

# GEPHE SUMMARY

	Gephebase Gene	GephelD
MC1R ( <a href="https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%MC1R%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%MC1R%#gephebase-summary-title</a> )	GP00001733	Main curator
Published	Entry Status	Courtier

## PHENOTYPIC CHANGE

Trait Category		Trait	
Morphology ( <a href="https://www.gephebase.org/search-criteria/?and+Trait+Category=%Morphology%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Trait+Category=%Morphology%#gephebase-summary-title</a> )			
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria/?and+Trait=%Coloration+(coat)%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Trait=%Coloration+(coat)%#gephebase-summary-title</a> )		Trait State in Taxon A	
Geoffroy's cat		Trait State in Taxon B	
Geoffroy's cat - melanistic		Ancestral State	
Taxon A		Taxonomic Status	
Intraspecific ( <a href="https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=%Intraspecific%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=%Intraspecific%#gephebase-summary-title</a> )			
Taxon A	Latin Name	Taxon B	Latin Name
Leopardus geoffroyi ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Leopardus+geoffroyi%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Leopardus+geoffroyi%#gephebase-summary-title</a> )		Leopardus geoffroyi ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Leopardus+geoffroyi%#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Leopardus+geoffroyi%#gephebase-summary-title</a> )	
Geoffroy's cat	Common Name	Geoffroy's cat	Common Name
Felis geoffroyi; Oncifelis geoffroyi; Geoffroy's cat; Oncifelis geoffroyi (d'Orbigny & Gervais, 1844); Oncifelis geoffrovi	Synonyms	Felis geoffroyi; Oncifelis geoffroyi; Geoffroy's cat; Oncifelis geoffroyi (d'Orbigny & Gervais, 1844); Oncifelis geoffrovi	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; Laurasiatheria; Carnivora; Feliformia; Felidae; Felinae; Leopardus	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; Leopardus	Lineage
Leopardus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841</a> )	Parent	Leopardus () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841</a> )	Parent
46844 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46844">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46844</a> )	NCBI Taxonomy ID	46844 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46844">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46844</a> )	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	GenebankID or UniProtKB
9606.ENSP00000451605 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000451605">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000451605</a> )	String	
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities	
GO:0008528 : G protein-coupled peptide receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008528">https://www.ebi.ac.uk/QuickGO/term/GO:0008528</a> )	GO - Molecular Function	
GO:0004977 : melanocortin receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004977">https://www.ebi.ac.uk/QuickGO/term/GO:0004977</a> )		
GO:0004980 : melanocyte-stimulating hormone receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004980">https://www.ebi.ac.uk/QuickGO/term/GO:0004980</a> )		

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

All melanistic individuals were heterozygous for four variants that predicted three nonsynonymous substitutions, p.C125R, p.T177I, and p.G194S - In the laboratory mouse a Cys to Arg substitution at the site homologous to felid residue 125 causes constitutive activation of the receptor and the exact same change is thought to be responsible for melanism in the Alaska silver fox - therefore C125R is considered to be the likely causative alteration in the Geoffroy's cat.

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Arg	125

Main Reference

Recurrent evolution of melanism in South American felids. (2015) (<https://pubmed.ncbi.nlm.nih.gov/25695801>)

Authors

Schneider A; Henegar C; Day K; Absher D; Napolitano C; Silveira L; David VA; O'Brien SJ; Menotti-Raymond M; Barsh GS; Eizirik E

Abstract

Morphological variation in natural populations is a genomic test bed for studying the interface between molecular evolution and population genetics, but some of the most interesting questions involve non-model organisms that lack well annotated reference genomes. Many felid species exhibit polymorphism for melanism but the relative roles played by genetic drift, natural selection, and interspecies hybridization remain uncertain. We identify mutations of Agouti signaling protein (ASIP) or the Melanocortin 1 receptor (MC1R) as independent causes of melanism in three closely related South American species: the pampas cat (*Leopardus colocolo*), the kodkod (*Leopardus guigna*), and Geoffroy's cat (*Leopardus geoffroyi*). To assess population level variation in the regions surrounding the causative mutations we apply genomic resources from the domestic cat to carry out clone-based capture and targeted resequencing of 299 kb and 251 kb segments that contain ASIP and MC1R, respectively, from 54 individuals (13-21 per species), achieving enrichment of ~500-2500-fold and ~150x coverage. Our analysis points to unique evolutionary histories for each of the three species, with a strong selective sweep in the pampas cat, a distinctive but short melanism-specific haplotype in the Geoffroy's cat, and reduced nucleotide diversity for both ancestral and melanism-bearing chromosomes in the kodkod. These results reveal an important role for natural selection in a trait of longstanding interest to ecologists, geneticists, and the lay community, and provide a platform for comparative studies of morphological variation in other natural populations.

Additional References

Related Genes

No matches found.

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS