

GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (ASIP) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Agouti (ASIP)^#gephebase-summary-title)	GP00002373	Main curator
Published	Entry Status	Santos

PHENOTYPIC CHANGE

	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait
Coloration (coat; dorso-ventral) (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

Taxon A	Latin Name	Taxon B	Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)		Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)	
rabbit	Common Name	rabbit	Common Name
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms	Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	Synonyms
species	Rank	species	Rank

Taxon A	Lineage	Taxon B	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	
Oryctolagus () - (Rank: genus)	Parent	Oryctolagus () - (Rank: genus)	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)	NCBI Taxonomy ID	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)	NCBI Taxonomy ID
9986		9986	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)	
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Mus musculus
Asip		
As; ASP; A<y>; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	0
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function	
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.gephbase.org/search-criteria?/and+Presumptive Null=%27Yes%27#gephbase-summary-title](https://www.gephbase.org/search-criteria?/and+Presumptive%20Null=%27Yes%27#gephbase-summary-title))

Molecular Type

Coding ([https://www.gephbase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephbase-summary-title](https://www.gephbase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephbase-summary-title))

Aberration Type

Insertion ([https://www.gephbase.org/search-criteria?/and+Aberration Type=%27Insertion%27#gephbase-summary-title](https://www.gephbase.org/search-criteria?/and+Aberration%20Type=%27Insertion%27#gephbase-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€“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.gephbase.org/search-criteria?/and+Experimental Evidence=%27Candidate Gene%27#gephbase-summary-title](https://www.gephbase.org/search-criteria?/and+Experimental%20Evidence=%27Candidate%20Gene%27#gephbase-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; Drâ¶gemâ¼ller 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 6Â 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 11Â 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 11Â kb deletion is the most likely causative variant for the black and tan phenotype in rabbits.

Â© 2019 Stichting International Foundation for Animal Genetics.

Additional References

RELATED GEPHE

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

Related Genes

1 ([https://www.gephbase.org/search-criteria?/or+Gene Gephebase=%27Agouti \(ASIP\)%27/and+Taxon ID=%9986^/or+Gene Gephebase=%27Agouti \(ASIP\)%27/and+Taxon ID=%9986^#gephbase-summary-title](https://www.gephbase.org/search-criteria?/or+Gene%20Gephebase=%27Agouti%20(ASIP)%27/and+Taxon%20ID=%9986^/or+Gene%20Gephebase=%27Agouti%20(ASIP)%27/and+Taxon%20ID=%9986^#gephbase-summary-title))

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Parallelism

