GEPHE SUMMARY

Gephebase Gene
SLC24A (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC24A^#gephebase-summary-title)

GepheID
GP00002277

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 (eyes) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (eyes)^#gephebase-summary-title)

Trait State in Taxon A
Paso Fino horses

Trait State in Taxon B
Paso Fino with "tiger eyes" with bright yellow amber or orange iris ; blue in homozygotes

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
Slc24a5

Synonyms
JSX; NCX5; Oca6; NCKX5; F630045L20Rik; Nckx5

String
10090.ENSMUSP00000063887 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000063887)

Sequence Similarities
Belongs to the Ca(2+):cation antiporter (CaCA) (TC 2.A.19) family. SLC24A subfamily.

GO - Molecular Function
GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)
GO:0005262 : calcium channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005262)
GO:0008273 : calcium, potassium:sodium antiporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008273)

GO - Biological Process

UniProtKB Mus musculus
Q8C261 (http://www.uniprot.org/uniprot/Q8C261)

GenebankID or UniProtKB
()

GO:0006874 : cellular calcium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0070588 : calcium ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0070588)
GO:0034220 : ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0034220)
GO:0048022 : negative regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048022)
GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0005802 : trans-Golgi network (https://www.ebi.ac.uk/QuickGO/term/GO:0005802)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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.272A>T p.Phe91Tyr

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Phe	Tyr	91

Main Reference

Two Variants in SLC24A5 Are Associated with "Tiger-Eye" Iris Pigmentation in Puerto Rican Paso Fino Horses. (2017) (https://pubmed.ncbi.nlm.nih.gov/28655738)

Authors

Mack M; Kowalski E; Grahn R; Bras D; Penedo MCT; Bellone R

Abstract

A unique eye color, called tiger-eye, segregates in the Puerto Rican Paso Fino (PRPF) horse breed and is characterized by a bright yellow, amber, or orange iris. Pedigree analysis identified a simple autosomal recessive mode of inheritance for this trait. A genome-wide association study (GWAS) with 24 individuals identified a locus on ECA 1 reaching genome-wide significance (P = 1.32 Ã— 10). This ECA1 locus harbors the candidate gene, Solute Carrier Family 24 (Sodium/Potassium/Calcium Exchanger), Member 5 (SLC24A5), with known roles in pigmentation in humans, mice, and zebrafish. Humans with compound heterozygous mutations in SLC24A5 have oculocutaneous albinism (OCA) type 6 (OCA6), which is characterized by dilute skin, hair, and eye pigmentation, as well as ocular anomalies. Twenty tiger-eye horses were homozygous for a nonsynonymous mutation in exon 2 (p.Phe91Tyr) of SLC24A5 (called here Tiger-eye 1), which is predicted to be deleterious to protein function. Additionally, eight of the remaining 12 tiger-eye horses heterozygous for the p.Phe91Tyr variant were also heterozygous for a 628 bp deletion encompassing all of exon 7 of SLC24A5 (c.875-340_1081+82del), which we will call here the Tiger-eye 2 allele. None of the 122 brown-eyed horses were homozygous for either tiger-eye-associated allele or were compound heterozygotes. Further, neither variant was detected in 196 horses from four related breeds not known to have the tiger-eye phenotype. Here, we propose that two mutations in SLC24A5 affect iris pigmentation in tiger-eye PRPF horses. Further, unlike OCA6 in humans, the Tiger-eye 1 mutation in its homozygous state or as a compound heterozygote (Tiger-eye 1/Tiger-eye 2) does not appear to cause ocular anomalies or a change in coat color in the PRPF horse.

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, SLC36A1, 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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLC24A^/and+Taxon ID=^9796^/or+Gene Gephebase=^SLC24A^/and+Taxon ID=^9796^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA002124/9796/ @AllelicSeries