

GEPHE SUMMARY

Gephebase Gene		GepheID
Melanophilin (MLPH) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Melanophilin (MLPH)"#gephebase-summary-title)		GP00000643
Entry Status		Martin
Published		Main curator

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	
Canis familiaris - various breeds		Trait State in Taxon B	
Canis familiaris -"dilute" in Chow Chow ; Sloughi ; Thai Ridgeback breeds		Ancestral State	
Taxon A		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated"#gephebase-summary-title)			
Taxon A		Taxon B	
Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Canis+lupus lupus"#gephebase-summary-title)		Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Canis+lupus+familiaris familiaris"#gephebase-summary-title)	
Common Name		Common Name	
gray wolf		dog	
Synonyms		Synonyms	
gray wolf; grey wolf; Canis lupus Linnaeus, 1758		Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	
Rank		Rank	
species		subspecies	
Lineage		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		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		Parent	
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)		Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)		9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Canis familiaris -"dilute"	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Mus musculus
Mlph		Q91V27 (http://www.uniprot.org/uniprot/Q91V27)
Synonyms		GenebankID or UniProtKB
In; lIRk3; Slac-2a; AW228792; D1Wsu84e; l(1)-3Rk; 2210418F23Rik; 5031433l09Rik; Ln; Slac2a		NP_001096689 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001096689)
String		
10090.ENSMUSP00000027528 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000027528)		
Sequence Similarities		
-		
GO - Molecular Function		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)		
GO:0017137 : Rab GTPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017137)		
GO:0003779 : actin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003779)		

GO:0030674 : protein binding, bridging
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030674>)
GO:0051010 : microtubule plus-end binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051010>)
GO:0017022 : myosin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0017022>)
GO:0031489 : myosin V binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0031489>)
GO - Biological Process
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0032400 : melanosome localization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032400>)
GO:0006605 : protein targeting (<https://www.ebi.ac.uk/QuickGO/term/GO:0006605>)
GO - Cellular Component

GO:0015629 : actin cytoskeleton (<https://www.ebi.ac.uk/QuickGO/term/GO:0015629>)
GO:0030425 : dendrite (<https://www.ebi.ac.uk/QuickGO/term/GO:0030425>)
GO:0048471 : perinuclear region of cytoplasm
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)
GO:0005815 : microtubule organizing center
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005815>)
GO:0030864 : cortical actin cytoskeleton
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030864>)
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)
GO:0001725 : stress fiber (<https://www.ebi.ac.uk/QuickGO/term/GO:0001725>)
GO:0016461 : unconventional myosin complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016461>)

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.705G>C p.Gln235His
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	Gln	His	235

Main Reference
A novel MLPH variant in dogs with coat colour dilution. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29349785>)
Authors
Bauer A; Kehl A; Jagannathan V; Leeb T
Abstract

Coat colour dilution may be the result of altered melanosome transport in melanocytes. Loss-of-function variants in the melanophilin gene (MLPH) cause a recessively inherited form of coat colour dilution in many mammalian and avian species including the dog. MLPH corresponds to the D locus in many domestic animals, and recessive alleles at this locus are frequently denoted with d. In this study, we investigated dilute coloured Chow Chows whose coat colour could not be explained by their genotype at the previously known MLPH:c.-22G>A variant. Whole genome sequencing of such a dilute Chow Chow revealed another variant in the MLPH gene: MLPH:c.705G>C. We propose to designate the corresponding mutant alleles at these two variants d and d. We performed an association study in a cohort of 15 dilute and 28 non-dilute Chow Chows. The dilute dogs were all either compound heterozygous d /d or homozygous d /d , whereas the non-dilute dogs carried at least one wildtype allele D. The d allele did not occur in 417 dogs from diverse other breeds. However, when we genotyped a Sloughi family, in which a dilute coloured puppy had been born out of non-dilute parents, we again observed perfect co-segregation of the newly discovered d allele with coat colour dilution. Finally, we identified a blue Thai Ridgeback with the d /d genotype. Thus, our data identify the MLPH:c.705G>C as a variant explaining a second canine dilution allele. Although relatively rare overall, this d allele is segregating in at least three dog breeds, Chow Chows, Sloughis and Thai Ridgebacks.

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Additional References

RELATED GEPHE

Related Genes
13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Microphthalmia-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
2 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Melanophilin \(MLPH\)~/and+Taxon ID=~9612~/or+Gene Gephebase=~Melanophilin \(MLPH\)~/and+Taxon ID=~9615~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Melanophilin (MLPH)~/and+Taxon ID=~9612~/or+Gene Gephebase=~Melanophilin (MLPH)~/and+Taxon ID=~9615~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@Parallelism @AllelicSeries <https://omia.org/OMIA000031/9615/>