GEPHE SUMMARY

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

GepheID
GP00002166

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Blood type (feline ABC) (https://www.gephebase.org/search-criteria?/and+Trait=^Blood type (feline ABC)^#gephebase-summary-title)

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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#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
Cmah

Synonyms
-

String
10090.ENSMUSP00000129007 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000129007)

Sequence Similarities
Belongs to the CMP-Neu5Ac hydroxylase family.

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0051537 : 2 iron, 2 sulfur cluster binding (https://www.ebi.ac.uk/QuickGO/term/GO:0051537)
GO:0030338 : CMP-N-acetylneuraminate monooxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0030338)

GO - Biological Process

UniProtKB Mus musculus
Q61419 (http://www.uniprot.org/uniprot/Q61419)

GenebankID or UniProtKB
()

GO:0046381 : CMP-N-acetylneuraminate metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0046381)
GO:0006054 : N-acetylneuraminate metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006054)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005783 : endoplasmic reticulum (https://www.ebi.ac.uk/QuickGO/term/GO:0005783)

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.139G>A p.V47M

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Val	Met	47

Main Reference

Molecular Characterization of the Cytidine Monophosphate-N-Acetylneuraminic Acid Hydroxylase (CMAH) Gene Associated with the Feline AB Blood Group System. (2016) (https://pubmed.ncbi.nlm.nih.gov/27755584)

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) (https://pubmed.ncbi.nlm.nih.gov/30854707)
Cytidine monophospho-N-acetylneuraminic acid hydroxylase (CMAH) mutations associated with the domestic cat AB blood group. (2007) (https://pubmed.ncbi.nlm.nih.gov/17553163)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

5 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^CMAH^/and+Taxon ID=^9685^/or+Gene Gephebase=^CMAH^/and+Taxon ID=^9685^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA000119/9685/