

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000572	GepheID
	Entry Status	Martin	Main curator
	Published		

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	
Bos taurus		Trait State in Taxon A	
Bos taurus		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		Taxon B	
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+taurus^#gephebase-summary-title)		Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Bos+taurus^#gephebase-summary-title)	
cattle		cattle	
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus		Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	
species		species	
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		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	
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)		Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)		9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q01726 (http://www.uniprot.org/uniprot/Q01726)
	String	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		NP_776533 (https://www.ncbi.nlm.nih.gov/nuccore/NP_776533)
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)		
Belongs to the G-protein coupled receptor 1 family.		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)		
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)		
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 - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
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:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
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:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger (<https://www.ebi.ac.uk/QuickGO/term/GO:0007187>)
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:0009650 : UV protection (<https://www.ebi.ac.uk/QuickGO/term/GO:0009650>)
GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

GO - Cellular Component

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>)

	Presumptive Null
No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	
	Molecular Type
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	
	Aberration Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	
	SNP Coding Change
Nonsynonymous	
	Molecular Details of the Mutation
L99P ; presumably constitutively active - g.14757910T>C c.296T>C p.L99P	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

	Main Reference
The role of melanocyte-stimulating hormone (MSH) receptor in bovine coat color determination. (1995) (https://pubmed.ncbi.nlm.nih.gov/8535072)	
	Authors
Klungland H; VÅge DI; Gomez-Raya L; Adalsteinsson S; Lien S	
	Abstract
The melanocyte-stimulating hormone (MSH) receptor has a major function in the regulation of black (eumelanin) versus red (phaeomelanin) pigment synthesis within melanocytes. We report three alleles of the MSH-receptor gene found in cattle. A point mutation in the dominant allele ED gives black coat color, whereas a frameshift mutation, producing a prematurely terminated receptor, in homozygous e/e animals, produces red coat color. The wild-type allele E+ produces a variety of colors, reflecting the possibilities for regulating the normal receptor. Microsatellite analysis, RFLP studies, and coat color information were used to localize the MSH-receptor to bovine Chromosome (Chr) 18.	
	Additional References

RELATED GEPHE

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

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001199/9913/>