

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000581	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	
Herpailurus yaguarondi		Trait State in Taxon B	
Herpailurus yaguarondi		Ancestral State	
Taxon A		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)			
Taxon A		Taxon B	
Puma yagouarundi (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Puma+yagouarundi^#gephebase-summary-title)		Puma yagouarundi (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Puma+yagouarundi^#gephebase-summary-title)	
Common Name		Common Name	
jaguarundi		jaguarundi	
Synonyms		Synonyms	
Herpailurus yagouarundi; Herpailurus yaguarondi; Puma yagouarundii; jaguarundi; Puma yagouarundi (E. Geoffroy Saint-Hilaire, 1803)		Herpailurus yagouarundi; Herpailurus yaguarondi; Puma yagouarundii; jaguarundi; Puma yagouarundi (E. Geoffroy Saint-Hilaire, 1803)	
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; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Puma		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; Felinae; Puma	
Parent		Parent	
Puma () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=146712)		Puma () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=146712)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
1608482 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=1608482)		1608482 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=1608482)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q01726 (http://www.uniprot.org/uniprot/Q01726)
	String	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		AAO62416 (https://www.ncbi.nlm.nih.gov/nuccore/AAO62416)
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 : adenylyl 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 (<a +no`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No`#gephebase-summary-title)	
Coding (<a +coding`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding`#gephebase-summary-title)	Molecular Type
Deletion (<a +deletion`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+Deletion`#gephebase-summary-title)	Aberration Type
10-99 bp	Deletion Size
Deletion of aa 95-102	Molecular Details of the Mutation
Candidate Gene (<a +candidate="" gene`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Experimental Evidence=">https://www.gephebase.org/search-criteria?/and+Experimental Evidence="+Candidate Gene`#gephebase-summary-title)	Experimental Evidence
Molecular genetics and evolution of melanism in the cat family. (2003) (https://pubmed.ncbi.nlm.nih.gov/12620197)	Main Reference
Eizirik E; Yuhki N; Johnson WE; Menotti-Raymond M; Hannah SS; O'Brien SJ	Authors
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.	Abstract
	Additional References

RELATED GEPHE

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

EXTERNAL LINKS

