

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~SLC45A2=MATP^#gephebase-summary-title)	Gephebase Gene	GP00002304	GepheID
Published	Entry Status	Martin	Main curator

Taxon A		Taxon B	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait=^Morphology^#gephebase-summary-title)	Trait Category	Equus caballus	Trait Category
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title)	Trait	Pearl coat color in American Paint Horse ; Lusitano ; Purebred Spanish horse ; Quarter Horse	Trait
Equus caballus	Trait State in Taxon A	Equus caballus	Trait State in Taxon B
	Trait State in Taxon A		Trait State in Taxon B
	Ancestral State		Ancestral State
Taxon A	Taxonomic Status	Equus caballus	Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)		Equus caballus	
Equus caballus	Latin Name	Equus caballus	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus caballus^#gephebase-summary-title)	
horse	Common Name	horse	Common Name
	Synonyms		Synonyms
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
species	Rank	species	Rank
	Lineage		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		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		Parent
Equus () - (Rank: subgenus)		Equus () - (Rank: subgenus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	
9796	NCBI Taxonomy ID	9796	NCBI Taxonomy ID
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

SLC45A2	Generic Gene Name	Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	AAO25647 (https://www.ncbi.nlm.nih.gov/nuccore/AAO25647)	GenebankID or UniProtKB
9606.ENSPP00000296589 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000296589)	String		
	Sequence Similarities		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.			
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)	GO - Molecular Function		
GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)	GO - Biological Process		

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.985G>A p.(Ala329Thr)

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	Ala	Thr	329

Main Reference

Explicit evidence for a missense mutation in exon 4 of SLC45A2 gene causing the pearl coat dilution in horses. (2019) (<https://pubmed.ncbi.nlm.nih.gov/30968968>)

Authors

Sevane N; Sanz CR; Dunner S

Abstract

Four loci seem responsible for the dilution of the basic coat colours in horse: Dun (D), Silver Dapple (Z), Champagne (CH) and Cream (C). Apart from the current phenotypes ascribed to these loci, pearl has been described as yet another diluted coat colour in this species. To date, this coat colour seems to segregate only in the Iberian breeds Purebred Spanish horse and Lusitano and has also been described in breeds of Iberian origin, such as Quarter Horses and Paint Horse, where it is referred to as the ‘Barlink Factor’. This phenotype segregates in an autosomal recessive manner and resembles some of the coat colours produced by the champagne CH and cream C alleles, sometimes being difficult to distinguish among them. The interaction between compound heterozygous for the pearl C and cream C alleles makes SLC45A2 the most plausible candidate gene for the pearl phenotype in horses. Our results provide documented evidence for the missense variation in exon 4 [SLC45A2:c.985G>A; SLC45A2:p.(Ala329Thr)] as the causative mutation for the pearl coat colour. In addition, it is most likely involved as well in the cremello, perlino and smoky cream like phenotypes associated with the compound C and C heterozygous genotypes (known as cream pearl in the Purebred Spanish horse breed). The characterization of the pearl mutation allows breeders to identify carriers of the C allele and to select this specific coat colour according to personal preferences, market demands or studbook requirements as well as to verify segregation within particular pedigrees.

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Additional References

RELATED GEPHE

Related Genes

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=~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

