GEPHE SUMMARY

Gephebase Gene

MC1R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC1R^#gephebase-summary-title)

GepheID

GP00002302

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 coat

Trait State in Taxon B

cream color in Australian Cattle Dog

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus^#gephebase-summary-title)

Common Name

gray wolf

Synonyms

gray wolf; grey wolf; Canis lupus 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; Caniformia; Canidae; Canis

Parent

Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)

NCBI Taxonomy ID

9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name

dog

Synonyms

Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank

subspecies

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; Caniformia; Canidae; Canis; Canis lupus

Parent

Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

NCBI Taxonomy ID

9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

MC1R

Synonyms

CMM5; MSH-R; SHEP2; MSHR

String

9606.ENSP00000451605 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000451605)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)
GO:0004980 : melanocyte-stimulating hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004980)

UniProtKB Homo sapiens

Q01726 (http://www.uniprot.org/uniprot/Q01726)

GenebankID or UniProtKB

()

GO:0031625 : ubiquitin protein ligase binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031625)

GO - Biological Process

GO:0007275 : multicellular organism development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
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:0007186 : G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0051897 : positive regulation of protein kinase B signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0051897)
GO:0019233 : sensory perception of pain
(https://www.ebi.ac.uk/QuickGO/term/GO:0019233)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007189)
GO:0035556 : intracellular signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0035556)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide
second messenger (https://www.ebi.ac.uk/QuickGO/term/GO:0007187)
GO:0032720 : negative regulation of tumor necrosis factor production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032720)
GO:0010739 : positive regulation of protein kinase A signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0010739)
GO:0090037 : positive regulation of protein kinase C signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0090037)
GO:0009650 : UV protection (https://www.ebi.ac.uk/QuickGO/term/GO:0009650)
GO:0070914 : UV-damage excision repair
(https://www.ebi.ac.uk/QuickGO/term/GO:0070914)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

single nucleotide variant within the MITF binding site of the canine MC1R promoter

Experimental Evidence

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

Main Reference

Two MC1R loss-of-function alleles in cream-coloured Australian Cattle Dogs and white Huskies. (2018) (https://pubmed.ncbi.nlm.nih.gov/29932470)

Authors

DÃ¼rig N; Letko A; Lepori V; Hadji Rasouliha S; Loechel R; Kehl A; HytÃ¶nen MK; Lohi H; Mauri N; Dietrich J; Wiedmer M; DrÃ¶gemÃ¼ller M; Jagannathan V; Schmutz SM; Leeb T

Abstract

Loss-of-function variants in the MC1R gene cause recessive red or yellow coat-colour phenotypes in many species. The canine MC1R:c.916C>T (p.Arg306Ter) variant is widespread and found in a homozygous state in many uniformly yellow- or red-coloured dogs. We investigated cream-coloured Australian Cattle Dogs whose coat colour could not be explained by this variant. A genome-wide association study with 10 cream and 123 red Australian Cattle Dogs confirmed that the cream locus indeed maps to MC1R. Whole-genome sequencing of cream dogs revealed a single nucleotide variant within the MITF binding site of the canine MC1R promoter. We propose to designate the mutant alleles at MC1R:c.916C>T as e and at the new promoter variant as e . Both alleles segregate in the Australian Cattle Dog breed. When we considered both alleles in combination, we observed perfect association between the MC1R genotypes and the cream coat colour phenotype in a cohort of 10 cases and 324 control dogs. Analysis of the MC1R transcript levels in an e /e compound heterozygous dog confirmed that the transcript levels of the e allele were markedly reduced with respect to the e allele. We further report another MC1R loss-of-function allele in Alaskan and Siberian Huskies caused by a 2-bp deletion in the coding sequence, MC1R:c.816_817delCT. We propose to term this allele e . Huskies that carry two copies of MC1R loss-of-function alleles have a white coat colour.

Â© 2018 Stichting International Foundation for Animal Genetics.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103), RALY (hnRNP associated with lethal yellow)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9612^/and+Trait=Coloration/or+Taxon ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

4 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9612^/or+Gene Gephebase=^MC1R^/and+Taxon ID=^9615^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001199/9615/ @Parallelism