

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^SLC45A2=MATP^#gephebase-summary-title)	Gephebase Gene	GP00001057	GepheID
Published	Entry Status	Martin	Main curator

Taxon A		Taxon B	
Taxon A	Taxon B	Taxon A	Taxon B
Equus caballus	Equus caballus	Equus caballus	Equus caballus
(#https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus)	(#https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus)	Equus caballus	Equus caballus
horse	horse	horse	horse
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	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	species	species	species
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	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
Equus () - (Rank: subgenus)	Equus () - (Rank: subgenus)	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)	(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	9796	9796	9796
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)	(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?	is Taxon A an Intraspecies?	is Taxon B an Intraspecies?
No	No	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 - 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)			

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.457G>A p.(Asp153Asn)

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Asn	153

Main Reference

A mutation in the MATP gene causes the cream coat colour in the horse. (2003 Jan-Feb) (<https://pubmed.ncbi.nlm.nih.gov/12605854>)

Authors

Mariat D; Taourit S; GuÃ©rin G

Abstract

In horses, basic colours such as bay or chestnut may be partially diluted to buckskin and palomino, or extremely diluted to cream, a nearly white colour with pink skin and blue eyes. This dilution is expected to be controlled by one gene and we used both candidate gene and positional cloning strategies to identify the "cream mutation". A horse panel including reference colours was established and typed for different markers within or in the neighbourhood of two candidate genes. Our data suggest that the causal mutation, a G to A transition, is localised in exon 2 of the MATP gene leading to an aspartic acid to asparagine substitution in the encoded protein. This conserved mutation was also described in mice and humans, but not in medaka.

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