

## GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase="Agouti">#gephebase-summary-title)	GP00000063	
	Entry Status	Main curator
Published	Martin	

## PHENOTYPIC CHANGE

	Trait Category		
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category="Morphology">#gephebase-summary-title)	Trait		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration</a> (coat)#gephebase-summary-title)	Trait State in Taxon A		
Felis catus - non-melanic	Trait State in Taxon B		
Felis catus - melanic	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status="Domesticated">#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Felis catus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus">#gephebase-summary-title</a> )		Felis catus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Felis+catus">#gephebase-summary-title</a> )	
domestic cat	Common Name	domestic cat	Common Name
	Synonyms		Synonyms
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.		Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	
species	Rank	species	Rank
	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; Felis		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; Felis	
Felis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682</a> )	Parent	Felis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682</a> )	Parent
9685 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685</a> )	NCBI Taxonomy ID	9685 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685</a> )	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Asip	Q03288 ( <a href="http://www.uniprot.org/uniprot/Q03288">http://www.uniprot.org/uniprot/Q03288</a> )	
As; ASP; A<y>; ASIP; a		GenebankID or UniProtKB
10090.ENSMUSP00000029123 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123</a> )	AHN64806 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AHN64806">https://www.ncbi.nlm.nih.gov/nuccore/AHN64806</a> )	
	Sequence Similarities	
-		
	GO - Molecular Function	
GO:0031779 : melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031779">https://www.ebi.ac.uk/QuickGO/term/GO:0031779</a> )		
GO:0031781 : type 3 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031781">https://www.ebi.ac.uk/QuickGO/term/GO:0031781</a> )		
GO:0031782 : type 4 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031782">https://www.ebi.ac.uk/QuickGO/term/GO:0031782</a> )		

## 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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

Deletion of nt 123-124

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=%27Linkage+Mapping%27#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

6 (Kit (type III receptor protein-tyrosine kinase), MC1R, Melanophilin (MLPH), Taqpep, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=%279685%27/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

<https://omnia.org/OMIA000201/9685/>

