

## GEPHE SUMMARY

|   |                |              |
|---|----------------|--------------|
|   | Gephebase Gene | GephelD      |
| Agouti (ASIP) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title</a> ) | GP00000061     | Main curator |
| Published   | Entry Status   | Martin       |
|   |                |              |

## PHENOTYPIC CHANGE

|   | Trait Category              |   |                             |
|---|-----------------------------|---|-----------------------------|
| 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> )  | Trait                       |   |                             |
| 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      |   |                             |
| Canis familiaris  | Trait State in Taxon B      |   |                             |
| German Shepherd Dog - Recessive black   | Ancestral State             |   |                             |
| Taxon A   | Taxonomic Status            |   |                             |
| Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title</a> )  |                             |   |                             |
| Taxon A   | Latin Name                  | Taxon B   | Latin Name                  |
| Canis lupus familiaris<br>( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title</a> )   |                             | Canis lupus familiaris<br>( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title</a> )   |                             |
| dog   | Common Name                 | dog   | Common Name                 |
| Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758;<br>Canis lupus familiaris Linnaeus, 1758   | Synonyms                    | Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758;<br>Canis lupus familiaris Linnaeus, 1758   | Synonyms                    |
| subspecies  | Rank                        | subspecies  | Rank                        |
| cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;<br>Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;<br>Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;<br>Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus | Lineage                     | cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;<br>Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;<br>Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;<br>Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus | Lineage                     |
| Canis lupus (gray wolf) - (Rank: species)<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612</a> )  | Parent                      | Canis lupus (gray wolf) - (Rank: species)<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612</a> )  | Parent                      |
| 9615<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615</a> )   | NCBI Taxonomy ID            | 9615<br>( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615</a> )   | 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<br>( <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> ) | String                  | AAW01462 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AAW01462">https://www.ncbi.nlm.nih.gov/nuccore/AAW01462</a> ) |
| -   | Sequence Similarities   |  |
| GO:0031779 : melanocortin receptor binding<br>( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031779">https://www.ebi.ac.uk/QuickGO/term/GO:0031779</a> )   | GO - Molecular Function |  |
| GO:0031781 : type 3 melanocortin receptor binding<br>( <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<br>( <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

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^No))

Molecular Type

Coding ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Coding))

Aberration Type

SNP ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=^SNP))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

R96C ; g.23393552C&gt;T c.286C&gt;T p.R96C

Experimental Evidence

Candidate Gene ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=^Candidate+Gene))

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

Characterization of the dog Agouti gene and a nonagoutimutation in German Shepherd Dogs. (2004) (<https://pubmed.ncbi.nlm.nih.gov/15520882>)

Main Reference

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

Authors

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.

Abstract

Association of an Agouti allele with fawn or sable coat color in domestic dogs. (2005) (<https://pubmed.ncbi.nlm.nih.gov/15965787>)

Additional References

## RELATED GEPHE

12 (GPR22, 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)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

2 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^Agouti+\(ASIP\)^/and+Taxon+ID=^9615^/or+Gene+Gephebase=^Agouti+\(ASIP\)^/and+Taxon+ID=^9615^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=^Agouti+(ASIP)^/and+Taxon+ID=^9615^/or+Gene+Gephebase=^Agouti+(ASIP)^/and+Taxon+ID=^9615^#gephebase-summary-title))

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

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