GEPHE SUMMARY

Gephebase Gene

Pax3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Pax3^#gephebase-summary-title)

GepheID

GP00002253

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

Trait State in Taxon A

Equus caballus - Appaloosa

Trait State in Taxon B

Equus caballus - white spotted Appaloosa ; leucism with deafness ; no homozygotes found

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

PAX3

Synonyms

WS1; WS3; CDHS; HUP2

String

9606.ENSP00000375921 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000375921)

Sequence Similarities

Belongs to the paired homeobox family.

GO - Molecular Function

GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)

UniProtKB Homo sapiens

P23760 (http://www.uniprot.org/uniprot/P23760)

GenebankID or UniProtKB

()

GO:0071837 : HMG box domain binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0071837)

GO - Biological Process

GO:0007399 : nervous system development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007399)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0009887 : animal organ morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0009887)
GO:0006915 : apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006915)
GO:0006366 : transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0006366)
GO:0007605 : sensory perception of sound
(https://www.ebi.ac.uk/QuickGO/term/GO:0007605)
GO:0007517 : muscle organ development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007517)

GO - Cellular Component

GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0000790 : nuclear chromatin (https://www.ebi.ac.uk/QuickGO/term/GO:0000790)

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.95C>G p.Pro32Arg

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Pro	Arg	32

Main Reference

Novel variants in the KIT and PAX3 genes in horses with white-spotted coat colour phenotypes. (2013) (https://pubmed.ncbi.nlm.nih.gov/23659293)

Authors

Hauswirth R; Jude R; Haase B; Bellone RR; Archer S; Holl H; Brooks SA; Tozaki T; Penedo MC; Rieder S; Leeb T

Abstract

Variants in the EDNRB, KIT, MITF, PAX3 and TRPM1 genes are known to cause white spotting phenotypes in horses, which can range from the common white markings up to completely white horses. In this study, we investigated these candidate genes in 169 horses with white spotting phenotypes not explained by the previously described variants. We identified a novel missense variant, PAX3:p.Pro32Arg, in Appaloosa horses with a splashed white phenotype in addition to their leopard complex spotting patterns. We also found three novel variants in the KIT gene. The splice site variant c.1346+1G>A occurred in a Swiss Warmblood horse with a pronounced depigmentation phenotype. The missense variant p.Tyr441Cys was present in several part-bred Arabians with sabino-like depigmentation phenotypes. Finally, we provide evidence suggesting that the common and widely distributed KIT:p.Arg682His variant has a very subtle white-increasing effect, which is much less pronounced than the effect of the other described KIT variants. We termed the new KIT variants W18-W20 to provide a simple and unambiguous nomenclature for future genetic testing applications.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphtalmia-associated transcription factor, PMEL17, SLC24A, 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=^Pax3^/and+Taxon ID=^9796^/or+Gene Gephebase=^Pax3^/and+Taxon ID=^9796^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @AllelicSeries https://omia.org/OMIA001688/9796/