

GEPHE SUMMARY

Gephebase Gene GepheID  
 Agouti (ASIP) ([https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Agouti+\(ASIP\)^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Agouti+(ASIP)^#gephebase-summary-title)) GP00001324  
 Entry Status Prigent  
 Published Main curator

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](https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title))  
 Trait State in Taxon A  
 Domestic sheep - ancient breed "Barbary" with wild-type pale belly phenotype  
 Trait State in Taxon B  
 Domestic sheep - Romanov and Texel breeds with dominant white allele  
 Ancestral State  
 Taxon A  
 Taxonomic Status  
 Domesticated (<https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title>)

Taxon A	Latin Name
Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title</a> )	Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title</a> )
Common Name	Common Name
sheep	sheep
Synonyms	Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758
Rank	Rank
species	species
Lineage	Lineage
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	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
Parent	Parent
Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )	Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )
NCBI Taxonomy ID	NCBI Taxonomy ID
9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )	9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )
is Taxon A an Intraspecies?	is Taxon B an Intraspecies?
Yes	Yes
Taxon A Description	Taxon B Description
Domestic sheep - ancient breed "Barbary" with wild-type pale belly phenotype	Domestic sheep - Romanov and Texel breeds with dominant white allele

Taxon B	Latin Name
Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title</a> )	Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title</a> )
Common Name	Common Name
sheep	sheep
Synonyms	Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758
Rank	Rank
species	species
Lineage	Lineage
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	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
Parent	Parent
Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )	Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )
NCBI Taxonomy ID	NCBI Taxonomy ID
9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )	9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )
is Taxon B an Intraspecies?	is Taxon B an Intraspecies?
Yes	Yes
Taxon B Description	Taxon B Description
Domestic sheep - Romanov and Texel breeds with dominant white allele	Domestic sheep - Romanov and Texel breeds with dominant white allele

GENOTYPIC CHANGE

Generic Gene Name UniProtKB Mus musculus  
 Asip Q03288 (<http://www.uniprot.org/uniprot/Q03288>)  
 Synonyms GenebankID or UniProtKB  
 As; ASP; A<y>; ASIP; a 0  
 String  
 10090.ENSMUSP00000029123  
 ([http://string-db.org/newstring.cgi/show\\_network\\_section.pl?identifier=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

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

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

Molecular Type

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

Aberration Type

100-1000 kb

Insertion Size

-190kb gene duplication causing dominant white phenotype. Duplication was facilitated by homologous recombination between two non-LTR SINE sequences flanking the duplicated segment ; possible involvement of gene conversion.

Molecular Details of the Mutation

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

Experimental Evidence

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>)

Main Reference

Norris BJ; Whan VA

Authors

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.

Abstract

Epistatic Interaction of the Melanocortin 1 Receptor and Agouti Signaling Protein Genes Modulates Wool Color in the Brazilian Creole Sheep. (2016)  
(<https://pubmed.ncbi.nlm.nih.gov/27288530>)

Additional References

RELATED GEPHE

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>)

Related Genes

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))

Related Haplotypes

EXTERNAL LINKS

COMMENTS

Dominance : dominant allele = duplication <https://omia.org/OMIA000201/9940/> @TEPpossibly

