

GEPHE SUMMARY

<p>Agouti (ASIP) (<a +agouti+(asip)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Agouti (ASIP)^#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001335</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (<a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category="Morphology^#gephebase-summary-title)</p> <p>Coloration (coat) (<a +coloration+(coat)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Coloration (coat)^#gephebase-summary-title)</p> <p>Donkey</p> <p>Donkey - no light points pattern (in Normand & miniature donkeys)</p> <p>Taxon A</p> <p>Domesticated (<a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated^#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Equus asinus (<a +equus+asinus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Equus asinus^#gephebase-summary-title)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508)</p> <p>9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793)</p> <p>is Taxon A an Intraspecies?</p> <p>Yes</p> <p>Donkey</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>	<p>Equus asinus (<a +equus+asinus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Equus asinus^#gephebase-summary-title)</p> <p>ass</p> <p>ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey</p> <p>species</p> <p>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</p> <p>Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35508)</p> <p>9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9793)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Donkey - no light points pattern (in Normand & miniature donkeys)</p>	<p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
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GENOTYPIC CHANGE

<p>Asip</p> <p>As; ASP; A<y>; ASIP; a</p> <p>10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)</p> <p>-</p> <p>GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)</p> <p>GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)</p> <p>GO:0031782 : type 4 melanocortin receptor binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>Q03288 (http://www.uniprot.org/uniprot/Q03288)</p> <p>0</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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(<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>)

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

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

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

Nonsynonymous SNP Coding Change

c.349T>C p.Cys117Arg Molecular Details of the Mutation

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

	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/>