GEPHE SUMMARY

Gephebase Gene

PMEL17 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PMEL17^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002072

Main curator

II

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

Shetland Sheepdog of wild-type color - nonmerle

Trait State in Taxon B

Shetland Sheepdog of merle color - patches of diluted pigment - autosomal and incompletely dominant

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

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 A an Infraspecies?

Yes

Taxon A Description

Shetland Sheepdog

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

Shetland Sheepdog

GENOTYPIC CHANGE

Generic Gene Name

Pmel

Synonyms

D10H12S53E; Pmel17; Si; Silv

String

10090.ENSMUSP00000051869 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000051869)

Sequence Similarities

Belongs to the PMEL/NMB family.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)

GO - Biological Process

UniProtKB Mus musculus

Q60696 (http://www.uniprot.org/uniprot/Q60696)

GenebankID or UniProtKB

()

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)
GO:0048023 : positive regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048023)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005783 : endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0032585 : multivesicular body membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0032585)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)

Insertion Size

100-999 bp

Molecular Details of the Mutation

due to a short interspersed element (SINE) insertion at the intron 10/exon 11 boundary. The SINE segregates with the merle phenotype in multiple breeds (Collie, Border Collie, Australian Shepherd, Cardigan Welsh Corgi, Dachshund, and Great Dane) and is absent from dogs representing breeds that do not have merle patterning. All examined harlequin Great Danes harbored the insertion in either a heterozygous or homozygous state.

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

Retrotransposon insertion in SILV is responsible for merle patterning of the domestic dog. (2006) (https://pubmed.ncbi.nlm.nih.gov/16407134)

Authors

Clark LA; Wahl JM; Rees CA; Murphy KE

Abstract

Merle is a pattern of coloring observed in the coat of the domestic dog and is characterized by patches of diluted pigment. This trait is inherited in an autosomal, incompletely dominant fashion. Dogs heterozygous or homozygous for the merle locus exhibit a wide range of auditory and ophthalmologic abnormalities, which are similar to those observed for the human auditory-pigmentation disorder Waardenburg syndrome. Mutations in at least five genes have been identified as causative for Waardenburg syndrome; however, the genetic bases for all cases have not been determined. Linkage disequilibrium was identified for a microsatellite marker with the merle phenotype in the Shetland Sheepdog. The marker is located in a region of CFA10 that exhibits conservation of synteny with HSA12q13. This region of the human genome contains SILV, a gene important in mammalian pigmentation. Therefore, this gene was evaluated as a candidate for merle patterning. A short interspersed element insertion at the boundary of intron 10/exon 11 was found, and this insertion segregates with the merle phenotype in multiple breeds. Another finding was deletions within the oligo(dA)-rich tail of the short interspersed element. Such deletions permit normal pigmentation. These data show that SILV is responsible for merle patterning and is associated with impaired function of the auditory and ophthalmologic systems. Although the mutant phenotype of SILV in the human is unknown, these results make it an intriguing candidate gene for human auditory-pigmentation disorders.

Additional References

RELATED GEPHE

Related Genes

12 (Agouti (ASIP), GPR22, MFSD12, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

5 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9615^/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9615^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @Splicing @TE - PMEL17 is also known as SILV and gp100 - Dogs heterozygous or homozygous for the merle locus exhibit a wide range of auditory and ophthalmologic abnormalities. Dogs homozygous for merle (MM) are known as double merles and are predominantly white - reversion rate (excision of the TE and reversion to the nonmerle color, which is then stable through the next progeny) in the germ line is quite high: 3â€“4%. @Pleiotropy with deleterious effect similar to the human auditoryâ€“pigmentation disorder Waardenburg syndrome @HeterozygoteAdvantage @AllelicSeries https://omia.org/OMIA000211/9615/