

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

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

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

1-10 kb Insertion Size

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 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."

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

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) Main Reference

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 Authors

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.

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Additional References

RELATED GEPHE

2 (Microphthalmia-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 Genes

No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Parallelism

