

GEPHE SUMMARY

	Gephebase Gene	GephelD
SLC45A2=MATP (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=^SLC45A2=MATP^#gephebase-summary-title)	GP00002305	
	Entry Status	Main curator
Published	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

Sunshine color in Standardbred Å— Tennessee and Walking Horse cross

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

1A1; AIM1; MATP; OCA4; SHEP5

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>)

No (https://www.gephebase.org/search-criteria/?and+Presumptive+Null=%22No%22#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria/?and+Molecular+Type=%22Coding%22#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria/?and+Aberration+Type=%22SNP%22#gephebase-summary-title)	Aberration Type
Nonsynonymous	SNP Coding Change
c.568G>A p.(Gly190Arg)	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria/?and+Experimental+Evidence=%22Candidate+Gene%22#gephebase-summary-title)	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Arg	190

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/>)

Holl HM; Pflug KM; Yates KM; Hoefs-Martin K; Shepard C; Cook DG; Lafayette C; Brooks SA	Authors
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-value = 6.5 × 10⁻¹⁰) 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.	Main Reference

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Additional References

RELATED GEPHE

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphthalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, syntaxin-17, T-box transcription factor (TBX3), TRPM1) (https://www.gephebase.org/search-criteria/?or+Taxon+ID=%229796%22+and+Trait=Coloration+and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
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3 (https://www.gephebase.org/search-criteria/?or+Gene+Gephebase=%22SLC45A2=MATP%22+and+Taxon+ID=%229796%22+or+Gene+Gephebase=%22SLC45A2=MATP%22+and+Taxon+ID=%229796%22#gephebase-summary-title)	Related Haplotypes
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EXTERNAL LINKS

COMMENTS

<https://omnia.org/OMIA001344/9796/> @Parallelism

