GEPHE SUMMARY

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

GepheID
GP00002252

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 - Quarter Horse

Trait State in Taxon B
Equus caballus - white spotted Quarter Horse ; 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

UniProtKB Homo sapiens
P23760 (http://www.uniprot.org/uniprot/P23760)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
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.209G>A p.C70Y

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	Cys	Tyr	70

Main Reference

Mutations in MITF and PAX3 cause "splashed white" and other white spotting phenotypes in horses. (2012) (https://pubmed.ncbi.nlm.nih.gov/22511888)

Authors

Hauswirth R; Haase B; Blatter M; Brooks SA; Burger D; DrÃ¶gemÃ¼ller C; Gerber V; Henke D; Janda J; Jude R; Magdesian KG; Matthews JM; Poncet PA; Svansson V; Tozaki T; Wilkinson-White L; Penedo MC; Rieder S; Leeb T

Abstract

During fetal development neural-crest-derived melanoblasts migrate across the entire body surface and differentiate into melanocytes, the pigment-producing cells. Alterations in this precisely regulated process can lead to white spotting patterns. White spotting patterns in horses are a complex trait with a large phenotypic variance ranging from minimal white markings up to completely white horses. The "splashed white" pattern is primarily characterized by an extremely large blaze, often accompanied by extended white markings at the distal limbs and blue eyes. Some, but not all, splashed white horses are deaf. We analyzed a Quarter Horse family segregating for the splashed white coat color. Genome-wide linkage analysis in 31 horses gave a positive LOD score of 1.6 in a region on chromosome 6 containing the PAX3 gene. However, the linkage data were not in agreement with a monogenic inheritance of a single fully penetrant mutation. We sequenced the PAX3 gene and identified a missense mutation in some, but not all, splashed white Quarter Horses. Genome-wide association analysis indicated a potential second signal near MITF. We therefore sequenced the MITF gene and found a 10 bp insertion in the melanocyte-specific promoter. The MITF promoter variant was present in some splashed white Quarter Horses from the studied family, but also in splashed white horses from other horse breeds. Finally, we identified two additional non-synonymous mutations in the MITF gene in unrelated horses with white spotting phenotypes. Thus, several independent mutations in MITF and PAX3 together with known variants in the EDNRB and KIT genes explain a large proportion of horses with the more extreme white spotting phenotypes.

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/