GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00001351

Entry Status

Published

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

Donkey ; wild type not red

Trait State in Taxon B

Donkey ; Red Normand and Red miniature ; red coat

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

ass

Synonyms

ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Perissodactyla; Equidae; Equus; Asinus

Parent

Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID

9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

ass

Synonyms

ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Perissodactyla; Equidae; Equus; Asinus

Parent

Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID

9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Donkey ; Red Normand and Red miniature ; red coat

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

UniProtKB Mus musculus

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

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0031625)
GO:0042562 : hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042562)

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)

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.629T>C p.Met210Thr

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

A missense mutation in melanocortin 1 receptor is associated with the red coat colour in donkeys. (2014) (https://pubmed.ncbi.nlm.nih.gov/25155046)

Authors

Abitbol M; Legrand R; Tiret L

Abstract

The seven donkey breeds recognised by the French studbook are characterised by few coat colours: black, bay and grey. Normand bay donkeys seldom give birth to red foals, a colour more commonly seen and recognised in American miniature donkeys. Red resembles the equine chestnut colour, previously attributed to a mutation in the melanocortin 1 receptor gene (MC1R). We used a panel of 124 donkeys to identify a recessive missense c.629T>C variant in MC1R that showed a perfect association with the red coat colour. This variant leads to a methionine to threonine substitution at position 210 in the protein. We showed that methionine 210 is highly conserved among vertebrate melanocortin receptors. Previous in silico and in vitro analyses predicted this residue to lie within a functional site. Our in vivo results emphasised the pivotal role played by this residue, the alteration of which yielded a phenotype fully compatible with a loss of function of MC1R. We thus propose to name the c.629T>C allele in donkeys the e allele, which further enlarges the panel of recessive MC1R loss-of-function alleles described in animals and humans.

Additional References

RELATED GEPHE

Related Genes

3 (Agouti (ASIP), Kit (type III receptor protein-tyrosine kinase), tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9793^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

recessive mutation ; evidence of loss of function