

## GEPHE SUMMARY

MFSD12 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~MFSD12^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~MFSD12^#gephebase-summary-title</a> )	Gephebase Gene	GP00002244	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology^#gephebase-summary-title</a> )	Trait Category		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat^#gephebase-summary-title</a> )	Trait		
sabre or red phaeomanlin in various dog breeds	Trait State in Taxon A		
light cream or white coat in various breeds	Trait State in Taxon B		
	Ancestral State		
	Taxon A		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated^#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris^#gephebase-summary-title</a> )	Latin Name	Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris^#gephebase-summary-title</a> )	Latin Name
dog	Common Name	dog	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms
subspecies	Rank	subspecies	Rank
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	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	Lineage
Canis lupus (gray wolf) - (Rank: species) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )	Parent	Canis lupus (gray wolf) - (Rank: species) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )	Parent
9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a> )	NCBI Taxonomy ID	9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

MFSD12	Generic Gene Name	Q6NUT3 ( <a href="http://www.uniprot.org/uniprot/Q6NUT3">http://www.uniprot.org/uniprot/Q6NUT3</a> )	UniProtKB Homo sapiens
PP3501; C19orf28	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000347583 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000347583">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000347583</a> )	String		
Belongs to the major facilitator superfamily.	Sequence Similarities		
GO:0015293 : symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0015293">https://www.ebi.ac.uk/QuickGO/term/GO:0015293</a> )	GO - Molecular Function		
GO:0048022 : negative regulation of melanin biosynthetic process ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0048022">https://www.ebi.ac.uk/QuickGO/term/GO:0048022</a> )	GO - Biological Process		
GO:0008643 : carbohydrate transport ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008643">https://www.ebi.ac.uk/QuickGO/term/GO:0008643</a> )			

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](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](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](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](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; Bothrel 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 Å– 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), Microphthalmia-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](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/>