GEPHE SUMMARY

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

GepheID
GP00001968

Entry Status
Published

Main curator
Martin

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
Oryctolagus cuniculus

Trait State in Taxon B
Oryctolagus cuniculus

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
CBJ17605 (https://www.ncbi.nlm.nih.gov/nuccore/CBJ17605)

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

1-9 bp

Molecular Details of the Mutation

an insertion in exon 2 of the black nonagouti allele = c.5_6insA mutation which causes a frameshift and a predicted truncated protein of only 21 amino acids

Experimental Evidence

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

Main Reference

Characterization of the rabbit agouti signaling protein (ASIP) gene: transcripts and phylogenetic analyses and identification of the causative mutation of the nonagouti black coat colour. (2010) (https://pubmed.ncbi.nlm.nih.gov/20004240)

Authors

Fontanesi L; Forestier L; Allain D; Scotti E; Beretti F; Deretz-Picoulet S; Pecchioli E; Vernesi C; Robinson TJ; Malaney JL; Russo V; Oulmouden A

Abstract

The agouti locus encodes the agouti signalling protein (ASIP) which is involved in determining the switch from eumelanin to pheomelanin synthesis in melanocytes. In the domestic rabbit (Oryctolagus cuniculus) early studies indicated three alleles at this locus: A, light-bellied agouti (wild type); a(t), black and tan; a, black nonagouti. We characterized the rabbit ASIP gene and identified the causative mutation (an insertion in exon 2) of the black nonagouti allele whose frequency was evaluated in 31 breeds. Phylogenetic analysis of ASIP sequences from Oryctolagus and 9 other species of the family Leporidae placed Oryctolagus as sister species to Pentalagus and Bunolagus. Transcription analysis in wild type agouti rabbits revealed the presence of two major transcripts with different 5′-untranslated regions having ventral or dorsal skin specific expression. ASIP gene transcripts were also detected in all examined rabbit tissues distinguishing the rabbit expression pattern from what was observed in wild type mice.

Additional References

Multiple Conserved Elements Structuring Inverted Repeats in the Mammalian Coat Color-Related Gene Asip. (2019) (https://pubmed.ncbi.nlm.nih.gov/31116535)

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

https://omia.org/OMIA000201/9986/