GEPHE SUMMARY

Gephebase Gene
MC1R

GephelD
GP00001330

Entry Status
Published

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Felis silvestris catus-Sable Burmese

Trait State in Taxon B
Felis silvestris catus-Russet Burmese

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

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 Infraspecies?
Yes

Taxon A Description
Felis silvestris catus-Sable Burmese

Taxon B

Latin Name
Felis catus

Common Name
domestic cat

Synonyms
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758

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 Infraspecies?
Yes

Taxon B Description
Felis silvestris catus-Russet Burmese

GENOTYPIC CHANGE

Generic Gene Name
MC1R

Synonyms
CMM5; MSH-R; SHEP2; MSHR

String
9606.ENSP00000451605

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0008528 : G protein-coupled peptide receptor activity
GO:0004977 : melanocortin receptor activity
GO:0004980 : melanocyte-stimulating hormone receptor activity
GO:0031625 : ubiquitin protein ligase binding

GO - Biological Process
GO:0007275 : multicellular organism development
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0042438 : melanin biosynthetic process

UniProtKB Homo sapiens
Q01726

GenebankID or UniProtKB

GO:0043473 : pigmentation
GO:0007186 : G protein-coupled receptor signaling pathway
GO:0051897 : positive regulation of protein kinase B signaling
GO:0019233 : sensory perception of pain
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
GO:0035556 : intracellular signal transduction
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger
GO:0032720 : negative regulation of tumor necrosis factor production
GO:0010739 : positive regulation of protein kinase A signaling
GO:0090037 : positive regulation of protein kinase C signaling
GO:0009650 : UV protection
GO:0070914 : UV-damage excision repair

GO - Cellular Component
GO:0005886 : plasma membrane
GO:0005887 : integral component of plasma membrane

Presumptive Null
No

Molecular Type
Coding

Aberration Type
Deletion

Deletion Size
1-9 bp

Molecular Details of the Mutation
3bp deletion at c.439-441del (p.Phe146del)

Experimental Evidence
Candidate Gene

Main Reference
Not another type of potato: MC1R and the russet coloration of Burmese cats. (2017)

Authors
Gustafson NA; Gandolfi B; Lyons LA

Abstract
The Burmese is a breed of domestic cat that originated in Southeast Asia and was further developed in the United States. Variants in melanocortin 1 receptor (MC1R) causes common coat colour phenotypes in a variety of mammalian species but only limited colour variation in the domestic cat. Known as the extension (E) locus, melanocortin 1 receptor (MC1R) interacts with the agouti locus to produce the eumelanin and pheomelanin pigments. Recently, a novel reddish coloration, which is termed russet, was identified in the Burmese cat breed. Because this russet Burmese coloration changes with aging, MC1R was suggested as candidate gene. The similar colouration in specific lineages of Norwegian Forest cat known as amber (e) (c.250G>A; p.Asp84Asn) was excluded for this Burmese phenotype. The complete 954-bp coding region of MC1R was directly sequenced in russet Burmese and suspected carriers. A 3-bp deletion (c.439_441del) associated with the deletion of a phenyalanine (p.Phe146del) in the protein sequence was identified. All russet coloured cats were homozygous for the variant, and all obligate carriers were heterozygous, confirming that the deletion segregated concordantly with colouring in Burmese cats from the New Zealand foundation lineage. The variant was not identified in 442 cats from 26 different breeds and random-bred cats. Twenty-six Burmese from the USA did not have the variant. This MC1R variant defines a unique coloration and the second breed-specific MC1R variant in cats. The interactions of the two recessive feline MC1R alleles (EÂ > Â e, e) is unknown.

Additional References

RELATED GEPHE

Related Genes
6 (Agouti, Kit (type III receptor protein-tyrosine kinase), Melanophilin (MLPH), Taqpep, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1))
Related Haplotypes
No matches found.

COMMENTS

autosomal recessive