GEPHE SUMMARY

Gephebase Gene

PSMB7 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PSMB7^#gephebase-summary-title)

GepheID

GP00000936

Entry Status

Published

Main curator

Martin

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

Trait State in Taxon B

Canis familiaris -Great Dane "Harlequin"

Ancestral State

Data not curated

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus^#gephebase-summary-title)

Common Name

gray wolf

Synonyms

gray wolf; grey wolf; Canis lupus Linnaeus, 1758

Rank

species

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

Parent

Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)

NCBI Taxonomy ID

9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name

dog

Synonyms

Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank

subspecies

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

Parent

Canis lupus (gray wolf) - (Rank: species) (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)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Canis familiaris -Great Dane "Harlequin"

GENOTYPIC CHANGE

Generic Gene Name

Psmb7

Synonyms

MC14; AU020723; Mmc14

String

10090.ENSMUSP00000028083 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000028083)

Sequence Similarities

Belongs to the peptidase T1B family.

GO - Molecular Function

GO:0004175 : endopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004175)
GO:0004298 : threonine-type endopeptidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004298)

GO - Biological Process

UniProtKB Mus musculus

P70195 (http://www.uniprot.org/uniprot/P70195)

GenebankID or UniProtKB

KHN77790 (https://www.ncbi.nlm.nih.gov/nuccore/KHN77790)

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 Î²2 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 Î²2 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), Microphtalmia-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/