## GEPHE SUMMARY

Gephebase Gene

tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001153

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

Felis catus

Trait State in Taxon B

Felis catus - cinnamon

Ancestral State

Taxon A

Taxonomic Status

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

### Taxon A

Latin Name

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

Common Name

domestic cat

Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.

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; Felinae; Felis

Parent

Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682 )

NCBI Taxonomy ID

9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685 )

is Taxon A an Infraspecies?

No

### Taxon B

Latin Name

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

Common Name

domestic cat

Synonyms

Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.

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; Felinae; Felis

Parent

Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682 )

NCBI Taxonomy ID

9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685 )

is Taxon B an Infraspecies?

Yes

Taxon B Description

Felis catus - cinnamon

## GENOTYPIC CHANGE

Generic Gene Name

Tyrp1

Synonyms

b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1

String

10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000006151 )

Sequence Similarities

Belongs to the tyrosinase family.

GO - Molecular Function

GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046982 : protein heterodimerization activity

UniProtKB Mus musculus

P07147 (http://www.uniprot.org/uniprot/P07147)

GenebankID or UniProtKB

AAY87043 (https://www.ncbi.nlm.nih.gov/nuccore/AAY87043)

(https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0004497 : monooxygenase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004497)

GO - Biological Process

GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)
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:0006583 : melanin biosynthetic process from tyrosine
(https://www.ebi.ac.uk/QuickGO/term/GO:0006583)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043438 : acetoacetic acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043438)
GO:0006582 : melanin metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006582)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030669)
GO:0010008 : endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0010008)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

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

premature UAG stop codon at position 100 of TYRP1; AAG>TAG c.298C>T p.R100*

Experimental Evidence

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

| | Taxon A | Taxon B | Position |
|---|---|---|---|
| Codon | AAG | TAG | 298 |
| Amino-acid | Arg | STP | 100 |

Main Reference

Tyrosinase and tyrosinase related protein 1 alleles specify domestic cat coat color phenotypes of the albino and brown loci. (2005 Jul-Aug) (https://pubmed.ncbi.nlm.nih.gov/15858157)

Authors

Schmidt-KÃ¼ntzel A; Eizirik E; O'Brien SJ; Menotti-Raymond M

Abstract

The genes encoding enzymes of the tyrosinase family are strong candidates for coat color variation in mammals. To investigate their influence in domestic cat coat color, we determined the complete nucleotide coding sequence of the domestic cat genes tyrosinase (TYR)--a plausible candidate gene for the albino (C) locus, and tyrosinase related protein 1 (TYRP1)--a candidate gene for the brown (B) locus. Sequence variants between individuals exhibiting variation in pigmentation were submitted to association studies. In TYR, two nonsynonymous substitutions encoding TYR-G301R and TYR-G227W were associated with the siamese and burmese phenotypes of the albino locus, respectively. TYRP1 was mapped on chromosome D4 within 5 cM of a highly polymorphic microsatellite, previously found to be fixed in a cat breed selected for the chocolate (b) allele of the B locus, which reinforced TYRP1 as a candidate gene for the B locus in the domestic cat. Two DNA polymorphisms, one leading to a TYRP1-A3G substitution in the signal peptide and another to an in-frame insertion TYRP1-421ins17/18 caused by a donor splice site mutation in intron 6, were associated with the chocolate (b) allele. A premature UAG stop codon at position 100 of TYRP1 was associated with a second allele of the B locus, cinnamon (b(l)). The results provide very strong evidence that the specific nucleotide variants of feline TYR (chromosome D1) are causative of the siamese (c(s)) and burmese (c(b)) alleles of the albino locus, as well as nucleotide variants of TYRP1 (chromosome D4) as specifying the chocolate (b) and cinnamon (b(l)) alleles of the B locus.

Additional References

Chocolate coated cats: TYRP1 mutations for brown color in domestic cats. (2005) (https://pubmed.ncbi.nlm.nih.gov/16104383)

## RELATED GEPHE

Related Genes

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

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9685^/or+Gene Gephebase=^tyrosinase-related protein 1 (TYRP1)^/and+Taxon ID=^9685^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA001249/9685/