GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00001105

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

Acinonyx jubatus - spotted

Trait State in Taxon B

Acinonyx jubatus - King

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

cheetah

Synonyms

cheetah

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; Acinonychinae; Acinonyx

Parent

Acinonyx () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32535)

NCBI Taxonomy ID

32536 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32536)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

cheetah

Synonyms

cheetah

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; Acinonychinae; Acinonyx

Parent

Acinonyx () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32535)

NCBI Taxonomy ID

32536 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32536)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Acinonyx jubatus - King

GENOTYPIC CHANGE

Generic Gene Name

Lvrn

Synonyms

Aqpep

String

10090.ENSMUSP00000025358 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000025358)

Sequence Similarities

Belongs to the peptidase M1 family.

GO - Molecular Function

GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)
GO:0042277 : peptide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042277)
GO:0070006 : metalloaminopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0070006)

GO - Biological Process

UniProtKB Mus musculus

Q2KHK3 (http://www.uniprot.org/uniprot/Q2KHK3)

GenebankID or UniProtKB

()

GO:0006508 : proteolysis (https://www.ebi.ac.uk/QuickGO/term/GO:0006508)
GO:0043171 : peptide catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043171)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)

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

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

1bp insertion resulting in frameshift p.N977Kfs110

Experimental Evidence

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

Main Reference

Specifying and sustaining pigmentation patterns in domestic and wild cats. (2012) (https://pubmed.ncbi.nlm.nih.gov/22997338)

Authors

Kaelin CB; Xu X; Hong LZ; David VA; McGowan KA; Schmidt-KÃ¼ntzel A; Roelke ME; Pino J; Pontius J; Cooper GM; Manuel H; Swanson WF; Marker L; Harper CK; van Dyk A; Yue B; Mullikin JC; Warren WC; Eizirik E; Kos L; O'Brien SJ; Barsh GS; Menotti-Raymond M

Abstract

Color markings among felid species display both a remarkable diversity and a common underlying periodicity. A similar range of patterns in domestic cats suggests a conserved mechanism whose appearance can be altered by selection. We identified the gene responsible for tabby pattern variation in domestic cats as Transmembrane aminopeptidase Q (Taqpep), which encodes a membrane-bound metalloprotease. Analyzing 31 other felid species, we identified Taqpep as the cause of the rare king cheetah phenotype, in which spots coalesce into blotches and stripes. Histologic, genomic expression, and transgenic mouse studies indicate that paracrine expression of Endothelin3 (Edn3) coordinates localized color differences. We propose a two-stage model in which Taqpep helps to establish a periodic pre-pattern during skin development that is later implemented by differential expression of Edn3.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA001721/32536/