## GEPHE SUMMARY

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

GepheID
GP00001143

Entry Status
Published

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 - siamese

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?
No

## GENOTYPIC CHANGE

Generic Gene Name
Tyr

Synonyms
c; Oca1; skc35; albino

String
10090.ENSMUSP00000004770 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770 )

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:0005507 : copper ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005507)
GO:0004503 : monophenol monooxygenase activity

UniProtKB Mus musculus
P11344 (http://www.uniprot.org/uniprot/P11344)

GenebankID or UniProtKB
AAY32311 (https://www.ncbi.nlm.nih.gov/nuccore/AAY32311)

(https://www.ebi.ac.uk/QuickGO/term/GO:0004503)

GO - Biological Process

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
GO:0033280 : response to vitamin D (https://www.ebi.ac.uk/QuickGO/term/GO:0033280)
GO:0051591 : response to cAMP (https://www.ebi.ac.uk/QuickGO/term/GO:0051591)
GO:0009411 : response to UV (https://www.ebi.ac.uk/QuickGO/term/GO:0009411)
GO:0048538 : thymus development (https://www.ebi.ac.uk/QuickGO/term/GO:0048538)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0048471 : perinuclear region of cytoplasm
(https://www.ebi.ac.uk/QuickGO/term/GO:0048471)
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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c. 940G>A p.G302R

Experimental Evidence

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

| | Taxon A | Taxon B | Position |
|---|---|---|---|
| Codon | - | - | - |
| Amino-acid | Gly | Arg | 302 |

Main Reference

Tyrosinase mutations associated with Siamese and Burmese patterns in the domestic cat (Felis catus). (2005) (https://pubmed.ncbi.nlm.nih.gov/15771720)

Authors

Lyons LA; Imes DL; Rah HC; Grahn RA

Abstract

The Siamese cat has a highly recognized coat colour phenotype that expresses pigment at the extremities of the body, such as the ears, tail and paws. This temperature-sensitive colouration causes a 'mask' on the face and the phenotype is commonly referred to as 'pointed'. Burmese is an allelic variant that is less temperature-sensitive, producing more pigment throughout the torso than Siamese. Tyrosinase (TYR) mutations have been suspected to cause these phenotypes because mutations in TYR are associated with similar phenotypes in other species. Linkage and synteny mapping in the cat has indirectly supported TYR as the causative gene for these feline phenotypes. TYR mutations associated with Siamese and Burmese phenotypes are described herein. Over 200 cats were analysed, representing 12 breeds as well as randomly bred cats. The SNP associated with the Siamese phenotype is an exon 2 G > A transition changing glycine to arginine (G302R). The SNP associated with the Burmese phenotype is an exon 1 G > T transversion changing glycine to tryptophan (G227W). The G302R mutation segregated concordantly within a pedigree of Himalayan (pointed) Persians. All cats that had 'pointed' or the Burmese coat colour phenotype were homozygous for the corresponding mutations, respectively, suggesting that these phenotypes are a result of the identified mutations or unidentified mutations that are in linkage disequilibrium. Because the same mutations were identified in different breeds with similar phenotypes, the mutations are likely to be identical by descent rather than multiple mutation events occurring at the same site.

Additional References

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)

## RELATED GEPHE

Related Genes

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

Related Haplotypes

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

## EXTERNAL LINKS

COMMENTS

@Parallelism @AllelicSeries https://omia.org/OMIA000202/9685/