

GEPHE SUMMARY

Kit ligand (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+ligand^#gephebase-summary-title)	Gephebase Gene	GP00000101	GepheID
Published	Entry Status	Martin	Main curator

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)	Trait		
Domesticated cattle	Trait State in Taxon A		
Roan phenotype in breeds Belgian Blue ; Shorthorn	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+taurus^#gephebase-summary-title)	Latin Name	Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Bos+taurus^#gephebase-summary-title)	Latin Name
cattle	Common Name	cattle	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	Synonyms	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	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; Artiodactyla; 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; Artiodactyla; 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
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	NCBI Taxonomy ID	9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
			Taxon B Description
		-	

GENOTYPIC CHANGE

KITLG	Generic Gene Name	P21583 (http://www.uniprot.org/uniprot/P21583)	UniProtKB Homo sapiens
SF; MGF; SCF; SLF; DCUA; FPH2; FPHH; KL-1; Kitl; SHEP7; DFNA69	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000228280 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000228280)	String		
Belongs to the SCF family.	Sequence Similarities		
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)	GO - Molecular Function		
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)			
GO:0046934 : phosphatidylinositol-4,5-bisphosphate 3-kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046934)			

GO:0005088 : Ras guanyl-nucleotide exchange factor activity
 (https://www.ebi.ac.uk/QuickGO/term/GO:0005088)
 GO:0005173 : stem cell factor receptor binding
 (https://www.ebi.ac.uk/QuickGO/term/GO:0005173)

GO - Biological Process

GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155)
 GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
 GO:0001755 : neural crest cell migration
 (https://www.ebi.ac.uk/QuickGO/term/GO:0001755)
 GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
 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:0097192 : extrinsic apoptotic signaling pathway in absence of ligand
 (https://www.ebi.ac.uk/QuickGO/term/GO:0097192)
 GO:0008584 : male gonad development
 (https://www.ebi.ac.uk/QuickGO/term/GO:0008584)
 GO:0000165 : MAPK cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0000165)
 GO:0033026 : negative regulation of mast cell apoptotic process
 (https://www.ebi.ac.uk/QuickGO/term/GO:0033026)
 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:1902035 : positive regulation of hematopoietic stem cell proliferation
 (https://www.ebi.ac.uk/QuickGO/term/GO:1902035)
 GO:0002687 : positive regulation of leukocyte migration
 (https://www.ebi.ac.uk/QuickGO/term/GO:0002687)
 GO:0043406 : positive regulation of MAP kinase activity
 (https://www.ebi.ac.uk/QuickGO/term/GO:0043406)
 GO:0070668 : positive regulation of mast cell proliferation
 (https://www.ebi.ac.uk/QuickGO/term/GO:0070668)
 GO:0045636 : positive regulation of melanocyte differentiation
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045636)
 GO:0002763 : positive regulation of myeloid leukocyte differentiation
 (https://www.ebi.ac.uk/QuickGO/term/GO:0002763)
 GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
 (https://www.ebi.ac.uk/QuickGO/term/GO:0050731)
 GO:0051897 : positive regulation of protein kinase B signaling
 (https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
 GO:0046579 : positive regulation of Ras protein signal transduction
 (https://www.ebi.ac.uk/QuickGO/term/GO:0046579)

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)
 GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
 GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
 GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
 GO:0005856 : cytoskeleton (https://www.ebi.ac.uk/QuickGO/term/GO:0005856)
 GO:0030175 : filopodium (https://www.ebi.ac.uk/QuickGO/term/GO:0030175)
 GO:0030027 : lamellipodium (https://www.ebi.ac.uk/QuickGO/term/GO:0030027)

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=[^]Yes[^]#gephebase-summary-title) Presumptive Null
 Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=[^]Coding[^]#gephebase-summary-title) Molecular Type
 SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=[^]SNP[^]#gephebase-summary-title) Aberration Type
 Nonsynonymous SNP Coding Change
 Ala193Asp ; missense mutation at position 654 - according to OMI: c.572C>A p.A191N Molecular Details of the Mutation
 Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=[^]Linkage Mapping[^]#gephebase-summary-title) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Asn	191

Main Reference

A missense mutation in the bovine MGF gene is associated with the roan phenotype in Belgian Blue and Shorthorn cattle. (1999) (https://pubmed.ncbi.nlm.nih.gov/10384045)

Authors

Seitz JJ; Schmutz SM; Thue TD; Buchanan FC

Abstract

The Roan locus is responsible for the coat coloration of Belgian Blue and Shorthorn cattle. The solid-colored and white animals are homozygotes, and the roan animals, with intermingled colored and white hairs, are heterozygous. The roan phenotype was mapped to cattle Chromosome (Chr) 5 with microsatellites, and a candidate gene was proposed (Charlier et al. Mamm Genome 7, 138, 1996). PCR primers to the exons of this candidate gene, the steel locus or mast cell growth factor (MGF) were designed. Solid-colored and white animals were sequenced. A missense mutation at 654 bp (amino acid 193, Ala --> Asp) was detected in these two groups. A PCR-RFLP was designed to this single base pair change, and 143 animals in total (Belgian Blue, Shorthorn, and various other breeds) were screened. In addition, the Canadian Beef Cattle Reference Herd (<http://skyway.usask.ca/> approximately schmutz) was used to verify Mendelian inheritance of this marker with the phenotypic inheritance of roan. Our data suggest that this mutation in the bovine MGF gene is responsible for the roan phenotype.

Additional References

Two polymorphisms in the bovine mast cell growth factor gene (MGF). (2000) (<https://pubmed.ncbi.nlm.nih.gov/11105228>)

RELATED GEPHE

Related Genes

11 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PMEL17, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+TaxonID=^9913^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001216/9913/>