

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)		GP00002373	
	Entry Status	Santos	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; dorso-ventral) (<a +coloration+(coat;+dorso-ventral)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat;+dorso-ventral)^#gephebase-summary-title)		
	Trait State in Taxon A	
agouti-coloured coat		
	Trait State in Taxon B	
black and tan coat colour		
	Ancestral State	
Taxon A		
	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	Latin Name	Taxon B	Latin Name
Oryctolagus cuniculus (<a +oryctolagus+cuniculus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryctolagus+cuniculus^#gephebase-summary-title)		Oryctolagus cuniculus (<a +oryctolagus+cuniculus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryctolagus+cuniculus^#gephebase-summary-title)	
	Common Name		Common Name
rabbit		rabbit	
	Synonyms		Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits		Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	
	Rank		Rank
species		species	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus	
	Parent		Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)		Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)		9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Asip		Q03288 (http://www.uniprot.org/uniprot/Q03288)	
	Synonyms		GenebankID or UniProtKB
As; ASP; A<y>; ASIP; a		()	
	String		
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)			
	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

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Insertion Size

10-100 kb

Molecular Details of the Mutation

"The structural variant represented an approximately 11 kb deletion (NC_013672.1:g.5,455,408_5,466,123del; Fig. 2). In rabbit, there are currently two ASIP transcript isoforms annotated (NCBI annotation release 102). The deletion removes the entire first 5â€²-untranslated exon of one of these transcripts (NM_001122939.1). (...) he deletion removed the transcription start site and the first untranslated exon of the presumably hair cycle-specific transcript isoform, suggesting that this is the most likely causative variant for the black and tan phenotype."

Experimental Evidence

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

Main Reference

A deletion spanning the promoter and first exon of the hair cycle-specific ASIP transcript isoform in black and tan rabbits. (2020) (https://pubmed.ncbi.nlm.nih.gov/31729778)

Authors

Letko A; Ammann B; Jagannathan V; Henkel J; Leuthard F; Schelling C; Carneiro M; DrÄ¼gemÄ¼ller C; Leeb T

Abstract

Black and tan animals have tan-coloured ventral body surfaces separated by sharp boundaries from black-coloured dorsal body surfaces. In the a mouse mutant, a retroviral 6Å kb insertion located in the hair cycle-specific promoter of the murine Asip gene encoding agouti signalling protein causes the black and tan phenotype. In rabbits, three ASIP alleles are thought to exist, including an a allele causing a black and tan coat colour that closely resembles the mouse black and tan phenotype. The goal of our study was to identify the functional genetic variant causing the rabbit a allele. We performed a WGS-based comparative analysis of the ASIP gene in one black and tan and three wt agouti-coloured rabbits. The analysis identified 75 a -associated variants including an 11Å kb deletion. The deletion is located in the region of the hair cycle-specific ASIP promoter and thus in a region homologous to the site of the retroviral insertion causing the a allele in mice. We observed perfect association of the genotypes at this deletion with the coat colour phenotype in 49 rabbits. The comparative analysis and the previous knowledge about the regulation of ASIP expression suggest that the 11Å kb deletion is the most likely causative variant for the black and tan phenotype in rabbits.

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Additional References

RELATED GEPHE

Related Genes

4 (MC1R, Melanophilin (MLPH), tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9986^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Agouti (ASIP)^/and+Taxon ID=^9986^/or+Gene Gephebase=^Agouti (ASIP)^/and+Taxon ID=^9986^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism

