

GEPHE SUMMARY

<p>Pax3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=[^]Pax3[^]#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002252</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=[^]Morphology[^]#gephebase-summary-title)</p> <p>Coloration (white-spotting ; leucism) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration (white-spotting ; leucism)<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=[^]Coloration (white-spotting ; leucism)[^]#gephebase-summary-title)</p> <p>Equus caballus - Quarter Horse</p> <p>Equus caballus - white spotted Quarter Horse ; leucism with deafness ; no homozygotes found</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=[^]Domesticated[^]#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Equus caballus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Equus caballus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Equus caballus[^]#gephebase-summary-title)</p> <p>Common Name</p> <p>horse</p> <p>Synonyms</p> <p>Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)</p> <p>NCBI Taxonomy ID</p> <p>9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)</p> <p>is Taxon A an Infrappecies?</p> <p>No</p>	<p>Equus caballus (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Equus caballus<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Equus caballus[^]#gephebase-summary-title)</p> <p>Common Name</p> <p>horse</p> <p>Synonyms</p> <p>Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35510)</p> <p>NCBI Taxonomy ID</p> <p>9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9796)</p> <p>is Taxon B an Infrappecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>PAX3</p> <p>WS1; WS3; CDHS; HUP2</p> <p>9606.ENSP00000375921 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000375921)</p> <p>Belongs to the paired homeobox family.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P23760 (http://www.uniprot.org/uniprot/P23760)</p> <p>0</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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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>)
 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>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>) Presumptive Null
 Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title>) Molecular Type
 SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>) Aberration Type
 Nonsynonymous SNP Coding Change
 c.209G>A p.C70Y Molecular Details of the Mutation
 Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title>) Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Tyr	70

Mutations in MITF and PAX3 cause "splashed white" and other white spotting phenotypes in horses. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22511888>) Main Reference
 Hauswirth R; Haase B; Blatter M; Brooks SA; Burger D; DrÄqgemÄ¼ller 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 Authors

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

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Microphthalmia-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 Genes
 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>) Related Haplotypes

EXTERNAL LINKS

COMMENTS

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