GEPHE SUMMARY

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

GepheID
GP00002311

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
WT

Trait State in Taxon B
Burmese Mocha coat from Thailand

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

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

Indel Size

100-999 bp

Molecular Details of the Mutation

c.820_936delinsAATCTC p.I274_L312delinsNL

Experimental Evidence

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

Main Reference

Mocha tyrosinase variant: a new flavour of cat coat coloration. (2019) (https://pubmed.ncbi.nlm.nih.gov/30716167)

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.I274_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 (CÂ >Â c Â =Â c Â >Â 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)) (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/