GEPHE SUMMARY

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

GepheID
GP00002336

Entry Status
Published

Main curator
Martin

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
WT

Trait State in Taxon B
Recessive black

Ancestral State
Taxon A

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

Taxon A

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

Common Name
alpaca

Synonyms
Lama guanicoe pacos; Lama pacos; alpaca

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; Tylopoda; Camelidae; Vicugna

Parent
Vicugna () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30539)

NCBI Taxonomy ID
30538 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30538)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
alpaca

Synonyms
Lama guanicoe pacos; Lama pacos; alpaca

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; Tylopoda; Camelidae; Vicugna

Parent
Vicugna () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30539)

NCBI Taxonomy ID
30538 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30538)

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
AAW01462 (https://www.ncbi.nlm.nih.gov/nuccore/AAW01462)

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

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

p.R98C likely affecting conformation in a cysteine-rich domain

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	98

Main Reference

The alpaca agouti gene: genomic locus, transcripts and causative mutations of eumelanic and pheomelanic coat color. (2013) (https://pubmed.ncbi.nlm.nih.gov/23558248)

Authors

Chandramohan B; Renieri C; La Manna V; La Terza A

Abstract

The agouti gene encodes the agouti signaling protein (ASIP) which regulates pheomelanin and eumelanin synthesis in mammals. To investigate the role of agouti in coat color variation of alpaca, we characterized the agouti gene and identified three mutations potentially involved with the determinism of eumelanic and pheomelanic phenotypes. The exon-4 hosts the mutations g.3836C>T, g.3896G>A and g.3866_3923del57. Further analysis of these mutations revealed two genotypes for black animals. The reverse transcription analysis of mRNA purified from skin biopsies of alpaca revealed the presence of three transcripts with different 5′ untranslated regions (UTRs) and color specific expression. The white specific transcript, possibly originating from a duplication event (intra-chromosomal recombination) of the agouti gene is characterise by a 5′UTR containing 142bp of the NCOA6 gene sequence. Furthermore, the relative level expression analysis of mRNA demonstrates that the agouti gene has up-regulated expression in white skin, suggesting a pleiotropic effect of agouti in the white phenotype. Our findings refine the structure of the agouti locus and transcripts and provide additional information in order to understand the role of agouti in the pigmentation of alpaca.

Additional References

RELATED GEPHE

Related Genes

1 (EDN3) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^30538^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Epistasis @Parallelism exact same mutation in dogs