GEPHE SUMMARY

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

GepheID
GP00002380

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

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 (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

10-100 kb

Molecular Details of the Mutation

"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."

Experimental Evidence

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

Main Reference

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)

Authors

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

Abstract

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.

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 52,619,845-72,176,538â€‰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 (CHI13:63,129,198-63,142,617â€‰bp) nearly 101â€‰kb upstream of ASIP, and this variant was strongly associated (Pâ€‰=â€‰1.48â€‰Ã—â€‰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.

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