

GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Agouti">#gephebase-summary-title)	GP00001731	Main curator
Published	Entry Status	Courtier

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category="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		
pampas cat	Trait State in Taxon B		
pampas cat - melanistic	Ancestral State		
Taxon A	Taxonomic Status		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Intraspecific">#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Leopardus colocolo (#gephebase-summary-title)		Leopardus colocolo (#gephebase-summary-title)	
Colocolo	Common Name	Colocolo	Common Name
Felis colocolo; Lynchailurus colocolo; Oncifelis colocolo; Colocolo; pampas cat; Oncifelis colocolo (Molina, 1782)	Synonyms	Felis colocolo; Lynchailurus colocolo; Oncifelis colocolo; Colocolo; pampas cat; Oncifelis colocolo (Molina, 1782)	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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841)	Parent	Leopardus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 46841)	Parent
61406 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 61406)	NCBI Taxonomy ID	61406 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 61406)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Asip	Generic Gene Name	UniProtKB Mus musculus
As; ASP; A<y>; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function	
GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)		
GO:0031782 : type 4 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031782)		

GO - Biological Process

GO:0008343 : adult feeding behavior
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
 GO:0006091 : generation of precursor metabolites and energy
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
 GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
 GO:0009755 : hormone-mediated signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
 GO:0042438 : melanin biosynthetic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
 GO:0032438 : melanosome organization
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
 GO:0032402 : melanosome transport
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
 GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
 GO:0048023 : positive regulation of melanin biosynthetic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
 GO:0040030 : regulation of molecular function, epigenetic
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
 GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

Presumptive Null

Yes (<https://www.gephebase.org/search-criteria/?and+Presumptive+Null=%27Yes%27#gephebase-summary-title>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria/?and+Molecular+Type=%27Coding%27#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria/?and+Aberration+Type=%27SNP%27#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Arg to Cys substitution in the C-terminal region of ASIP (p.R120C) lies in the critical RFF loop (e.g. a triplet motif made of one arginine and two phenylalanine residues) required for binding to the MC1R

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria/?and+Experimental+Evidence=%27Candidate+Gene%27#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	120

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 GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA000201/61406/>