

GEPHE SUMMARY

Gephebase Gene
tyrosinase (TYR)

Entry Status
Published

GepheID
GP00002311

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
WT

Trait State in Taxon B
Burmese Mocha coat from Thailand

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

No

Molecular Type

Coding

Aberration Type

Indel

Indel Size

100-999 bp

Molecular Details of the Mutation

c.820_936delinsAATCTC p.L274_L312delinsNL

Experimental Evidence

Candidate Gene

Main Reference

Mocha tyrosinase variant: a new flavour of cat coat coloration. (2019)

Authors

Yu Y; Grahn RA; Lyons LA

Abstract

A novel coloration named 'mocha' has been identified in the Burmese cat breed from Thailand. Tyrosinase (TYR) mutations are known to be associated with coat coloration in cats, such as the sable Burmese, the points of the Siamese and albino cats. Additionally, sable Burmese that produced mocha-colored cats had unexpected genotypes for TYR. Therefore, TYR was considered a candidate gene for mocha in cats. Sanger sequencing for genomic DNA revealed NC_018732.3:chromosome D1:45â 898â 609_45â 898â 771dup in exon 2 and intron 2 of TYR. Transcription analysis using cDNA detected c.820_936delinsAATCTC (p.L274_L312delinsNL), which caused a 111-bp (37 amino acid) deletion in the reading frame of TYR. The identified variant was concordant with the phenotype and segregated with TYR variants in a pedigree of 12 Burmese cats. This findings of this study suggest that TYR is associated with the mocha coloration in cats. The new color variant adds to the allelic series for TYR (C \hat{A} > \hat{A} c \hat{A} = \hat{A} c \hat{A} > \hat{A} c, c) and is recessive to full color (C); however, interactions with the c and c alleles are unclear due to the temperature-sensitivity of the alleles.

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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

4

EXTERNAL LINKS

COMMENTS

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