GEPHE SUMMARY

Gephebase Gene

MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC1R^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001350

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

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

Trait

Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)

Trait State in Taxon A

Reindeer ; wild type light ventral color

Trait State in Taxon B

Reindeer ; dark ventral color

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

Rangifer tarandus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Rangifer tarandus^#gephebase-summary-title)

Common Name

reindeer

Synonyms

reindeer; caribou; Rangifer tarandus Linnaeus

Rank

species

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

Parent

Rangifer () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9869)

NCBI Taxonomy ID

9870 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9870)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Rangifer tarandus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Rangifer tarandus^#gephebase-summary-title)

Common Name

reindeer

Synonyms

reindeer; caribou; Rangifer tarandus Linnaeus

Rank

species

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

Parent

Rangifer () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9869)

NCBI Taxonomy ID

9870 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9870)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Mc1r

Synonyms

e; Tob; Mcr1; Mshra; Msh-r

String

10090.ENSMUSP00000095929 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000095929)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

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:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562)

UniProtKB Mus musculus

Q01727 (http://www.uniprot.org/uniprot/Q01727)

GenebankID or UniProtKB

()

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

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

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

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	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 Tâ†’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 Tâ†’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

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

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

Experimental Evidence

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

	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)

Authors

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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