

GEPHE SUMMARY

MC1R ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase= <sup>^</sup> MC1R <sup>^</sup> #gephebase-summary-title)	Gephebase Gene	GP00001350	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Morphology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> = <sup>^</sup> Coloration (coat) <sup>^</sup> #gephebase-summary-title)	Trait		
Reindeer ; wild type light ventral color	Trait State in Taxon A		
Reindeer ; dark ventral color	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status= <sup>^</sup> Domesticated <sup>^</sup> #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Rangifer tarandus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Rangifer tarandus <sup>^</sup> #gephebase-summary-title)	Latin Name	Rangifer tarandus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Rangifer tarandus <sup>^</sup> #gephebase-summary-title)	Latin Name
reindeer	Common Name	reindeer	Common Name
reindeer; caribou; Rangifer tarandus Linnaeus	Synonyms	reindeer; caribou; Rangifer tarandus Linnaeus	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; Cervidae; Odocoileinae; Rangifer	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; Cervidae; Odocoileinae; Rangifer	Lineage
Rangifer () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9869">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9869</a> )	Parent	Rangifer () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9869">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9869</a> )	Parent
9870 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9870">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9870</a> )	NCBI Taxonomy ID	9870 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9870">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9870</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

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

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>)

Mutation #1

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

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

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

Nonsynonymous

c.218T>C p.Met73Thr and c.839T>G p.Phe280Cys

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Met	Thr	73

Main Reference

Two missense mutations in melanocortin 1 receptor (MC1R) are strongly associated with dark ventral coat color in reindeer (*Rangifer tarandus*). (2014)

(<https://pubmed.ncbi.nlm.nih.gov/25039753>)

Authors

VÃ¶ge DI; Nieminen M; Anderson DG; RÃed KH

Abstract

The protein-coding region of melanocortin 1 receptor (MC1R) was sequenced to identify potential variation affecting coat color in reindeer (*Rangifer tarandus*). A TÃC sequence variation at nucleotide position 218 (c.218T>C) causing an amino acid (aa) change from methionine to threonine at aa position 73 (p.Met73Thr) was identified. In addition, a TÃG sequence variation was found at nucleotide position 839 (c.839T>G), causing phenylalanine to be exchanged by cysteine at aa position 280 (p.Phe280Cys). The two sequence variants (c.218C and c.839G) were found to be closely associated with a darker belly coat compared with animals not having any of these two variants. The aa acid change p.Met73Thr affects the same position as p.Met73Lys previously reported to give constitutive activation of MC1R in black sheep (*Ovis aries*), whereas p.Phe280Cys is identical to one of two variants previously reported to be associated with dark coat color in Arctic fox (*Alopex lagopus*), supporting that the two variants found in reindeer are functional. The complete absence of Thr73 and Cys280 among the 51 wild reindeer analyzed provides some evidence that these variants are more common in the domestic herds.

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Additional References

Mutation #2

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

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

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

Presumptive Null

Molecular Type

Aberration Type

Nonsynonymous

Molecular Details of the Mutation

c.218T&gt;C p.Met73Thr and c.839T&gt;G p.Phe280Cys

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Phe	Cys	280

Main Reference

Two missense mutations in melanocortin 1 receptor (MC1R) are strongly associated with dark ventral coat color in reindeer (*Rangifer tarandus*). (2014) (<https://pubmed.ncbi.nlm.nih.gov/25039753>)

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Additional References

## RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

p.Phe280Cys is also known in Arctic fox (*Alopex lagopus*) ; p.Met73 is also mutated in black sheep (*Ovis aries*)