GEPHE SUMMARY

Gephebase Gene

MFSD12 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MFSD12^#gephebase-summary-title)

GepheID

GP00002244

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

sabre or red phaeomanlin in various dog breeds

Trait State in Taxon B

light cream or white coat in various breeds

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 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 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

MFSD12

Synonyms

PP3501; C19orf28

String

9606.ENSP00000347583 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000347583)

Sequence Similarities

Belongs to the major facilitator superfamily.

GO - Molecular Function

GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)

GO - Biological Process

GO:0048022 : negative regulation of melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0048022)
GO:0008643 : carbohydrate transport (https://www.ebi.ac.uk/QuickGO/term/GO:0008643)

UniProtKB Homo sapiens

Q6NUT3 (http://www.uniprot.org/uniprot/Q6NUT3)

GenebankID or UniProtKB

()

GO:0071702 : organic substance transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0071702)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)
GO:0005770 : late endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005770)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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.151C>T p.Arg51Cys

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	51

Main Reference

Identification of a Missense Variant in MFSD12 Involved in Dilution of Phaeomelanin Leading to White or Cream Coat Color in Dogs. (2019) (https://pubmed.ncbi.nlm.nih.gov/31117290)

Authors

HÃ©dan B; Cadieu E; Botherel N; Dufaure de Citres C; Letko A; Rimbault M; DrÃ¶gemÃ¼ller C; Jagannathan V; Derrien T; Schmutz S; Leeb T; AndrÃ© C

Abstract

White coat color in mammals has been selected several times during the domestication process. Numerous dog breeds are fixed for one form of white coat color that involves darkly pigmented skin. The genetic basis of this color, due to the absence of pigment in the hairs, was suggested to correspond to extreme dilution of the phaeomelanin, by both the expression of only phaeomelanin (locus E) and its extreme dilution (locus I). To go further, we performed genome-wide association studies (GWAS) using a multiple breed approach. The first GWAS, using 34 white dogs and 128 non-white dogs, including White Shepherds, Poodles, Cotons de Tulear and Bichons allowed us to identify two significantly associated loci on the locus E and a novel locus on chromosome 20. A second GWAS using 15 other breeds presenting extreme phaeomelanin dilution confirmed the position of locus I on the chromosome 20 (position 55 Mb p = 6 Ã— 10). Using whole-genome sequencing, we identified a missense variant in the first exon of MFSD12, a gene recently identified to be involved in human, mouse and horse pigmentation. We confirmed the role of this variant in phaeomelanin dilution of numerous canine breeds, and the conserved role of MFSD12 in mammalian pigmentation.

Additional References

RELATED GEPHE

Related Genes

12 (Agouti (ASIP), GPR22, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA002197/9615/