

GEPHE SUMMARY

Gephebase Gene
tyrosinase (TYR)

Entry Status
Published

GepheID
GP00002313

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
WT

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

SNP

SNP Coding Change

Nonsense

Molecular Details of the Mutation

c.1204C>T p.R402*

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	STP	402

Main Reference

Allelic heterogeneity of albinism in the domestic cat. (2017)

Authors

Abitbol M; BossÃ© P; Grimard B; Martignat L; Turet L

Abstract

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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

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EXTERNAL LINKS

COMMENTS

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