

GEPHE SUMMARY

PSMB7 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~PSMB7~#gephebase-summary-title)	Gephebase Gene	GP00000936	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)		Trait Category	
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(coat)~#gephebase-summary-title)		Trait	
Canis familiaris		Trait State in Taxon A	
Canis familiaris -Great Dane “Harlequin”		Trait State in Taxon B	
Data not curated		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~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)		Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Canis+lupus+familiaris~#gephebase-summary-title)	
gray wolf		dog	
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	
species		subspecies	
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	
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)	
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)	
No		Yes	
		Taxon B Description	
		Canis familiaris -Great Dane “Harlequin”	

GENOTYPIC CHANGE

Psmb7		Generic Gene Name		UniProtKB Mus musculus
MC14; AU020723; Mmc14		Synonyms		GenebankID or UniProtKB
10090.ENSMUSP00000028083 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000028083)		String		KHN77790 (https://www.ncbi.nlm.nih.gov/nuccore/KHN77790)
Belongs to the peptidase T1B family.		Sequence Similarities		
GO:0004175 : endopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004175)		GO - Molecular Function		
GO:0004298 : threonine-type endopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004298)		GO - Biological Process		

GO:0043161 : proteasome-mediated ubiquitin-dependent protein catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043161>)
GO:0010498 : proteasomal protein catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010498>)
GO:0010499 : proteasomal ubiquitin-independent protein catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010499>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0000502 : proteasome complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0000502>)
GO:0005839 : proteasome core complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005839>)
GO:0019774 : proteasome core complex, beta-subunit complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019774>)

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.146T>G p.V6G

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	Val	Gly	49

Main Reference

A missense mutation in the 20S proteasome β^2 subunit of Great Danes having harlequin coat patterning. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21256207>)

Authors

Clark LA; Tsai KL; Starr AN; Nowend KL; Murphy KE

Abstract

Harlequin is a pigmentary trait of the domestic dog that is controlled by two autosomal loci: the melanosomal gene, SILV, and a modifier gene, harlequin (H), previously localized to chromosome 9. Heterozygosity for a retrotransposon insertion in SILV and a mutation in H causes a pattern of black patches on a white background. Homozygosity for H is embryonic lethal. Fine mapping of the harlequin locus revealed a 25 kb interval wherein all harlequin Great Danes are heterozygous for a common haplotype. This region contains one gene, PSMB7, which encodes the β^2 catalytic subunit of the proteasome. Sequence analysis identified a coding variant in exon 2 that segregates with harlequin patterning. The substitution predicts the replacement of a highly conserved valine with a glycine. Described herein is the identification of a naturally-occurring mutation of the ubiquitin proteasome system that is associated with a discernable phenotype of dogs.

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Additional References

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, 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

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001454/9615/>

