

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000593	GepheID
	Entry Status	Martin	Main curator
	Published		

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	
Panthera onca		Trait State in Taxon A	
Panthera onca - black		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Panthera onca (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Panthera+onca^#gephebase-summary-title)		Panthera onca (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Panthera+onca^#gephebase-summary-title)	
jaguar		jaguar	
jaguar		jaguar	
species		species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Pantherinae; Panthera		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Pantherinae; Panthera	
Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688)		Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688)	
9690 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9690)		9690 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9690)	
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
	Synonyms	AAO62414 (https://www.ncbi.nlm.nih.gov/nuccore/AAO62414)	GenebankID or UniProtKB
	String		
9606.ENSPO0000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000451605)			
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
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
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title)	
	Deletion Size
10-99 bp	
	Molecular Details of the Mutation
Deletion of aa 101-105 and L106T	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	
	Main Reference
Molecular genetics and evolution of melanism in the cat family. (2003) (https://pubmed.ncbi.nlm.nih.gov/12620197)	
	Authors
Eizirik E; Yuhki N; Johnson WE; Menotti-Raymond M; Hannah SS; O’Brien SJ	
	Abstract
Melanistic coat coloration occurs as a common polymorphism in 11 of 37 felid species and reaches high population frequency in some cases but never achieves complete fixation. To investigate the genetic basis, adaptive significance, and evolutionary history of melanistic variants in the Felidae, we mapped, cloned, and sequenced the cat homologs of two putative candidate genes for melanism (ASIP [agouti] and MC1R) and identified three independent deletions associated with dark coloration in three different felid species. Association and transmission analyses revealed that a 2 bp deletion in the ASIP gene specifies black coloration in domestic cats, and two different “in-frame” deletions in the MC1R gene are implicated in melanism in jaguars and jaguarundis. Melanistic individuals from five other felid species did not carry any of these mutations, implying that there are at least four independent genetic origins for melanism in the cat family. The inferred multiple origins and independent historical elevation in population frequency of felid melanistic mutations suggest the occurrence of adaptive evolution of this visible phenotype in a group of related free-ranging species.	
	Additional References

RELATED GEPHE

	Related Genes
No matches found.	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

