

GEPHE SUMMARY

Gephebase Gene		GepheID
Melanophilin (MLPH) (<a +melanophilin+(mlph)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Melanophilin+(MLPH)+"#gephebase-summary-title)		GP00002331
Entry Status		Martin
Published		Main curator

PHENOTYPIC CHANGE

Trait Category			
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)			
Trait			
Coloration (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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 various breeds including Chihuahua ; Pekingese			
Ancestral State			
Taxon A			
Taxonomic Status			
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Canis lupus (<a +canis+lupus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+"#gephebase-summary-title)		Canis lupus familiaris (<a +canis+lupus+familiaris+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+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
Yes (#gephebase-summary-title)	
	Molecular Type
Coding (#gephebase-summary-title)	
	Aberration Type
Insertion (#gephebase-summary-title)	
	Insertion Size
1-9 bp	
	Molecular Details of the Mutation
c.667_668insC p.(His223Profs*41)	
	Experimental Evidence
Candidate Gene (#gephebase-summary-title)	
	Main Reference
A Third MLPH Variant Causing Coat Color Dilution in Dogs. (2020) (https://pubmed.ncbi.nlm.nih.gov/32531980)	
	Authors
Van Buren SL; Minor KM; Grahm RA; Mickelson JR; Grahm JC; Malvick J; Colangelo JR; Mueller E; Kuehnlein P; Kehl A	
	Abstract
Altered melanosome transport in melanocytes, resulting from variants in the melanophilin (MLPH) gene, are associated with inherited forms of coat color dilution in many species. In dogs, the MLPH gene corresponds to the D locus and two variants, c.-22G > A (d) and c.705G > C (d), leading to the dilution of coat color, as described. Here, we describe the independent investigations of dogs whose coat color dilution could not be explained by known variants, and who report a third MLPH variant, (c.667_668insC) (d), which leads to a frameshift and premature stop codon (p.His223Profs*41). The d allele is found at low frequency in multiple dog breeds, as well as in wolves, wolf-dog hybrids, and indigenous dogs. Canids in which the d allele contributed to the grey (dilute) phenotype were d/d compound heterozygotes or d homozygotes, and all non-dilute related dogs had one or two D alleles, consistent with a recessive inheritance. Similar to other loci responsible for coat colors in dogs, this, alongside likely additional allelic heterogeneity at the D locus, or other loci, must be considered when performing and interpreting genetic testing.	
	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)	

EXTERNAL LINKS

COMMENTS

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

