

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)	Gephebase Gene	GP00002276	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)	Trait Category		
Coloration (eyes) (<a +coloration+(eyes)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(eyes)+"#gephebase-summary-title)	Trait		
Paso Fino horses	Trait State in Taxon A		
Paso Fino with "tiger eyes" with bright yellow amber or orange iris ; blue in homozygotes	Trait State in Taxon B		
	Ancestral State		
	Taxon A		
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)	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)	Latin Name	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)	Latin Name
horse	Common Name	horse	Common Name
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms	Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	Synonyms
species	Rank	species	Rank
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	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	Lineage
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	Parent	Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	Parent
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	NCBI Taxonomy ID	9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	NCBI Taxonomy ID
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

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

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](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](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding~#gephebase-summary-title))

Aberration Type

Deletion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion~#gephebase-summary-title))

Deletion Size

100-999 bp

Molecular Details of the Mutation

628 bp deletion encompassing all of exon 7 of SLC24A5 (c.875-340_1081+82del)

Experimental Evidence

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

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; Grahm 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, MFS12, 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](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](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>

