

## GEPHE SUMMARY

### Gephebase Gene

Agouti

### Entry Status

Published

### GepheID

GP00000063

### Main curator

Martin

## PHENOTYPIC CHANGE

### Trait Category

Morphology

### Trait

Coloration (coat)

### Trait State in Taxon A

Felis catus - non-melanic

### Trait State in Taxon B

Felis catus - melanic

### Ancestral State

Taxon A

### Taxonomic Status

Domesticated

### Taxon A

#### Latin Name

*Felis catus*

#### Common Name

domestic cat

#### Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758

#### Rank

species

#### 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

#### Parent

Felis () - (Rank: genus)

#### NCBI Taxonomy ID

9685

#### is Taxon A an Intraspecies?

No

### Taxon B

#### Latin Name

*Felis catus*

#### Common Name

domestic cat

#### Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758

#### Rank

species

#### 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

#### Parent

Felis () - (Rank: genus)

#### NCBI Taxonomy ID

9685

#### is Taxon B an Intraspecies?

No

## GENOTYPIC CHANGE

### Generic Gene Name

Asip

### Synonyms

As; ASP; A $\gamma$ ; ASIP; a

### String

10090.ENSMUSP00000029123

### Sequence Similarities

-

### GO - Molecular Function

GO:0031779 : melanocortin receptor binding

GO:0031781 : type 3 melanocortin receptor binding

GO:0031782 : type 4 melanocortin receptor binding

### GO - Biological Process

GO:0008343 : adult feeding behavior

GO:0006091 : generation of precursor metabolites and energy

GO:0071514 : genetic imprinting

GO:0009755 : hormone-mediated signaling pathway

GO:0042438 : melanin biosynthetic process

GO:0032438 : melanosome organization

### UniProtKB Mus musculus

Q03288

### GenebankID or UniProtKB

AHN64806

GO:0032402 : melanosome transport  
GO:0043473 : pigmentation  
GO:0048023 : positive regulation of melanin biosynthetic process  
GO:0040030 : regulation of molecular function, epigenetic

GO - Cellular Component  
GO:0005576 : extracellular region  
GO:0005623 : cell

Presumptive Null  
Yes

Molecular Type  
Coding

Aberration Type  
Deletion

Deletion Size  
1-9 bp

Molecular Details of the Mutation  
Deletion of nt 123-124

Experimental Evidence  
Linkage Mapping

Main Reference  
Molecular genetics and evolution of melanism in the cat family. (2003)

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))  
**Related Haplotypes**  
No matches found.

## COMMENTS

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