

GEPHE SUMMARY

<p>Taqpep (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~Taqpep~#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001108</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
--	---	---------------------------------	------------------------------------

PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)</p> <p>Felis catus - western breeds</p> <p>Felis catus - western breeds</p> <p>Data not curated</p> <p>Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific~#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Felis catus</p> <p>domestic cat</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>species</p> <p>Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)</p> <p>9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)</p> <p>No</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Felis catus</p> <p>domestic cat</p> <p>Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.</p> <p>species</p> <p>Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)</p> <p>9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p>
---	---	---	---	---	---

GENOTYPIC CHANGE

<p>Lvrn</p> <p>Aqpep</p> <p>10090.ENSMUSP00000025358 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000025358)</p> <p>Belongs to the peptidase M1 family.</p> <p>GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)</p> <p>GO:0042277 : peptide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042277)</p> <p>GO:0070006 : metalloaminopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0070006)</p> <p>GO:0006508 : proteolysis (https://www.ebi.ac.uk/QuickGO/term/GO:0006508)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>Q2KHK3 (http://www.uniprot.org/uniprot/Q2KHK3)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
---	---	--

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

W841stop

Experimental Evidence

Linkage Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Linkage+Mapping~#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Trp	STP	841

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-Käntzel 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

6 (Agouti, Kit (type III receptor protein-tyrosine kinase), MC1R, Melanophilin (MLPH), tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9685~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9685~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

2 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Taqpep~/and+Taxon ID=~9685~/or+Gene Gephebase=~Taqpep~/and+Taxon ID=~9685~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~Taqpep~/and+Taxon+ID=~9685~/or+Gene+Gephebase=~Taqpep~/and+Taxon+ID=~9685~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@AllelicSeries <https://omia.org/OMIA001429/9685/>