GEPHE SUMMARY

Gephebase Gene
SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC45A2=MATP^#gephebase-summary-title)

GepheID
GP00002306

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
Equus caballus

Trait State in Taxon B
Dilute snowdrop in Gypsy

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
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)

Sequence Similarities
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.

GO - Molecular Function
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)

GO - Biological Process
GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)

UniProtKB Homo sapiens
Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)

GenebankID or UniProtKB
AAO25647 (https://www.ncbi.nlm.nih.gov/nuccore/AAO25647)

GO:0048066 : developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)
GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)
GO:0015770 : sucrose transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015770)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

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.305G>A p.(Arg102Gln)

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Gln	102

Main Reference

A candidate gene approach identifies variants in SLC45A2 that explain dilute phenotypes, pearl and sunshine, in compound heterozygote horses. (2019)
(https://pubmed.ncbi.nlm.nih.gov/31006892)

Authors

Holl HM; Pflug KM; Yates KM; Hoefs-Martin K; Shepard C; Cook DG; Lafayette C; Brooks SA

Abstract

Variations in the SLC45A2 gene are responsible for the dilution phenotypes cream and pearl in domestic horses. Cream dilution is inherited in an incomplete dominant manner, diluting only red in the heterozygous state but both red and black pigments when two alleles are present. The pearl dilution is recessive and dilutes only the red and black pigment in the homozygous state or when paired with a cream allele. Horses that inherit one copy of pearl (C) and one copy of the dominant cream allele (C) display a dilution phenotype similar to that of homozygous cream, suggesting that pearl is the result of a different variation in the same gene responsible for cream. We sequenced SLC45A2 in two 'false double dilute' horses that appeared phenotypically homozygous cream but tested as possessing only a single C allele. We also sequenced one known pearl carrier to screen for putative causal variants. The missense variant ECA21:SLC45A2:c.985G>A; p.Ala329Thr (C) was present in one false double dilute and the pearl carrier and was also genotyped in an additional 126 horses for statistical evaluation. The genotype matched the expected phenotype in all horses (P-valueÂ =Â 6.5Â Ã—Â 10) and is identical to a pearl variant found previously. The second false double dilute horse and one non-dilute offspring genotyped as heterozygous for a novel missense variant ECA21:SLC45A2:c.568G>A (p.Gly190Arg), the proposed C variant (for the name of the horse). This variant produces a recessive dilution similar to pearl and indicates that multiple alleles of SLC45A2 result in dilution phenotypes in the domestic horse.

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Additional References

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphtalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, 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

3 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9796^/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9796^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001344/9796/ @Parallelism