

GEPHE SUMMARY

<p>Gephebase Gene</p> <p>Agouti (ASIP) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Agouti+(ASIP)^#gephebase-summary-title)</p> <p>Published</p>	<p>GP00001325</p> <p>Entry Status</p> <p>Prigent</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title)</p> <p>Domestic sheep - dominant white phenotype in Australian Merino breed</p> <p>Domestic sheep - recessive "self-color black" phenotype in Australian Merino breed</p> <p>Taxon A</p> <p>Domesticated (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>
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<p>Taxon A</p> <p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon A an Intraspecies?</p> <p>Yes</p> <p>Domestic sheep - dominant white phenotype in Australian Merino breed</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 A an Intraspecies?</p> <p>Taxon A Description</p>
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<p>Taxon B</p> <p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Domestic sheep - recessive "self-color black" phenotype in Australian Merino breed</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>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>
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<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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GO:0031782 : type 4 melanocortin receptor binding
(<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](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](https://www.gephebase.org/search-criteria?/and+Molecular+Type+^Cis-regulatory^#gephebase-summary-title))

Aberration Type

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

Molecular Details of the Mutation

unidentified promoter silencing

Experimental Evidence

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

Main Reference

A gene duplication affecting expression of the ovine ASIP gene is responsible for white and black sheep. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18493018>)

Authors

Norris BJ; Whan VA

Abstract

Agouti signaling protein (ASIP) functions to regulate pigmentation in mice, while its role in many other animals and in humans has not been fully determined. In this study, we identify a 190-kb tandem duplication encompassing the ovine ASIP and AHCY coding regions and the ITCH promoter region as the genetic cause of white coat color of dominant white/tan (A(Wt)) agouti sheep. The duplication 5' breakpoint is located upstream of the ASIP coding sequence. Ubiquitous expression of a second copy of the ASIP coding sequence regulated by a duplicated copy of the nearby ITCH promoter causes the white sheep phenotype. A single copy ASIP gene with a silenced ASIP promoter occurs in recessive black sheep. In contrast, a single copy functional wild-type (A(+)) ASIP is responsible for the ancient Barbary sheep coat color phenotype. The gene duplication was facilitated by homologous recombination between two non-LTR SINE sequences flanking the duplicated segment. This is the first sheep trait attributable to gene duplication and shows nonallelic homologous recombination and gene conversion events at the ovine ASIP locus could have an important role in the evolution of sheep pigmentation.

Additional References

RELATED GEPHE

Related Genes

2 (MC1R, tyrosinase-related protein 1 (TYRP1)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=^9940^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID+^9940^/and+Trait+Coloration/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

2 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Agouti \(ASIP\)^/and+Taxon ID=^9940^/or+Gene Gephebase=^Agouti \(ASIP\)^/and+Taxon ID=^9940^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase+^Agouti+(ASIP)^/and+Taxon+ID+^9940^/or+Gene+Gephebase+^Agouti+(ASIP)^/and+Taxon+ID+^9940^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

The mutation is recessive and probably induces a loss-of-function

