GEPHE SUMMARY

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

GepheID
GP00002381

Entry Status
Published

Main curator
Santos

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)

Trait State in Taxon A
Valais Blacknecked goat ; black forequarters and white hindquarters

Trait State in Taxon B
white Valais Capra Sempione

Ancestral State
Taxon A

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

Taxon A

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

Common Name
goat

Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
goat

Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra

Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9922)

NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9925)

is Taxon B an Infraspecies?
No

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

UniProtKB Mus musculus
Q03288 (http://www.uniprot.org/uniprot/Q03288)

GenebankID or UniProtKB
()

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)

Molecular Type

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

Aberration Type

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

Insertion Size

100-1000 kb

Molecular Details of the Mutation

"The white Capra Sempione carried one copy of the AWt allele, which is commonly found in Saanen and Appenzell goats. AWt involves a triplication of ~155 kb comprising the coding sequence of the ASIP, AHCY and ITCH genes (Fontanesi et al. 2009; Henkel et al. 2019). Closer inspection of the short-read alignments revealed that the sequenced Capra Sempione had acquired additional smaller structural alterations compared to the original AWt allele, which comprised a duplication of 1053 bp inserted into a 10â€‰194 bp deletion of the original AWt CNV allele (Supplementary Figure S1)."

Experimental Evidence

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

Main Reference

Introgression of ASIP and TYRP1 Alleles Explains Coat Color Variation in Valais Goats. (2021) (https://pubmed.ncbi.nlm.nih.gov/34050662)

Authors

Henkel J; Dubacher A; Bangerter E; Herren U; Ammann P; DrÃ¶gemÃ¼ller C; Flury C; Leeb T

Abstract

The Valais Blackneck goat is a Swiss goat breed with a characteristic coat color phenotype. Before the revision of the breed standard in 1938, 4 different color varieties of Valais goats were known. Besides Blackneck animals resembling the modern breed standard, the brown and white Copperneck goat, the white Capra Sempione, and the greyish GrÃ¼enochte comprised the historic Valais goats. The brown pigmentation of Copperneck goats had previously been traced back to an introgression of a mutant TYRP1 allele from Toggenburg goats. In the present study, we identified additional introgression events of distinct ASIP alleles causing the remaining 2 rare coat color patterns within the Valais Blackneck goat breed. We identified the introgression of the AWt allele from Appenzell or Saanen goats in white Capra Sempione goats. Similarly, introgression of the Apc allele from Peacock goats resulted in the greyish GrÃ¼enochte phenotype. These results demonstrate past hybridization events between breeds that are separated today. A perfect genotype-phenotype association in 393 Valais goats supported the causality of the genotyped variants for the different coat color phenotypes. Our study gives insights into the introgression of functionally relevant copy number variant (CNV) alleles controlling pigmentation between goat breeds with strikingly different coat color patterns.

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Additional References

RELATED GEPHE

Related Genes

3 (EDNRA, MC1R, tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9925^/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=^9925^/or+Gene Gephebase=^Agouti (ASIP)^/and+Taxon ID=^9925^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Introgression The duplicated region also contains two other genes AHCY and ITCH, which do not appear to be involved in pigmentation: AHCY is an enzyme involved in L-homocysteine biosynthesis and ITCH is an E3 ubiquitin-protein ligase.