

Gephebase Gene		GepheID
Melanophilin (MLPH) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^Melanophilin+(MLPH)^#gephebase-summary-title)	GP00002330	Main curator
Published	Entry Status: Martin	

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

Generic Gene Name	UniProtKB Mus musculus
Mlph Synonyms In; I1Rk3; Slac-2a; AW228792; D1Wsu84e; I(1)-3Rk; 2210418F23Rik; 5031433I09Rik; Ln; Slac2a 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)	Q91V27 (http://www.uniprot.org/uniprot/Q91V27) GenebankID or UniProtKB NP_001096689 (https://www.ncbi.nlm.nih.gov/nuccore/NP_001096689)

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
-	Molecular Details of the Mutation
GA substitution at splice donor site causing Splicing defect	Experimental Evidence
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title)	

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

A noncoding melanophilin gene (MLPH) SNP at the splice donor of exon 1 represents a candidate causal mutation for coat color dilution in dogs. (2007)
(<https://pubmed.ncbi.nlm.nih.gov/17519392>)

DrÃ¶gemÃ¶ller C; Philipp U; Haase B; GÃ¶nzl-Apel AR; Leeb T

Coat color dilution in several breeds of dog is characterized by a specific pigmentation phenotype and sometimes accompanied by hair loss and recurrent skin inflammation, the so-called color dilution alopecia or black hair follicular dysplasia. Coat color dilution (d) is inherited as a Mendelian autosomal recessive trait. In a previous study, MLPH polymorphisms showed perfect cosegregation with the dilute phenotype within breeds. However, different dilute haplotypes were found in different breeds, and no single polymorphism was identified in the coding sequence that was likely to be causative for the dilute phenotype. We resequenced the 5'-region of the canine MLPH gene and identified a strong candidate single nucleotide polymorphism within the nontranslated exon 1, which showed perfect association to the dilute phenotype in 65 dilute dogs from 7 different breeds. The A/G polymorphism is located at the last nucleotide of exon 1 and the mutant A-allele is predicted to reduce splicing efficiency 8-fold. An MLPH mRNA expression study using quantitative reverse transcriptase-polymerase chain reaction confirmed that dd animals had only about approximately 25% of the MLPH transcript compared with DD animals. These results provide preliminary evidence that the reported regulatory MLPH mutation might represent a causal mutation for coat color dilution in dogs.

Additional References

RELATED GEPHE

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>)

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

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