GEPHE SUMMARY

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

GepheID
GP00002373

Entry Status
Published

Main curator
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
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)

Common Name
rabbit

Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

Rank
species

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

Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryctolagus cuniculus^#gephebase-summary-title)

Common Name
rabbit

Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

Rank
species

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

Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Asip

Synonyms
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

UniProtKB Mus musculus
Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB
()

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; DrÃ¶gemÃ¼ller 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.

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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