

GEPHE SUMMARY

Gephebase Gene		GepheID
Agouti (ASIP) (<a +agouti+(asip)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase= "Agouti (ASIP)" #gephebase-summary-title)	GP00000060	
Entry Status		Main curator
Published		

PHENOTYPIC CHANGE

Trait Category			
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category= "Morphology" #gephebase-summary-title)		Trait	
Coloration (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait= "Coloration (coat)" #gephebase-summary-title)		Trait State in Taxon A	
Canis familiaris		Trait State in Taxon B	
Canis familiaris - 8 breeds with a black-and-tan or tricolor phenotype		Ancestral State	
Data not curated		Taxonomic Status	
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status= "Domesticated" #gephebase-summary-title)			
Taxon A		Taxon B	
Canis lupus familiaris (<a +canis+lupus+familiaris+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= "Canis lupus familiaris" #gephebase-summary-title)		Canis lupus familiaris (<a +canis+lupus+familiaris+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= "Canis lupus familiaris" #gephebase-summary-title)	
Common Name		Common Name	
dog		dog	
Synonyms		Synonyms	
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758		Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	
Rank		Rank	
subspecies		subspecies	
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; Caniformia; Canidae; Canis; Canis lupus		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		Parent	
Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)		Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)		9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Mus musculus
Asip	Synonyms	Q03288 (http://www.uniprot.org/uniprot/Q03288)
As; ASP; A<y>; ASIP; a	String	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000029123)		AAW01462 (https://www.ncbi.nlm.nih.gov/nuccore/AAW01462)
Sequence Similarities		
-		
GO - Molecular Function		
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)		
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: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 (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.244G>T + 248G>A ; p.A82S + p.R83H
A82S and R83H in Linkage Disequilibrium
a G-to-T transversion and a G-to-A transition in exon 4 at positions corresponding to residues 244 and 248 of the cDNA sequence
It is unknown if one or both substitutions alter Agouti protein function

Experimental Evidence

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

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

Main Reference

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

Authors

Berryere TG; Kerns JA; Barsh GS; Schmutz SM

Abstract

The type of pigment synthesized in mammalian hair, yellow-red pheomelanin or black-brown eumelanin, depends on the interaction between Agouti protein and the Melanocortin 1 receptor. Although the genetics of pigmentation is broadly conserved across most mammalian species, pigment type-switching in domestic dogs is unusual because a yellow-tan coat with variable amounts of dark hair is thought to be caused by an allele of the Agouti locus referred to as fawn or sable (a(y)). In a large survey covering thirty seven breeds, we identified an Agouti allele with two missense alterations, A82S and R83H, which was present (heterozygous or homozygous) in 41 dogs (22 breeds) with a fawn or sable coat, but was absent from 16 dogs (8 breeds) with a black-and-tan or tricolor phenotype. In an additional 33 dogs (14 breeds) with a eumelanic coat, 8 (German Shepherd Dogs, Groenendaels, Schipperkes, or Shetland Sheepdogs) were homozygous for a previously reported mutation, non-agouti R96C; the remainder are likely to have carried dominant black, which is independent of and epistatic to Agouti. This work resolves some of the complexity in dog coat color genetics and provides diagnostic opportunities and practical guidelines for breeders.

Additional References

RELATED GEPHE

Related Genes

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 Haplotypes

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))

EXTERNAL LINKS

COMMENTS

@Epistasis <https://omia.org/OMIA000201/9615/>