

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="MC1R"#gephebase-summary-title)	Gephebase Gene	GP00001338	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category="Morphology"#gephebase-summary-title)	Trait Category		
Coloration (coat) (<a coloration"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Coloration (coat)#gephebase-summary-title)	Trait		
Antarctic fur seal-dark colored	Trait State in Taxon A		
Antarctic fur seal-cream colored	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Intraspecific"#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Arctocephalus gazella (<a arctocephalus"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="Arctocephalus gazella"#gephebase-summary-title)	Latin Name	Arctocephalus gazella (<a arctocephalus"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="Arctocephalus gazella"#gephebase-summary-title)	Latin Name
antarctic fur seal	Common Name	antarctic fur seal	Common Name
antarctic fur seal	Synonyms	antarctic fur seal	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Otariidae; Arctocephalus	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Otariidae; Arctocephalus	Lineage
Arctocephalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=29083)	Parent	Arctocephalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=29083)	Parent
37190 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=37190)	NCBI Taxonomy ID	37190 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=37190)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	()	GenebankID or UniProtKB
9606.ENSP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	GO - Molecular Function		
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)			
GO:0004980 : melanocyte-stimulating hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004980)			
GO:0031625 : ubiquitin protein ligase binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031625>)

GO - Biological Process

- GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
- GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
- GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
- GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
- GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
- GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
- GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)
- GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)
- GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)
- GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger (<https://www.ebi.ac.uk/QuickGO/term/GO:0007187>)
- GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
- GO:0010739 : positive regulation of protein kinase A signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010739>)
- GO:0090037 : positive regulation of protein kinase C signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090037>)
- GO:0009650 : UV protection (<https://www.ebi.ac.uk/QuickGO/term/GO:0009650>)
- GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

GO - Cellular Component

- GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
- GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null+No+g#gepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No+g#gepbase-summary-title))

Presumptive Null

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type+Coding+g#gepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type+Coding+g#gepbase-summary-title))

Molecular Type

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

Aberration Type

Nonsynonymous

SNP Coding Change

c.C>T p.Ser291Phe

Molecular Details of the Mutation

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

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

the mutation is recessive and probably induces a loss of function