

GEPHE SUMMARY

<p>PMEL17 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=~PMEL17~#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002101</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+coat~#gephebase-summary-title)</p> <p>wild-type color</p> <p>silver color</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Domesticated~#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Mus+musculus~#gephebase-summary-title)</p> <p>Common Name</p> <p>house mouse</p> <p>Synonyms</p> <p>house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=862507)</p> <p>NCBI Taxonomy ID</p> <p>10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10090)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Mus+musculus~#gephebase-summary-title)</p> <p>Common Name</p> <p>house mouse</p> <p>Synonyms</p> <p>house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=862507)</p> <p>NCBI Taxonomy ID</p> <p>10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10090)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>Pmel</p> <p>D10H12S53E; Pmel17; Si; Silv</p> <p>10090.ENSMUSP00000051869 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000051869)</p> <p>Belongs to the PMEL/NMB family.</p> <p>GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)</p> <p>GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)</p> <p>GO:0032438 : melanosome organization (https://www.ebi.ac.uk/QuickGO/term/GO:0032438)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>UniProtKB Mus musculus</p> <p>Q60696 (http://www.uniprot.org/uniprot/Q60696)</p> <p>GenebankID or UniProtKB</p> <p>0</p>
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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

3 (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