GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00002386

Main curator

Santos

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

yellow agouti (A) phenotype

Trait State in Taxon B

black non-agouti phenotype

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

Mus musculus molossinus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus molossinus^#gephebase-summary-title)

Common Name

Japanese wild mouse

Synonyms

Mus molossinus; Japanese wild mouse; Mus musculus molosinus

Rank

subspecies

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; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus; Mus musculus

Parent

Mus musculus (house mouse) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)

NCBI Taxonomy ID

57486 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 57486)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Mus musculus molossinus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus molossinus^#gephebase-summary-title)

Common Name

Japanese wild mouse

Synonyms

Mus molossinus; Japanese wild mouse; Mus musculus molosinus

Rank

subspecies

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; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus; Mus musculus

Parent

Mus musculus (house mouse) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)

NCBI Taxonomy ID

57486 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 57486)

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)

UniProtKB Mus musculus

Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB

()

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

1-10 kb

Molecular Details of the Mutation

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Experimental Evidence

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

Main Reference

Nested retrotransposition in the East Asian mouse genome causes the classical nonagouti mutation. (2019) (https://pubmed.ncbi.nlm.nih.gov/31396563)

Authors

Tanave A; Imai Y; Koide T

Abstract

Black coat color (nonagouti) is a widespread classical mutation in laboratory mouse strains. The intronic insertion of endogenous retrovirus VL30 in the nonagouti (a) allele of agouti gene was previously reported as the cause of the nonagouti phenotype. Here, we report agouti mouse strains from East Asia that carry the VL30 insertion, indicating that VL30 alone does not cause the nonagouti phenotype. We find that a rare type of endogenous retrovirus, Î²4, was integrated into the VL30 region at the a allele through nested retrotransposition, causing abnormal splicing. Targeted complete deletion of the Î²4 element restores agouti gene expression and agouti coat color, whereas deletion of Î²4 except for a single long terminal repeat results in black-and-tan coat color. Phylogenetic analyses show that the a allele and the Î²4 retrovirus originated from an East Asian mouse lineage most likely related to Japanese fancy mice. These findings reveal the causal mechanism and historic origin of the classical nonagouti mutation.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS