodium assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031274>)

GO:0120072 : positive regulation of pyloric antrum smooth muscle contraction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0120072>)

GO:1905065 : positive regulation of vascular smooth muscle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1905065>)

GO:1904251 : regulation of bile acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904251>)

GO:0009314 : response to radiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0009314>)

GO:0048103 : somatic stem cell division
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048103>)

GO:0035019 : somatic stem cell population maintenance
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035019>)

GO:0007286 : spermatid development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007286>)

GO:0030217 : T cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030217>)

GO:0043586 : tongue development (<https://www.ebi.ac.uk/QuickGO/term/GO:0043586>)

GO:0008542 : visual learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0008542>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)

GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)

GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)

GO:0009898 : cytoplasmic side of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009898>)
GO:0009897 : external side of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009897>)
GO:0001669 : acrosomal vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0001669>)
GO:0005911 : cell-cell junction (<https://www.ebi.ac.uk/QuickGO/term/GO:0005911>)
GO:0042629 : mast cell granule (<https://www.ebi.ac.uk/QuickGO/term/GO:0042629>)

Presumptive Null

Unknown (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Unknown^#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

serial translocations Cs6 and Cs29

Experimental Evidence

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

Main Reference

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

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.

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Additional References

RELATED GEPHE

Related Genes

2 (MC1R, PMEL17) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~30521^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^Kit+\(type+III+receptor+protein-tyrosine+kinase\)+\[pseudoreplicate+with+two+Bos+taurus+KIT+entries\]^/and+Taxon+ID=~30521^/or+Gene+Gephebase=^Kit+\(type+III+receptor+protein-tyrosine+kinase\)+\[pseudoreplicate+with+two+Bos+taurus+KIT+entries\]^/and+Taxon+ID=~30521^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)+[pseudoreplicate+with+two+Bos+taurus+KIT+entries]^/and+Taxon+ID=~30521^/or+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)+[pseudoreplicate+with+two+Bos+taurus+KIT+entries]^/and+Taxon+ID=~30521^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

©Introgression Dominant mutations - mutations are identical in cattle which are the likely source through hybridization