GEPHE SUMMARY

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

GepheID
GP00002370

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
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
Bubalus bubalis (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bubalus bubalis^#gephebase-summary-title)

Common Name
water buffalo

Synonyms
Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bubalus

Parent
Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9918)

NCBI Taxonomy ID
89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 89462)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
water buffalo

Synonyms
Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bubalus

Parent
Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9918)

NCBI Taxonomy ID
89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 89462)

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)

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

GenebankID or UniProtKB
()

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 (3â€² truncated and containing only 5â€² UTR) functions as a strong proximal promoter that leads to a 10-fold increase in the transcription of ASIP in white buffalo skin. The 165â€‰bp of 5â€² 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 165â€‰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.

Additional References

RELATED GEPHE

Related Genes

2 (Microphtalmia-associated transcription factor, tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^89462^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism