

## GEPHE SUMMARY

**Gephebase Gene**  
Agouti (ASIP)

**Entry Status**  
Published

**GepheID**  
GP00000061

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Coloration (coat)

**Trait State in Taxon A**  
Canis familiaris

**Trait State in Taxon B**  
German Shepherd Dog - Recessive black

**Ancestral State**  
Taxon A

**Taxonomic Status**  
Domesticated

### Taxon A

**Latin Name**  
*Canis lupus familiaris*

**Common Name**  
dog

**Synonyms**  
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

**Rank**  
subspecies

**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; Caniformia; Canidae; Canis; Canis lupus

**Parent**  
Canis lupus (gray wolf) - (Rank: species)

**NCBI Taxonomy ID**  
9615

**is Taxon A an Intraspecies?**  
No

### Taxon B

**Latin Name**  
*Canis lupus familiaris*

**Common Name**  
dog

**Synonyms**  
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

**Rank**  
subspecies

**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; Caniformia; Canidae; Canis; Canis lupus

**Parent**  
Canis lupus (gray wolf) - (Rank: species)

**NCBI Taxonomy ID**  
9615

**is Taxon B an Intraspecies?**  
No

## GENOTYPIC CHANGE

**Generic Gene Name**  
Asip

**Synonyms**  
As; ASP; A<sub>cy</sub>; 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

**UniProtKB Mus musculus**  
Q03288

**GenebankID or UniProtKB**  
AAW01462

GO:0032438 : melanosome organization  
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**

No

**Molecular Type**

Coding

**Aberration Type**

SNP

**SNP Coding Change**

Nonsynonymous

**Molecular Details of the Mutation**

R96C ; g.23393552C>T c.286C>T p.R96C

**Experimental Evidence**

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

**Main Reference**

Characterization of the dog Agouti gene and a nonagoutimutation in German Shepherd Dogs. (2004)

**Authors**

Kerns JA; Newton J; Berryere TG; Rubin EM; Cheng JF; Schmutz SM; Barsh GS

**Abstract**

The interaction between two genes, Agouti and Melanocortin-1 receptor ( Mc1r), produces diverse pigment patterns in mammals by regulating the type, amount, and distribution pattern of the two pigment types found in mammalian hair: eumelanin (brown/black) and pheomelanin (yellow/red). In domestic dogs ( Canis familiaris), there is a tremendous variation in coat color patterns between and within breeds; however, previous studies suggest that the molecular genetics of pigment-type switching in dogs may differ from that of other mammals. Here we report the identification and characterization of the Agouti gene from domestic dogs, predicted to encode a 131-amino-acid secreted protein 98% identical to the fox homolog, and which maps to chromosome CFA24 in a region of conserved linkage. Comparative analysis of the Doberman Pinscher Agouti cDNA, the fox cDNA, and 180 kb of Doberman Pinscher genomic DNA suggests that, as with laboratory mice, different pigment-type-switching patterns in the canine family are controlled by alternative usage of different promoters and untranslated first exons. A small survey of Labrador Retrievers, Greyhounds, Australian Shepherds, and German Shepherd Dogs did not uncover any polymorphisms, but we identified a single nucleotide variant in black German Shepherd Dogs predicted to cause an Arg-to-Cys substitution at codon 96, which is likely to account for recessive inheritance of a uniform black coat.

**Additional References**

Association of an Agouti allele with fawn or sable coat color in domestic dogs. (2005)

**RELATED GEPHE**

**Related Genes**

11 (MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103))

**Related Haplotypes**

2

**EXTERNAL LINKS**

**COMMENTS**

@Epistasis @Parallelism exact same mutation in alpaca <https://omia.org/OMIA000201/9615/>

