GEPHE SUMMARY

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

GepheID
GP00001356

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait #1

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
Sheep ; Soay sheep on St Kilda ; wild type dark upper body and pale belly

Trait State in Taxon B
Sheep ; Soay sheep on St Kilda ; uniformly coloured

Trait #2

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

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

Trait State in Taxon A
-

Trait State in Taxon B
-

Ancestral State
Taxon A

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

Taxon A

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

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon A an Infraspecies?
Yes

Taxon A Description
Sheep ; Soay sheep on St Kilda ; wild type dark upper body and pale belly

Taxon B

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

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Sheep ; Soay sheep on St Kilda ; uniformly coloured

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 (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)

UniProtKB Mus musculus

Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB

()

Mutation #1

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

deletion of 5 bp - g.100-104delAGGAA in exon2 - frame-shift mutation that introduces a premature stop codon at amino acid position 64 resulting in a mature peptide that lacks the functionally important cysteine-signalling domain.

Experimental Evidence

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

Main Reference

The genetic basis of recessive self-colour pattern in a wild sheep population. (2010) (https://pubmed.ncbi.nlm.nih.gov/19672282)

Authors

Gratten J; Pilkington JG; Brown EA; Beraldi D; Pemberton JM; Slate J

Abstract

Bridging the genotype-phenotype gap for traits of ecological and evolutionary importance in natural populations can provide a novel insight into the origin and maintenance of variation. Here, we identify the gene and putative causal mutations underlying a recessive colour pattern phenotype ('self' or uniform colour) in a wild population of primitive Soay sheep. We targeted the agouti signalling protein (ASIP) gene, a positional candidate based on previous study that mapped the Coat pattern locus to a presumptive region on chromosome 13. We found evidence for three recessive mutations, including two functional changes in the coding sequence and a putative third cis-regulatory mutation that inactivates the promoter. These mutations define up to five haplotypes in Soays, which collectively explained the coat pattern in all but one member of a complex multi-generational pedigree containing 621 genotyped individuals. The functional mutations are in strong linkage disequilibrium in the study population, and are identical to those known to underlie the self phenotype in domestic sheep. This is indicative of a recent (and simultaneous) origin in Soay sheep, possibly as a consequence of past interbreeding with modern domestic breeds. This is only the second study in which ASIP has been linked to variation in pigmentation in a natural population. Knowledge of the genetic basis of self-colour pattern in Soay sheep, and the recognition that several mutations are segregating in the population, will aid future studies investigating the role of selection in the maintenance of the polymorphism.

Additional References

Selection and microevolution of coat pattern are cryptic in a wild population of sheep. (2012) (https://pubmed.ncbi.nlm.nih.gov/22432567)

Mutation #2

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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

g.5172T-A in exon 4 - non-synonymous mutation that is predicted to cause a cysteine to serine substitution at codon 126 within the signalling domain of the protein

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	5172
Amino-acid	Cys	Ser	126

Main Reference

The genetic basis of recessive self-colour pattern in a wild sheep population. (2010) (https://pubmed.ncbi.nlm.nih.gov/19672282)

Authors

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Abstract

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Additional References

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RELATED GEPHE

Related Genes

2 (MC1R, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9940^/and+Trait=Coloration/or+Taxon ID=^9940^/and+Trait=Lifespan/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)

EXTERNAL LINKS

COMMENTS

The three mutations define up to five haplotypes in the population- Recessive ; homozygous with reduction in juvenile survival - evidence of natural selection @Fitness - A putative cis-regulatory mutation that inactivates the promoter is proposed to explain mismatch between coding sequence and coat phenotype and ASIP expression levels. However this third mutation may not map to the same locus.