

GEPHE SUMMARY

Gephebase Gene
CMAH

Entry Status
Published

GepheID
GP00002168

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Blood type (feline ABC)

Trait State in Taxon A
Blood antigen A

Trait State in Taxon B
Reduction (type B) or loss (type C) of antigen A

Ancestral State
Taxon A

Taxonomic Status
Intraspecific

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
Cmah

Synonyms
-

String
10090.ENSMUSP00000129007

Sequence Similarities
Belongs to the CMP-Neu5Ac hydroxylase family.

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0051537 : 2 iron, 2 sulfur cluster binding
GO:0030338 : CMP-N-acetylneuraminate monooxygenase activity

GO - Biological Process
GO:0046381 : CMP-N-acetylneuraminate metabolic process
GO:0006054 : N-acetylneuraminate metabolic process

GO - Cellular Component
GO:0005737 : cytoplasm
GO:0005783 : endoplasmic reticulum

UniProtKB Mus musculus
Q61419

GenebankID or UniProtKB

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1600G>A p.D534N

Experimental Evidence

Candidate Gene

| | Taxon A | Taxon B | Position |
|------------|---------|---------|----------|
| Codon | - | - | - |
| Amino-acid | Asp | Asn | 534 |

Main Reference

Molecular Characterization of the Cytidine Monophosphate-N-Acetylneuraminic Acid Hydroxylase (CMAH) Gene Associated with the Feline AB Blood Group System. (2016)

Authors

Omi T; Nakazawa S; Udagawa C; Tada N; Ochiai K; Chong YH; Kato Y; Mitsui H; Gin A; Oda H; Azakami D; Tamura K; Sako T; Inagaki T; Sakamoto A; Tsutsui T; Bonkobara M; Tsuchida S; Ikemoto S

Abstract

Cat's AB blood group system (blood types A, B, and AB) is of major importance in feline transfusion medicine. Type A and type B antigens are Neu5Gc and Neu5Ac, respectively, and the enzyme CMAH participating in the synthesis of Neu5Gc from Neu5Ac is associated with this cat blood group system. Rare type AB erythrocytes express both Neu5Gc and Neu5Ac. Cat serum contains naturally occurring antibodies against antigens occurring in the other blood types. To understand the molecular genetic basis of this blood group system, we investigated the distribution of AB blood group antigens, CMAH gene structure, mutation, diplotypes, and haplotypes of the cat CMAH genes. Blood-typing revealed that 734 of the cats analyzed type A (95.1%), 38 cats were type B (4.9%), and none were type AB. A family of three Ragdoll cats including two type AB cats and one type A was also used in this study. CMAH sequence analyses showed that the CMAH protein was generated from two mRNA isoforms differing in exon 1. Analyses of the nucleotide sequences of the 16 exons including the coding region of CMAH examined in the 34 type B cats and in the family of type AB cats carried the CMAH variants, and revealed multiple novel diplotypes comprising several polymorphisms. Haplotype inference, which was focused on non-synonymous SNPs revealed that eight haplotypes carried one to four mutations in CMAH, and all cats with type B (n = 34) and AB (n = 2) blood carried two alleles derived from the mutated CMAH gene. These results suggested that double haploids selected from multiple recessive alleles in the cat CMAH loci were highly associated with the expression of the Neu5Ac on erythrocyte membrane in types B and AB of the feline AB blood group system.

Additional References

CMAH genotyping survey for blood types A, B and C (AB) in purpose-bred cats. (2019)

Cytidine monophospho-N-acetylneuraminic acid hydroxylase (CMAH) mutations associated with the domestic cat AB blood group. (2007)

RELATED GEPHE**Related Genes**

No matches found.

Related Haplotypes

5

EXTERNAL LINKS**COMMENTS**

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