

GEPHE SUMMARY

	Gephebase Gene		GepheID
MC1R (<a +mc1r+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+MC1R+"#gephebase-summary-title)		GP00000583	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)			
	Trait		
Coloration (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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 poschiavinus (tobacco mouse)			
	Ancestral State		
Data not curated			
	Taxonomic Status		
Interspecific (<a +interspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Interspecific+"#gephebase-summary-title)			

Taxon A		Taxon B	
	Latin Name		Latin Name
Mus musculus (<a +mus+musculus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Mus+musculus+"#gephebase-summary-title)		Mus poschiavinus (<a +mus+poschiavinus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Mus+poschiavinus+"#gephebase-summary-title)	
	Common Name		Common Name
house mouse		-	
	Synonyms		Synonyms
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; unclassified Mus	
	Parent		Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)		unclassified Mus () - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 941326)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)		100383 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 100383)	
	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		BAQ22650 (https://www.ncbi.nlm.nih.gov/nuccore/BAQ22650)	
	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>)

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title>)

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title>)

Nonsynonymous

S69L

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title>)

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

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>)

Robbins LS; Nadeau JH; Johnson KR; Kelly MA; Roselli-Reh fuss L; Baack E; Mountjoy KG; Cone RD

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 melanin 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 (Eto b) 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 Eto b receptor remains hormone responsive and produces a greater activation of its effector, adenylyl cyclase, than does the wild-type allele.

Main Reference

Authors

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/or+Taxon ID=~100383^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

4 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~MC1R^/and+Taxon ID=~10090^/or+Gene Gephebase=~MC1R^/and+Taxon ID=~100383^#gephebase-summary-title>)

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS