GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00002324

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Equus caballus

Trait State in Taxon B

American Paint Horse with white spotting / blue eyes / deafenss

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?

Yes

Taxon B Description

-

GENOTYPIC CHANGE

Generic Gene Name

Mitf

Synonyms

Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit

String

10090.ENSMUSP00000044938 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938)

Sequence Similarities

Belongs to the MiT/TFE family.

GO - Molecular Function

GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)
GO:0003700 : DNA-binding transcription factor activity

UniProtKB Mus musculus

Q08874 (http://www.uniprot.org/uniprot/Q08874)

GenebankID or UniProtKB

()

(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:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0003682 : chromatin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003682)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA
polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0070888 : E-box binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070888)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-
specific binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003705)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006357 : regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0006355 : regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0006355)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0045893 : positive regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0006351 : transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0006351)
GO:0045165 : cell fate commitment (https://www.ebi.ac.uk/QuickGO/term/GO:0045165)
GO:0010468 : regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043010 : camera-type eye development
(https://www.ebi.ac.uk/QuickGO/term/GO:0043010)
GO:0030316 : osteoclast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030316)
GO:0042127 : regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0042127)
GO:0046849 : bone remodeling (https://www.ebi.ac.uk/QuickGO/term/GO:0046849)
GO:0044336 : canonical Wnt signaling pathway involved in negative regulation of apoptotic
process (https://www.ebi.ac.uk/QuickGO/term/GO:0044336)
GO:0030336 : negative regulation of cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0030336)
GO:2000144 : positive regulation of DNA-templated transcription, initiation
(https://www.ebi.ac.uk/QuickGO/term/GO:2000144)
GO:0065003 : protein-containing complex assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0065003)
GO:0045670 : regulation of osteoclast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045670)
GO:2001141 : regulation of RNA biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:2001141)
GO:0016055 : Wnt signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0016055)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0032991 : protein-containing complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0032991)

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

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

Deletion Size

10-100 kb

Molecular Details of the Mutation

~63-kb deletion spanning exons 6-9

Experimental Evidence

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

Main Reference

Whole-genome sequencing reveals a large deletion in the MITF gene in horses with white spotted coat colour and increased risk of deafness. (2019)
(https://pubmed.ncbi.nlm.nih.gov/30644113)

Authors

Henkel J; Lafayette C; Brooks SA; Martin K; Patterson-Rosa L; Cook D; Jagannathan V; Leeb T

Abstract

White spotting phenotypes in horses are highly valued in some breeds. They are quite variable and may range from the common white markings up to completely white horses. EDNRB, KIT, MITF, PAX3 and TRPM1 represent known candidate genes for white spotting phenotypes in horses. For the present study, we investigated an American Paint Horse family segregating a phenotype involving white spotting and blue eyes. Six of eight horses with the white-spotting phenotype were deaf. We obtained whole-genome sequence data from an affected horse and specifically searched for structural variants in the known candidate genes. This analysis revealed a heterozygous ~63-kb deletion spanning exons 6-9 of the MITF gene (chr16:21Â 503Â 211-21Â 566Â 617). We confirmed the breakpoints of the deletion by PCR and Sanger sequencing. PCR-based genotyping revealed that all eight available affected horses from the family carried the deletion. The finding of an MITF variant fits well with the syndromic phenotype involving both depigmentation and an increased risk for deafness and corresponds to human Waardenburg syndrome type 2A. Our findings will enable more precise genetic testing for depigmentation phenotypes in horses.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, Kit (type III receptor protein-tyrosine kinase), MC1R, MFSD12, Pax3, 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

5 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Microphtalmia-associated transcription factor^/and+Taxon ID=^9796^/or+Gene Gephebase=^Microphtalmia-associated transcription factor^/and+Taxon ID=^9796^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism @HeterozygoteAdvantage @Pleiotropy https://omia.org/OMIA000214/9796/