

GEPHE SUMMARY

	Gephebase Gene	GephelD
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)	GP00001153	Main curator
	Entry Status	
Published	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	Taxon B	Latin Name
Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Felis catus^#gephebase-summary-title)		Felis catus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Felis catus^#gephebase-summary-title)	
domestic cat	Common Name	domestic cat	Common Name
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	Synonyms	Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	Synonyms
species	Rank	species	Rank
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	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	Lineage
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)	Parent	Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)	Parent
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)	NCBI Taxonomy ID	9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
		Felis catus - cinnamon	Taxon B Description

GENOTYPIC CHANGE

	Generic Gene Name		
Tyrp1			UniProtKB Mus musculus
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1	Synonyms	P07147 (http://www.uniprot.org/uniprot/P07147)	GenebankID or UniProtKB
10090.ENSMUSP00000006151 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)	String	AAY87043 (https://www.ncbi.nlm.nih.gov/nuccore/AAY87043)	
Belongs to the tyrosinase family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		
GO:0046982 : protein heterodimerization activity			

(<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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Yes))

Molecular Type

Coding ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Coding))

Aberration Type

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

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

	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-Kuntzel 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)) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+TaxonID=^9685^/and+Trait=Coloration/and+groupHaplotypes=true))

Related Haplotypes

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

EXTERNAL LINKS

COMMENTS

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