

GEPHE SUMMARY

	Gephebase Gene	GephelD
Agouti (ASIP) (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%Agouti+(ASIP)%#gephebase-summary-title)	GP00002370	Main curator
Published	Entry Status	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
dark gray to black coat	Trait State in Taxon B
white coat	Ancestral State
Taxon A	Taxonomic Status

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

Taxon A	Latin Name	Taxon B	Latin Name
Bubalus bubalis (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Bubalus+bubalis%#gephebase-summary-title)	Common Name	Bubalus bubalis (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Bubalus+bubalis%#gephebase-summary-title)	Common Name
water buffalo	Synonyms	water buffalo	Synonyms
Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus		Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus	
species	Rank	species	Rank
	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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bubalus		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; Bovinae; Bubalus	
Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9918)	Parent	Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9918)	Parent
89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 89462)	NCBI Taxonomy ID	89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 89462)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Asip	Generic Gene Name	UniProtKB Mus musculus
As; ASP; A<y>; ASIP; a	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000029123 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)	String	0
-	Sequence Similarities	
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)	GO - Molecular Function	
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>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria/?and+Presumptive+Null=%No%#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria/?and+Molecular+Type=%Cis-regulatory%#gephebase-summary-title>)

Aberration Type

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

Insertion Size

1-10 kb

Molecular Details of the Mutation

"2,809-bp-long LINE-1 insertion in the ASIP (agouti signaling protein) gene is the causative mutation for the white coat phenotype in swamp buffalo (*Bubalus bubalis*). This LINE-1 insertion (3' truncated and containing only 5' UTR) functions as a strong proximal promoter that leads to a 10-fold increase in the transcription of ASIP in white buffalo skin. The 165 bp of 5' UTR transcribed from the LINE-1 is spliced into the first coding exon of ASIP, resulting in a chimeric transcript. The increased expression of ASIP prevents melanocyte maturation, leading to the absence of pigment in white buffalo skin and hairs. Phylogenetic analyses indicate that the white buffalo-specific ASIP allele originated from a recent genetic transposition event in swamp buffalo. Interestingly, as a similar LINE-1 insertion has been identified in the cattle ASIP gene, we discuss the convergent mechanism of coat color evolution in the Bovini tribe."

Experimental Evidence

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

Main Reference

Genomic Analysis Revealed a Convergent Evolution of LINE-1 in Coat Color: A Case Study in Water Buffaloes (*Bubalus bubalis*). (2021) (<https://pubmed.ncbi.nlm.nih.gov/33212507/>)

Authors

Liang D; Zhao P; Si J; Fang L; Pairo-Castineira E; Hu X; Xu Q; Hou Y; Gong Y; Liang Z; Tian B; Mao H; Yindee M; Faruque MO; Kongvongxay S; Khamphoumee S; Liu GE; Wu DD; Barker JSF; Han J; Zhang Y

Abstract

Visible pigmentation phenotypes can be used to explore the regulation of gene expression and the evolution of coat color patterns in animals. Here, we performed whole-genome and RNA sequencing and applied genome-wide association study, comparative population genomics and biological experiments to show that the 2,809-bp-long LINE-1 insertion in the ASIP (agouti signaling protein) gene is the causative mutation for the white coat phenotype in swamp buffalo (*Bubalus bubalis*). This LINE-1 insertion (3' truncated and containing only 5' UTR) functions as a strong proximal promoter that leads to a 10-fold increase in the transcription of ASIP in white buffalo skin. The 165 bp of 5' UTR transcribed from the LINE-1 is spliced into the first coding exon of ASIP, resulting in a chimeric transcript. The increased expression of ASIP prevents melanocyte maturation, leading to the absence of pigment in white buffalo skin and hairs. Phylogenetic analyses indicate that the white buffalo-specific ASIP allele originated from a recent genetic transposition event in swamp buffalo. Interestingly, as a similar LINE-1 insertion has been identified in the cattle ASIP gene, we discuss the convergent mechanism of coat color evolution in the Bovini tribe.

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Additional References

RELATED GEPHE

Related Genes

2 (Microphthalmia-associated transcription factor, tyrosinase (TYR)) (<https://www.gephebase.org/search-criteria/?or+TaxonID=%89462%and+Trait=Colorationand+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism

