GEPHE SUMMARY

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

GepheID
GP00001335

Entry Status
Published

Main curator
Prigent

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
Donkey

Trait State in Taxon B
Donkey - no light points pattern (in Normand & miniature donkeys)

Ancestral State
Taxon A

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

Taxon A

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

Common Name
ass

Synonyms
ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Laurasiatheria; Perissodactyla; Equidae; Equus; Asinus

Parent
Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID
9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

is Taxon A an Infraspecies?
Yes

Taxon A Description
Donkey

Taxon B

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

Common Name
ass

Synonyms
ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Laurasiatheria; Perissodactyla; Equidae; Equus; Asinus

Parent
Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID
9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Donkey - no light points pattern (in Normand & miniature donkeys)

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

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

GenebankID or UniProtKB
()

(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

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.349T>C p.Cys117Arg

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Arg`	117

Main Reference

A missense mutation in the agouti signaling protein gene (ASIP) is associated with the no light points coat phenotype in donkeys. (2015) (https://pubmed.ncbi.nlm.nih.gov/25887951)

Authors

Abitbol M; Legrand R; Tiret L

Abstract

Seven donkey breeds are recognized by the French studbook and are characterized by a black, bay or grey coat colour including light cream-to-white points (LP). Occasionally, Normand bay donkeys give birth to dark foals that lack LP and display the no light points (NLP) pattern. This pattern is more frequent and officially recognized in American miniature donkeys. The LP (or pangare) phenotype resembles that of the light bellied agouti pattern in mouse, while the NLP pattern resembles that of the mammalian recessive black phenotype; both phenotypes are associated with the agouti signaling protein gene (ASIP).

We used a panel of 127 donkeys to identify a recessive missense c.349 T > C variant in ASIP that was shown to be in complete association with the NLP phenotype. This variant results in a cysteine to arginine substitution at position 117 in the ASIP protein. This cysteine is highly-conserved among vertebrate ASIP proteins and was previously shown by mutagenesis experiments to lie within a functional site. Altogether, our results strongly support that the identified mutation is causative of the NLP phenotype.

Thus, we propose to name the c.[349 T > C] allele in donkeys, the a(nlp) allele, which enlarges the panel of coat colour alleles in donkeys and ASIP recessive loss-of-function alleles in animals.

Additional References

RELATED GEPHE

Related Genes

3 (Kit (type III receptor protein-tyrosine kinase), MC1R, tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9793^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

mutation is recessive; https://omia.org/OMIA000201/9793/