GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00000065

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

Panthera pardus

Trait State in Taxon B

Panthera pardus - melanic

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

leopard

Synonyms

Leo pardus; leopard

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; Carnivora; Feliformia; Felidae; Pantherinae; Panthera

Parent

Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9688)

NCBI Taxonomy ID

9691 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9691)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

leopard

Synonyms

Leo pardus; leopard

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; Carnivora; Feliformia; Felidae; Pantherinae; Panthera

Parent

Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9688)

NCBI Taxonomy ID

9691 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9691)

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

AGB56420 (https://www.ncbi.nlm.nih.gov/nuccore/AGB56420)

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

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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

Nonsense

Molecular Details of the Mutation

nonsense mutation located in exon 4 (C333A) predicted to introduce a stop codon at amino acid position 111

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	TGC	TGA	333
Amino-acid	Cys	STP	111

Main Reference

How the leopard hides its spots: ASIP mutations and melanism in wild cats. (2012) (https://pubmed.ncbi.nlm.nih.gov/23251368)

Authors

Schneider A; David VA; Johnson WE; O'Brien SJ; Barsh GS; Menotti-Raymond M; Eizirik E

Abstract

The occurrence of melanism (darkening of the background coloration) is documented in 13 felid species, in some cases reaching high frequencies at the population level. Recent analyses have indicated that it arose multiple times in the Felidae, with three different species exhibiting unique mutations associated with this trait. The causative mutations in the remaining species have so far not been identified, precluding a broader assessment of the evolutionary dynamics of melanism in the Felidae. Among these, the leopard (Panthera pardus) is a particularly important target for research, given the iconic status of the 'black panther' and the extremely high frequency of melanism observed in some Asian populations. Another felid species from the same region, the Asian golden cat (Pardofelis temminckii), also exhibits frequent records of melanism in some areas. We have sequenced the coding region of the Agouti Signaling Protein (ASIP) gene in multiple leopard and Asian golden cat individuals, and identified distinct mutations strongly associated with melanism in each of them. The single nucleotide polymorphism (SNP) detected among the P. pardus individuals was caused by a nonsense mutation predicted to completely ablate ASIP function. A different SNP was identified in P. temminckii, causing a predicted amino acid change that should also induce loss of function. Our results reveal two additional cases of species-specific mutations implicated in melanism in the Felidae, and indicate that ASIP mutations may play an important role in naturally-occurring coloration polymorphism.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA000201/9691/