

tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)	Gephebase Gene	GP00001154	GepheID
Published	Entry Status	Courtier	Main curator

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)		Trait Category	
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)		Trait	
Felis catus		Trait State in Taxon A	
Felis catus - chocolate		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Latin Name		Latin Name	
https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Felis+catus~#gephebase-summary-title		https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Felis+catus~#gephebase-summary-title	
Common Name		Common Name	
domestic cat		domestic cat	
Synonyms		Synonyms	
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.		Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	
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; Felis		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		Parent	
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)		Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9682)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)		9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9685)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Felis catus - chocolate	

	Generic Gene Name	UniProtKB Mus musculus
Tyrp1		P07147 (http://www.uniprot.org/uniprot/P07147)
	Synonyms	GenebankID or UniProtKB
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-1		AA Y87043 (https://www.ncbi.nlm.nih.gov/nucore/AA Y87043)
	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		

(<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

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

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

Molecular Details of the Mutation

Two mutations associated with the chocolate (b) allele - 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 -exact causing change unknown

Experimental Evidence

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

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

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](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

