

GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (ASIP) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title)	GP00001336	Main curator
Published	Entry Status	Prigent

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait		
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)	Trait State in Taxon A		
Dog-wolf sable	Trait State in Taxon B		
Dog black-and-tan + saddle phenotypes	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
Canis lupus	Latin Name	Canis lupus familiaris	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)	
gray wolf	Common Name	dog	Common Name
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758;	Synonyms
	Rank	Canis lupus familiaris Linnaeus, 1758	Rank
species	Lineage	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		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	
Canis () - (Rank: genus)	Parent	Canis lupus (gray wolf) - (Rank: species)	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)	NCBI Taxonomy ID	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	NCBI Taxonomy ID
9612		9615	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	
No	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
	No		

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Asip	Q03288 (http://www.uniprot.org/uniprot/Q03288)	GenebankID or UniProtKB
As; ASP; A<y>; ASIP; a	Synonyms	
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	
-	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 - 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 (<https://www.gephebase.org/search-criteria/?and+Presumptive+Null=%No%#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

Insertion Size

100-999 bp

Molecular Details of the Mutation

A 239bp SINE insertion in reverse orientation in intron 1

Experimental Evidence

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

Main Reference

A SINE insertion causes the black-and-tan and saddle tan phenotypes in domestic dogs. (2011 Sep-Oct) (<https://pubmed.ncbi.nlm.nih.gov/21846741>)

Authors

Dreger DL; Schmutz SM

Abstract

Agouti Signaling Protein (ASIP) controls the localized expression of red and black pigment in the domestic dog through interaction with other genes, such as Melanocortin 1 Receptor and Beta-Defensin 103. Specific ASIP alleles are necessary for many of the coat color patterns, such as black-and-tan and saddle tan. Mutations in 2 ASIP alleles, a(y) and a, have previously been identified. Here, we characterize a mutation consisting of a short interspersed nuclear element (SINE) insertion in intron 1 of ASIP that allows for the differentiation of the a(w) wolf sable and a(t) black-and-tan alleles. The SINE insertion is present in dogs with the a(t) and a alleles but absent from dogs with the a(w) and a(y) alleles. Dogs with the saddle tan phenotype were all a(t)/a(t). Schnauzers were all a(w)/a(w). Genotypes of 201 dogs of 35 breeds suggest that there are only 4 ASIP alleles, as opposed to the 5 or 6 predicted in previous literature. These data demonstrate that the dominance hierarchy of ASIP is a(y) > a(w) > a(t) > a.

Additional References

RELATED GEPHE

Related Genes

13 (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), RALY (hnRNP associated with lethal yellow)) (<https://www.gephebase.org/search-criteria/?or+Taxon+ID=%9612%and+Trait=Coloration/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=%9612%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=%9612%or+Gene+Gephebase=%Agouti+(ASIP)%and+Taxon+ID=%9615%#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

this mutation might be involved in the solid hair color (versus banded hair color) and the black and tan phenotype. Dreger and Schmutz (2011) "characterize[d] a mutation consisting of a short interspersed nuclear element (SINE) insertion in intron 1 of ASIP that allows for the differentiation of the a(w) wolf sable and a(t) black-and-tan alleles. The SINE insertion is present in dogs with the a(t) and a alleles but absent from dogs with the a(w) and a(y) alleles. Dogs with the saddle tan phenotype were all a(t)/a(t). Schnauzers were all a(w)/a(w)." <https://omia.org/OMIA000201/9615/>

