

## GEPHE SUMMARY

<p>MC1R (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001351</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Coloration (coat) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+coat^#gephebase-summary-title</a>)</p> <p>Donkey ; wild type not red</p> <p>Donkey ; Red Normand and Red miniature ; red coat</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Equus asinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+asinus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+asinus^#gephebase-summary-title</a>)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508</a>)</p> <p>9793 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793</a>)</p> <p>No</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Equus asinus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+asinus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+asinus^#gephebase-summary-title</a>)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508</a>)</p> <p>9793 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793</a>)</p> <p>Yes</p> <p>Donkey ; Red Normand and Red miniature ; red coat</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
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## GENOTYPIC CHANGE

<p>Mcr1</p> <p>e; Tob; Mcr1; Mshra; Msh-r</p> <p>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>)</p> <p>Belongs to the G-protein coupled receptor 1 family.</p> <p>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>)</p> <p>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>)</p> <p>GO:0031625 : ubiquitin protein ligase binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>Q01727 (<a href="http://www.uniprot.org/uniprot/Q01727">http://www.uniprot.org/uniprot/Q01727</a>)</p> <p>0</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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(<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>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#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  
 c.629T>C p.Met210Thr Molecular Details of the Mutation  
 Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title>) Experimental Evidence

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

A missense mutation in melanocortin 1 receptor is associated with the red coat colour in donkeys. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25155046>) Main Reference  
 Abitbol M; Legrand R; Tired L Authors  
 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. Abstract

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Additional References

RELATED GEPHE

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 Genes

No matches found.

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

recessive mutation ; evidence of loss of function