## GEPHE SUMMARY

Gephebase Gene
SLC36A1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC36A1^#gephebase-summary-title)

GepheID
GP00002278

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
Champagne color coat

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
SLC36A1

Synonyms
Dct1; PAT1; LYAAT1; TRAMD3

String
9606.ENSP00000243389 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000243389 )

Sequence Similarities
Belongs to the amino acid/polyamine transporter 2 family.

GO - Molecular Function
GO:0015171 : amino acid transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015171)
GO:0015193 : L-proline transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015193)
GO:0005280 : amino acid:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005280)

UniProtKB Homo sapiens
Q7Z2H8 (http://www.uniprot.org/uniprot/Q7Z2H8)

GenebankID or UniProtKB
()

GO:0015187 : glycine transmembrane transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0015187)
GO:0015180 : L-alanine transmembrane transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0015180)
GO:0015078 : proton transmembrane transporter activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0015078)

GO - Biological Process

GO:0006811 : ion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006811)
GO:0003333 : amino acid transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0003333)
GO:1902600 : proton transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:1902600)
GO:0006865 : amino acid transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006865)
GO:0015816 : glycine transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015816)
GO:0015808 : L-alanine transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015808)
GO:0035524 : proline transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0035524)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005774 : vacuolar membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005774)
GO:0005783 : endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)

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.188C>G p.T63R

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 | Tyr | Arg | 63 |

Main Reference

Missense mutation in exon 2 of SLC36A1 responsible for champagne dilution in horses. (2008) (https://pubmed.ncbi.nlm.nih.gov/18802473)

Authors

Cook D; Brooks S; Bellone R; Bailey E

Abstract

Champagne coat color in horses is controlled by a single, autosomal-dominant gene (CH). The phenotype produced by this gene is valued by many horse breeders, but can be difficult to distinguish from the effect produced by the Cream coat color dilution gene (CR). Three sires and their families segregating for CH were tested by genome scanning with microsatellite markers. The CH gene was mapped within a 6 cM region on horse chromosome 14 (LOD = 11.74 for theta = 0.00). Four candidate genes were identified within the region, namely SPARC [Secreted protein, acidic, cysteine-rich (osteonectin)], SLC36A1 (Solute Carrier 36 family A1), SLC36A2 (Solute Carrier 36 family A2), and SLC36A3 (Solute Carrier 36 family A3). SLC36A3 was not expressed in skin tissue and therefore not considered further. The other three genes were sequenced in homozygotes for CH and homozygotes for the absence of the dilution allele (ch). SLC36A1 had a nucleotide substitution in exon 2 for horses with the champagne phenotype, which resulted in a transition from a threonine amino acid to an arginine amino acid (T63R). The association of the single nucleotide polymorphism (SNP) with the champagne dilution phenotype was complete, as determined by the presence of the nucleotide variant among all 85 horses with the champagne dilution phenotype and its absence among all 97 horses without the champagne phenotype. This is the first description of a phenotype associated with the SLC36A1 gene.

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, 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

https://omia.org/OMIA001263/9796/