## GEPHE SUMMARY

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

GepheID
GP00002101

Entry Status
Published

Main curator
Courtier

## 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
wild-type color

Trait State in Taxon B
silver color

Ancestral State
Taxon A

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

### Taxon A

Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title)

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507 )

NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090 )

is Taxon A an Infraspecies?
No

### Taxon B

Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title)

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507 )

NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090 )

is Taxon B an Infraspecies?
No

## 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
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)

UniProtKB Mus musculus
Q60696 (http://www.uniprot.org/uniprot/Q60696)

GenebankID or UniProtKB
()

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

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

Aberration Type

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

single nucleotide (A) insertion in the putative cytoplasmic tail of the si/si Pmel 17 cDNA clone. This insertion is predicted to alter the last 24 amino acids at the C-terminus. Also predicted is the extension of the Pmel 17 protein by 12 residues because a new termination signal is created downstream from the wild-type reading frame. The silver pmel 17 protein has a major defect at the carboxyl terminus.

Experimental Evidence

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

Main Reference

Mouse silver mutation is caused by a single base insertion in the putative cytoplasmic domain of Pmel 17. (1995) (https://pubmed.ncbi.nlm.nih.gov/7870580)

Authors

Kwon BS; Halaban R; Ponnazhagan S; Kim K; Chintamaneni C; Bennett D; Pickard RT

Abstract

This laboratory has established in previous studies that Pmel 17, a gene expressed specifically in melanocytes, maps near the silver coat color locus (si/si) on mouse chromosome 10. In the current study, we have focused on determining whether or not the si allele carries a mutation in Pmel 17. Pmel 17 cDNA clones, isolated from wild-type and si/si murine melanocyte cDNA libraries, were sequenced and compared. A single nucleotide (A) insertion was found in the putative cytoplasmic tail of the si/si Pmel 17 cDNA clone. This insertion is predicted to alter the last 24 amino acids at the C-terminus. Also predicted is the extension of the Pmel 17 protein by 12 residues because a new termination signal created downstream from the wild-type reading frame. The mutation was confirmed by the sequence of the PCR-amplified genomic region flanking and including the mutation site. The fact that si/si Pmel 17 was not recognized by antibodies directed toward the C-terminal 15 amino acids of wild-type Pmel 17, indicated a defect in this region. We conclude from these results that silver pmel 17 protein has a major defect at the carboxyl terminus. The chromosomal location and the identification of a potentially pathologic mutation in si-Pmel 17 support our conclusion that Pmel 17 is encoded at the silver locus.

Additional References

## RELATED GEPHE

Related Genes

4 (Agouti, Agouti (ASIP), MC1R, SLC45A2=MATP) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^10090^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

PMEL17 is also known as SILV and gp100