

GEPHE SUMMARY

SLC24A ( <a +slc24a+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+SLC24A+"#gephebase-summary-title</a> )	Gephebase Gene	GP00002277	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology ([https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Trait+Category=))

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

Paso Fino horses

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

Domesticated ([https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=))

Taxon A	Taxon B
Equus caballus ( <a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus+"#gephebase-summary-title</a> )	Equus caballus ( <a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus+"#gephebase-summary-title</a> )
Common Name	Common Name
horse	horse
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
Rank	Rank
species	species
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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a> )	Equus () - (Rank: subgenus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a> )
NCBI Taxonomy ID	NCBI Taxonomy ID
9796 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a> )	9796 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a> )
is Taxon A an Intraspecies?	is Taxon B an Intraspecies?
No	No

GENOTYPIC CHANGE

SLC24A5	Generic Gene Name	Q8C261 ( <a href="http://www.uniprot.org/uniprot/Q8C261">http://www.uniprot.org/uniprot/Q8C261</a> )	UniProtKB Mus musculus
JSX; NCX5; Oca6; NCKX5; F630045L20Rik; Nckx5	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000063887 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000063887">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000063887</a> )	String		
Belongs to the Ca(2+):cation antiporter (CaCA) (TC 2.A.19) family. SLC24A subfamily.	Sequence Similarities		
GO:0015293 : symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0015293">https://www.ebi.ac.uk/QuickGO/term/GO:0015293</a> )	GO - Molecular Function		
GO:0005262 : calcium channel activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005262">https://www.ebi.ac.uk/QuickGO/term/GO:0005262</a> )			
GO:0008273 : calcium, potassium:sodium antiporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008273">https://www.ebi.ac.uk/QuickGO/term/GO:0008273</a> )			
	GO - Biological Process		

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 ( $P = 1.32 \times 10^{-10}$ ). 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.

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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, 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

