

GEPHE SUMMARY

	Gephebase Gene		GepheID
Agouti (ASIP) (<a +agouti+(asip)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Agouti+(ASIP)^#gephebase-summary-title)		GP00002381	
	Entry Status	Santos	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (<a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title)			
	Trait		
Coloration (coat) (<a +coloration+(coat)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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 (<a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated^#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Capra hircus (<a +capra+hircus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Capra+hircus^#gephebase-summary-title)		Capra hircus (<a +capra+hircus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Capra+hircus^#gephebase-summary-title)	
	Common Name		Common Name
goat		goat	
	Synonyms		Synonyms
Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat		Capra aegagrus hircus; goat; domestic goat; goats; Carpa hircus; South African angora goat	
	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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Capra		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		Parent
Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)		Capra () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)		9925 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

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		()	
	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)

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

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 1053 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

Introgession 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; DrÄqgemÄ¼ller 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 GrÄ¼enochte 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 GrÄ¼enochte 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

Ä 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.

