

GEPHE SUMMARY

|                                                                                                                                                                                                                      |  |              |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|--------------|
| Gephebase Gene                                                                                                                                                                                                       |  | GepheID      |
| Agouti ( <a href="https://www.gephebase.org/search-criteria?/and+GeneGephebase=^Agouti^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+GeneGephebase=^Agouti^#gephebase-summary-title</a> ) |  | GP00000066   |
| Entry Status                                                                                                                                                                                                         |  | Main curator |
| Published                                                                                                                                                                                                            |  |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                           |  |                                                                                                                                                                                                                                                                                                                                           |  |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|
| Trait Category                                                                                                                                                                                                                                                                                                                            |  |                                                                                                                                                                                                                                                                                                                                           |  |
| Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title</a> )                                                                                                          |  | Trait                                                                                                                                                                                                                                                                                                                                     |  |
| Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)^#gephebase-summary-title</a> )                                                                                                       |  | Trait State in Taxon A                                                                                                                                                                                                                                                                                                                    |  |
| Pardofelis temminckii - non-melanic                                                                                                                                                                                                                                                                                                       |  | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                    |  |
| Pardofelis temminckii - melanic                                                                                                                                                                                                                                                                                                           |  | Ancestral State                                                                                                                                                                                                                                                                                                                           |  |
| Data not curated                                                                                                                                                                                                                                                                                                                          |  | Taxonomic Status                                                                                                                                                                                                                                                                                                                          |  |
| Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title</a> )                                                                                             |  |                                                                                                                                                                                                                                                                                                                                           |  |
| Taxon A                                                                                                                                                                                                                                                                                                                                   |  | Taxon B                                                                                                                                                                                                                                                                                                                                   |  |
| Latin Name                                                                                                                                                                                                                                                                                                                                |  | Latin Name                                                                                                                                                                                                                                                                                                                                |  |
| Catopuma temminckii ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Catopumatemminckii^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Catopumatemminckii^#gephebase-summary-title</a> )                                                                       |  | Catopuma temminckii ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Catopumatemminckii^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Catopumatemminckii^#gephebase-summary-title</a> )                                                                       |  |
| Common Name                                                                                                                                                                                                                                                                                                                               |  | Common Name                                                                                                                                                                                                                                                                                                                               |  |
| Asiatic golden cat                                                                                                                                                                                                                                                                                                                        |  | Asiatic golden cat                                                                                                                                                                                                                                                                                                                        |  |
| Synonyms                                                                                                                                                                                                                                                                                                                                  |  | Synonyms                                                                                                                                                                                                                                                                                                                                  |  |
| Felis temminckii; Pardofelis temminckii; Profelis temmincki; Profelis temminckii; Asiatic golden cat; Catopuma temminckii (Vigors & Horsfield, 1827)                                                                                                                                                                                      |  | Felis temminckii; Pardofelis temminckii; Profelis temmincki; Profelis temminckii; Asiatic golden cat; Catopuma temminckii (Vigors & Horsfield, 1827)                                                                                                                                                                                      |  |
| Rank                                                                                                                                                                                                                                                                                                                                      |  | Rank                                                                                                                                                                                                                                                                                                                                      |  |
| species                                                                                                                                                                                                                                                                                                                                   |  | species                                                                                                                                                                                                                                                                                                                                   |  |
| 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; Carnivora; Feliformia; Felidae; Felinae; Catopuma |  | 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; Felinae; Catopuma |  |
| Parent                                                                                                                                                                                                                                                                                                                                    |  | Parent                                                                                                                                                                                                                                                                                                                                    |  |
| Catopuma () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61453">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61453</a> )                                                                                                                                                         |  | Catopuma () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61453">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61453</a> )                                                                                                                                                         |  |
| NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                          |  | NCBI Taxonomy ID                                                                                                                                                                                                                                                                                                                          |  |
| 61455 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61455">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61455</a> )                                                                                                                                                                               |  | 61455 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61455">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=61455</a> )                                                                                                                                                                               |  |
| is Taxon A an Intraspecies?                                                                                                                                                                                                                                                                                                               |  | is Taxon B an Intraspecies?                                                                                                                                                                                                                                                                                                               |  |
| No                                                                                                                                                                                                                                                                                                                                        |  | No                                                                                                                                                                                                                                                                                                                                        |  |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                                          |  |                                                                                                                        |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--|------------------------------------------------------------------------------------------------------------------------|
| Generic Gene Name                                                                                                                                                                                                                        |  | UniProtKB Mus musculus                                                                                                 |
| Asip                                                                                                                                                                                                                                     |  | Q03288 ( <a href="http://www.uniprot.org/uniprot/Q03288">http://www.uniprot.org/uniprot/Q03288</a> )                   |
| Synonyms                                                                                                                                                                                                                                 |  | GenebankID or UniProtKB                                                                                                |
| As; ASP; A<y>; ASIP; a                                                                                                                                                                                                                   |  | AGB56420 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AGB56420">https://www.ncbi.nlm.nih.gov/nuccore/AGB56420</a> ) |
| String                                                                                                                                                                                                                                   |  |                                                                                                                        |
| 10090.ENSMUSP00000029123 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123</a> ) |  |                                                                                                                        |
| Sequence Similarities                                                                                                                                                                                                                    |  |                                                                                                                        |
| -                                                                                                                                                                                                                                        |  |                                                                                                                        |
| GO - Molecular Function                                                                                                                                                                                                                  |  |                                                                                                                        |
| GO:0031779 : melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031779">https://www.ebi.ac.uk/QuickGO/term/GO:0031779</a> )                                                                                 |  |                                                                                                                        |
| GO:0031781 : type 3 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031781">https://www.ebi.ac.uk/QuickGO/term/GO:0031781</a> )                                                                          |  |                                                                                                                        |
| GO:0031782 : type 4 melanocortin receptor binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031782">https://www.ebi.ac.uk/QuickGO/term/GO:0031782</a> )                                                                          |  |                                                                                                                        |

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

C128W (384C>G)

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 | Cys     | Trp     | 128      |

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

