

GEPHE SUMMARY

CMAH (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=%CMAH%#gephebase-summary-title)	Gephebase Gene	GP00002163	GephelD
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category	Trait	
Physiology (https://www.gephebase.org/search-criteria/?and+Trait+Category=%Physiology%#gephebase-summary-title)			
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	Taxon B	Latin Name
Felis catus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Felis+catus%#gephebase-summary-title)		Felis catus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=%Felis+catus%#gephebase-summary-title)	
domestic cat	Common Name	domestic cat	Common Name
	Synonyms		Synonyms
Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.		Felis domesticus; Felis silvestris catus; domestic cat; cat; cats; Felis catus Linnaeus, 1758; Korat cats L.	
	Rank		Rank
species	Lineage	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		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		Parent
Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)		Felis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9682)	
9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)	NCBI Taxonomy ID	9685 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9685)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

Cmah	Generic Gene Name	UniProtKB Mus musculus
-	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000129007 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000129007)	0	
	String	
Belongs to the CMP-Neu5Ac hydroxylase family.	Sequence Similarities	
	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	

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.179G>T p.G60V

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	Glu	Val	60

Main Reference

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

(<https://pubmed.ncbi.nlm.nih.gov/27755584>)

Authors

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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>)

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/>

