

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000584	GepheID
	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	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
Mus musculus		Mus musculus	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Mus musculus^#gephebase-summary-title)		(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)		Mus () - (Rank: subgenus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
10090		10090	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 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	
MC1R		Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
		Synonyms	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		CH466525 (https://www.ncbi.nlm.nih.gov/nuccore/CH466525)	
		String	
9606.ENSPP00000451605			
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)			
		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>)

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
E92K	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	-	-	-

Pigmentation phenotypes of variant extension locus alleles result from point mutations that alter MSH receptor function. (1993) (https://pubmed.ncbi.nlm.nih.gov/8458079)	Main Reference
Robbins LS; Nadeau JH; Johnson KR; Kelly MA; Roselli-Reh fuss L; Baack E; Mountjoy KG; Cone RD	Authors
Coat colors in the chestnut horse, the yellow Labrador retriever, the red fox, and one type of yellow mouse are due to recessive alleles at the extension locus. Similarly, dominant alleles at this locus are often responsible for dark coat colors in mammals, such as the melanistic form of the leopard, Panthera pardus. We show here that the murine extension locus encodes the melanocyte-stimulating hormone (MSH) receptor. In mice, the recessive yellow allele (e) results from a frameshift that produces a prematurely terminated, nonfunctioning receptor. The sombre (Eso and Eso-3J) and tobacco darkening (Etob) alleles, which both have dominant melanizing effects, results from point mutations that produce hyperactive MSH receptors. The Eso-3J receptor is constitutively activated, while the Etob receptor remains hormone responsive and produces a greater activation of its effector, adenylyl cyclase, than does the wild-type allele.	Abstract
	Additional References

RELATED GEPHE

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 Genes
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)	Related Haplotypes

EXTERNAL LINKS

COMMENTS