GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00002271

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

WT coat

Trait State in Taxon B

Silver dapple coat in breeds: American Miniature Horse ; Icelandic; Rocky Mountain

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

horse

Synonyms

Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus 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; Perissodactyla; Equidae; Equus; Equus

Parent

Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID

9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

horse

Synonyms

Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus 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; Perissodactyla; Equidae; Equus; Equus

Parent

Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)

NCBI Taxonomy ID

9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)

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

UniProtKB Mus musculus

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

GenebankID or UniProtKB

()

(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

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

g.73665304C>T p.R625C

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	625

Main Reference

A missense mutation in PMEL17 is associated with the Silver coat color in the horse. (2006) (https://pubmed.ncbi.nlm.nih.gov/17029645)

Authors

Brunberg E; Andersson L; Cothran G; Sandberg K; Mikko S; Lindgren G

Abstract

The Silver coat color, also called Silver dapple, in the horse is characterized by dilution of the black pigment in the hair. This phenotype shows an autosomal dominant inheritance. The effect of the mutation is most visible in the long hairs of the mane and tail, which are diluted to a mixture of white and gray hairs. Herein we describe the identification of the responsible gene and a missense mutation associated with the Silver phenotype.

Segregation data on the Silver locus (Z) were obtained within one half-sib family that consisted of a heterozygous Silver colored stallion with 34 offspring and their 29 non-Silver dams. We typed 41 genetic markers well spread over the horse genome, including one single microsatellite marker (TKY284) close to the candidate gene PMEL17 on horse chromosome 6 (ECA6q23). Significant linkage was found between the Silver phenotype and TKY284 (theta = 0, z = 9.0). DNA sequencing of PMEL17 in Silver and non-Silver horses revealed a missense mutation in exon 11 changing the second amino acid in the cytoplasmic region from arginine to cysteine (Arg618Cys). This mutation showed complete association with the Silver phenotype across multiple horse breeds, and was not found among non-Silver horses with one clear exception; a chestnut colored individual that had several Silver offspring when mated to different non-Silver stallions also carried the exon 11 mutation. In total, 64 Silver horses from six breeds and 85 non-Silver horses from 14 breeds were tested for the exon 11 mutation. One additional mutation located in intron 9, only 759 bases from the missense mutation, also showed complete association with the Silver phenotype. However, as one could expect to find several non-causative mutations completely associated with the Silver mutation, we argue that the missense mutation is more likely to be causative.

The present study shows that PMEL17 causes the Silver coat color in the horse and enable genetic testing for this trait.

Additional References

Two SNPs in the SILV gene are associated with silver coat colour in ponies. (2007) (https://pubmed.ncbi.nlm.nih.gov/17257181)
Pleiotropic effects of pigmentation genes in horses. (2010) (https://pubmed.ncbi.nlm.nih.gov/21070283)
Mutations in or near the transmembrane domain alter PMEL amyloid formation from functional to pathogenic. (2011) (https://pubmed.ncbi.nlm.nih.gov/21949659)
Equine multiple congenital ocular anomalies and silver coat colour result from the pleiotropic effects of mutant PMEL. (2013) (https://pubmed.ncbi.nlm.nih.gov/24086599)

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphtalmia-associated transcription factor, Pax3, SLC24A, SLC36A1, SLC45A2=MATP, syntaxin-17, T-box transcription factor (TBX3), TRPM1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9796^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Pleiotropy @Parallelism @HeterozygoteAdvantage https://omia.org/OMIA001438/9796/