

GEPHE SUMMARY

	Gephebase Gene		GepheID
MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R~#gephebase-summary-title)		GP00000586	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~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		
Mus musculus		Trait State in Taxon B	
Mus musculus			
	Ancestral State		
Data not curated		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific~#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Mus+musculus~#gephebase-summary-title)		Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Mus+musculus~#gephebase-summary-title)	
	Common Name		Common Name
house mouse		house mouse	
	Synonyms		Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid		house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	
	Rank		Rank
species		species	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus	
	Parent		Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)		Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)		10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
MC1R		Q01726 (http://www.uniprot.org/uniprot/Q01726)	
	Synonyms		GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		BC119294 (https://www.ncbi.nlm.nih.gov/nucore/BC119294)	
	String		
9606.ENSP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605)			
	Sequence Similarities		
Belongs to the G-protein coupled receptor 1 family.			
	GO - Molecular Function		
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

W252C

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 nucleotide substitution responsible for the tawny coat color mutation carried by the MSKR inbred strain of mice. (2005 Mar-Apr) (<https://pubmed.ncbi.nlm.nih.gov/15653560>)

Authors

Wada A; Kunieda T; Nishimura M; Kakizoe-Ishida Y; Watanabe N; Ohkawa K; Tsudzuki M

Abstract

“Tawny” is an autosomal recessive coat color mutation found in a wild population of *Mus musculus molossinus*. The inbred strain MSKR carries the mutation. The causative gene *Mc1r*(taw) of the tawny phenotype is the second recessive allele at the melanocortin 1 receptor locus and is dominant to the first recessive allele, “recessive yellow” (*Mc1r*(e)). The *Mc1r*(taw) gene has six nucleotide substitutions, and its forecasted transcript has three amino acid substitutions (i.e., V101A, V216A, W252C). Though the nucleotide substitutions leading to V101A and V216A exist in various mouse strains, the nucleotide substitution leading to W252C exists in only tawny-colored mice. Thus this substitution is considered to be responsible for the expression of the tawny coat color. The frequency of the allele having this nucleotide substitution was 9.21% in the wild *M. m. molossinus* population inhabiting Sakai City, Osaka Prefecture, Japan, where the ancestral mice of the MSKR strain were captured.

Additional References

RELATED GEPHE

Related Genes

4 (Agouti, Agouti (ASIP), PMEL17, SLC45A2=MATP) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~10090~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

4 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~MC1R~/and+Taxon ID=~10090~/or+Gene Gephebase=~MC1R~/and+Taxon ID=~10090~#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS