

## 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</a> )		GP00002380	
	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</a> )			
	Trait		
Coloration ( <a +coloration^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration^#gephebase-summary-title</a> )			
	Trait State in Taxon A		
solid coloured coat			
	Trait State in Taxon B		
depigmentation phenotype of non-classical swiss markings			
	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</a> )			
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</a> )		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</a> )	
	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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922</a> )		Capra () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9922</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9925 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925</a> )		9925 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9925</a> )	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Asip		Q03288 ( <a href="http://www.uniprot.org/uniprot/Q03288">http://www.uniprot.org/uniprot/Q03288</a> )	
	Synonyms		GenebankID or UniProtKB
As; ASP; A<y>; ASIP; a		()	
	String		
10090.ENSMUSP00000029123 ( <a href="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</a> )			
	Sequence Similarities		
-			
	GO - Molecular Function		
GO:0031779 : melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031779">https://www.ebi.ac.uk/QuickGO/term/GO:0031779</a> )			
GO:0031781 : type 3 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031781">https://www.ebi.ac.uk/QuickGO/term/GO:0031781</a> )			
GO:0031782 : type 4 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031782">https://www.ebi.ac.uk/QuickGO/term/GO:0031782</a> )			
	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

10-100 kb Insertion Size

"We confirmed that a genomic region harboring the ASIP gene is a major locus affecting the coat color phenotype of Swiss markings in goats. Although the molecular genetic mechanisms remain unsolved, the 13,420-bp duplication upstream of ASIP is a necessary but not sufficient condition for this phenotype in goats. Moreover, the variations in the copy number of the duplication across different goat breeds do not lead to phenotypic heterogeneity."  
Molecular Details of the Mutation

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

A 13.42-kb tandem duplication at the ASIP locus is strongly associated with the depigmentation phenotype of non-classic Swiss markings in goats. (2022)  
 (https://pubmed.ncbi.nlm.nih.gov/35698044) Main Reference

Guo J; Sun X; Mao A; Liu H; Zhan S; Li L; Zhong T; Wang L; Cao J; Liu GE; Zhang H Authors

The pigmentation phenotype diversity is rich in domestic goats, and identification of the genetic loci affecting coat color in goats has long been of interest. Via the detections of selection signatures, a duplication upstream ASIP was previously reported to be a variant affecting the Swiss markings depigmentation phenotype in goats. Abstract

We conducted a genome-wide association study using whole-genome sequencing (WGS) data to identify the genetic loci and causal variants affecting the pigmentation phenotype in 65 Jintang black (JT) goats (i.e., 48 solid black vs. 17 non-classic Swiss markings). Although a single association peak harboring the ASIP gene at 52,619,845-72,176,538 bp on chromosome 13 was obtained using a linear mixed model approach, all the SNPs and indels in this region were excluded as causal variants for the pigmentation phenotype. We then found that all 17 individuals with non-classic Swiss markings carried a 13,420-bp duplication (CH13:63,129,198-63,142,617 bp) nearly 101 kb upstream of ASIP, and this variant was strongly associated (P=1.48e-10) with the coat color in the 65 JT goats. The copy numbers obtained from the WGS data also showed that the duplication was present in all 53 goats from three European breeds with Swiss markings and absent in 45 of 51 non-Swiss markings goats from four other breeds and 21 Bezoars, which was further validated in 314 samples from seven populations based on PCR amplification. The copy numbers of the duplication vary in different goat breeds with Swiss markings, indicating a threshold effect instead of a dose-response effect at the molecular level. Furthermore, breakpoint flanking repeat analysis revealed that the duplication was likely to be a result of the Bov-B-mediated nonallelic homologous recombination.

We confirmed that a genomic region harboring the ASIP gene is a major locus affecting the coat color phenotype of Swiss markings in goats. Although the molecular genetic mechanisms remain unsolved, the 13,420-bp duplication upstream of ASIP is a necessary but not sufficient condition for this phenotype in goats. Moreover, the variations in the copy number of the duplication across different goat breeds do not lead to phenotypic heterogeneity.

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Additional References

RELATED GEPHE

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 Genes

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) Related Haplotypes

EXTERNAL LINKS

