

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000575	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=^Morphology^#gephebase-summary-title)	Trait Category		
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration(coat)^#gephebase-summary-title)	Trait		
Cavia porcellus	Trait State in Taxon A		
Cavia porcellus	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=^Domesticated^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Cavia porcellus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Cavia+porcellus^#gephebase-summary-title)	Latin Name	Cavia porcellus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Cavia+porcellus^#gephebase-summary-title)	Latin Name
domestic guinea pig	Common Name	domestic guinea pig	Common Name
Cavia aperea porcellus; Cavia cobaya; domestic guinea pig; guinea pig; Cavia cobya	Synonyms	Cavia aperea porcellus; Cavia cobaya; domestic guinea pig; guinea pig; Cavia cobya	Synonyms
species	Rank	species	Rank
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; Hystricomorpha; Caviidae; Cavia	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; Hystricomorpha; Caviidae; Cavia	Lineage
Cavia (guinea pigs) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10140)	Parent	Cavia (guinea pigs) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10140)	Parent
10141 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10141)	NCBI Taxonomy ID	10141 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10141)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	0	GenebankID or UniProtKB
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	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			

(<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
Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)	
	Molecular Type
Gene Loss (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Gene Loss^#gephebase-summary-title)	
	Aberration Type
Complex Change (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Complex Change^#gephebase-summary-title)	
	Molecular Details of the Mutation
Deletion of part or all of gene	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)	
	Main Reference
The melanocortin receptors: agonists, antagonists, and the hormonal control of pigmentation. (1996) (https://pubmed.ncbi.nlm.nih.gov/8701084)	
	Authors
Cone RD; Lu D; Koppula S; Vage DI; Klunland H; Boston B; Chen W; Orth DN; Pouton C; Kesterson RA	
	Abstract
Molecular cloning experiments have led to the identification and characterization of a family of five receptors for the melanocortin (melanotropic and adrenocorticotropic) peptides. The first two members of the family cloned were the well-characterized melanocyte-stimulating hormone receptor (MSH-R) and adrenocorticotropin receptor (ACTH-R). The three new melanocortin receptors have been termed the MC3-R, MC4-R, and MC5-R, according to the order of their discovery, and little is known at this point concerning their function. Agouti and extension are two genetic loci known to control the amounts of eumelanin (brown-black) and pheomelanin (yellow-red) pigments. Chromosomal mapping demonstrated that the MSH-R, now termed MCI-R, mapped to extension. Extension was shown to encode the MCI-R, and mutations in the MCI-R are responsible for the different pigmentation phenotypes caused by this locus. Functional variants of the MCI-R, originally characterized in the mouse, have now also been identified in the guinea pig and cow. Dominant constitutive mutants of the MCI-R are responsible for causing dark black coat colors while recessive alleles result in yellow or red coat colors. Agouti, a secreted 108 amino acid peptide produced within the hair follicle, acts on follicular melanocytes to inhibit alpha-MSH-induced eumelanin production. Experiments demonstrate that agouti is a high-affinity antagonist, acting at the MCI-R to block alpha-MSH stimulation of adenylyl cyclase, the effector through which alpha-MSH induces eumelanin synthesis. The MCI-R is thus a unique bifunctionally controlled receptor, activated by alpha-MSH and antagonized by agouti, both contributing to the variability seen in mammalian coat colors. The variable tan and black coat color patterns seen in the German Shepherd, for example, can now be understood on the molecular level as the interaction of a number of extension and agouti alleles encoding variably functioning receptors and a differentially expressed antagonist of the receptor, respectively.	
	Additional References

RELATED GEPHE

	Related Genes
1 (tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^10141^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

