

GEPHE SUMMARY

Gephebase Gene
tyrosinase (TYR)

Entry Status
Published

GepheID
GP00002312

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Siamese cat

Trait State in Taxon B
Complete albinism

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Felis catus

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)

NCBI Taxonomy ID
9685

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Felis catus

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)

NCBI Taxonomy ID
9685

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Tyr

Synonyms
c; Ocat; skc35; albino

String
10090.ENSMUSP00000004770

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity
GO:0046982 : protein heterodimerization activity
GO:0005507 : copper ion binding
GO:0004503 : monophenol monooxygenase activity

GO - Biological Process
GO:0042438 : melanin biosynthetic process
GO:0043473 : pigmentation
GO:0008283 : cell proliferation
GO:0033280 : response to vitamin D

UniProtKB Mus musculus
P11344

GenebankID or UniProtKB
AA32311

GO:0051591 : response to cAMP
GO:0009411 : response to UV
GO:0048538 : thymus development

GO - Cellular Component

GO:0016021 : integral component of membrane
GO:0005737 : cytoplasm
GO:0005829 : cytosol
GO:0005634 : nucleus
GO:0043231 : intracellular membrane-bounded organelle
GO:0048471 : perinuclear region of cytoplasm
GO:0042470 : melanosome
GO:0033162 : melanosome membrane

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

Deletion

Deletion Size

1-9 bp

Molecular Details of the Mutation

a cytosine deletion in TYR at position 975 in exon 2

Experimental Evidence

Candidate Gene

Main Reference

Albinism in the domestic cat (*Felis catus*) is associated with a tyrosinase (TYR) mutation. (2006)

Authors

Imes DL; Geary LA; Grahn RA; Lyons LA

Abstract

Albino phenotypes are documented in a variety of species including the domestic cat. As albino phenotypes in other species are associated with tyrosinase (TYR) mutations, TYR was proposed as a candidate gene for albinism in cats. An Oriental and Colourpoint Shorthair cat pedigree segregating for albinism was analysed for association with TYR by linkage and sequence analyses. Microsatellite FCA931, which is closely linked to TYR and TYR sequence variants were tested for segregation with the albinism phenotype. Sequence analysis of genomic DNA from wild-type and albino cats identified a cytosine deletion in TYR at position 975 in exon 2, which causes a frame shift resulting in a premature stop codon nine residues downstream from the mutation. The deletion mutation in TYR and an allele of FCA931 segregated concordantly with the albino phenotype. Taken together, our results suggest that the TYR gene corresponds to the colour locus in cats and its alleles, from dominant to recessive, are as follows: C (full colour) > c(b) (burmese) > or = c(s) (siamese) > c (albino).

Additional References

RELATED GEPHE

Related Genes

6 (Agouti, Kit (type III receptor protein-tyrosine kinase), MC1R, Melanophilin (MLPH), Taqpep, tyrosinase-related protein 1 (TYRP1))

Related Haplotypes

4

EXTERNAL LINKS

COMMENTS

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