GEPHE SUMMARY

Gephebase Gene

Kit (type III receptor protein-tyrosine kinase) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002213

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

American Paint Horse; Appaloosa; German Riding Pony; Gipsy; Noriker; Old-Tori; Oldenberg; Quarter Horse; Thoroughbred; Warmblood; Welsh Pony

Trait State in Taxon B

American Paint Horse; Appaloosa; German Riding Pony; Gipsy; Noriker; Old-Tori; Oldenberg; Quarter Horse; Thoroughbred; Warmblood; Welsh Pony - white

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

Kit

Synonyms

W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsco1; Gsfsco5; Gsfsow3; Sl

String

10090.ENSMUSP00000005815 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815)

Sequence Similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.

GO - Molecular Function

GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)

UniProtKB Mus musculus

P05532 (http://www.uniprot.org/uniprot/P05532)

GenebankID or UniProtKB

()

GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0042803 : protein homodimerization activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0002020 : protease binding (https://www.ebi.ac.uk/QuickGO/term/GO:0002020)
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004714)
GO:0004713 : protein tyrosine kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004713)
GO:0019955 : cytokine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019955)
GO:0005020 : stem cell factor receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0005020)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
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:0070374 : positive regulation of ERK1 and ERK2 cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:0035234 : ectopic germ cell programmed cell death
(https://www.ebi.ac.uk/QuickGO/term/GO:0035234)
GO:0035162 : embryonic hemopoiesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035162)
GO:0008584 : male gonad development
(https://www.ebi.ac.uk/QuickGO/term/GO:0008584)
GO:0001541 : ovarian follicle development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001541)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0043406 : positive regulation of MAP kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043406)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0043410 : positive regulation of MAPK cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0043410)
GO:0007283 : spermatogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0007283)
GO:0008360 : regulation of cell shape
(https://www.ebi.ac.uk/QuickGO/term/GO:0008360)
GO:0048070 : regulation of developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048070)
GO:0006468 : protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0006468)
GO:0060326 : cell chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0060326)
GO:0006935 : chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0006935)
GO:0048565 : digestive tract development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048565)
GO:0006954 : inflammatory response
(https://www.ebi.ac.uk/QuickGO/term/GO:0006954)
GO:0019221 : cytokine-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0019221)
GO:0048863 : stem cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048863)
GO:0048066 : developmental pigmentation
(https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0009968 : negative regulation of signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0009968)
GO:0046777 : protein autophosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0046777)
GO:0030218 : erythrocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030218)
GO:0018108 : peptidyl-tyrosine phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0018108)
GO:0097067 : cellular response to thyroid hormone stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0097067)
GO:1904349 : positive regulation of small intestine smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:1904349)
GO:0000187 : activation of MAPK activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0000187)
GO:0046427 : positive regulation of JAK-STAT cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0046427)
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
(https://www.ebi.ac.uk/QuickGO/term/GO:0042531)
GO:0030335 : positive regulation of cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0030335)
GO:0046686 : response to cadmium ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0046686)

GO:0035556 : intracellular signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0035556)
GO:0031532 : actin cytoskeleton reorganization
(https://www.ebi.ac.uk/QuickGO/term/GO:0031532)
GO:0002371 : dendritic cell cytokine production
(https://www.ebi.ac.uk/QuickGO/term/GO:0002371)
GO:0050910 : detection of mechanical stimulus involved in sensory perception of sound
(https://www.ebi.ac.uk/QuickGO/term/GO:0050910)
GO:0050673 : epithelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050673)
GO:0038162 : erythropoietin-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0038162)
GO:0038093 : Fc receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0038093)
GO:0007281 : germ cell development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007281)
GO:0008354 : germ cell migration (https://www.ebi.ac.uk/QuickGO/term/GO:0008354)
GO:0006687 : glycosphingolipid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006687)
GO:0035701 : hematopoietic stem cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0035701)
GO:0030097 : hemopoiesis (https://www.ebi.ac.uk/QuickGO/term/GO:0030097)
GO:0002327 : immature B cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002327)
GO:0038109 : Kit signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0038109)
GO:0030032 : lamellipodium assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0030032)
GO:0002320 : lymphoid progenitor cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002320)
GO:0002551 : mast cell chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0002551)
GO:0032762 : mast cell cytokine production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032762)
GO:0043303 : mast cell degranulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043303)
GO:0060374 : mast cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0060374)
GO:0035855 : megakaryocyte development
(https://www.ebi.ac.uk/QuickGO/term/GO:0035855)
GO:0097326 : melanocyte adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0097326)
GO:0097324 : melanocyte migration (https://www.ebi.ac.uk/QuickGO/term/GO:0097324)
GO:0002573 : myeloid leukocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002573)
GO:0002318 : myeloid progenitor cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0002318)
GO:0043069 : negative regulation of programmed cell death
(https://www.ebi.ac.uk/QuickGO/term/GO:0043069)
GO:1904343 : positive regulation of colon smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:1904343)
GO:0051091 : positive regulation of DNA-binding transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051091)
GO:0048170 : positive regulation of long-term neuronal synaptic plasticity
(https://www.ebi.ac.uk/QuickGO/term/GO:0048170)
GO:0045747 : positive regulation of Notch signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0045747)
GO:0031274 : positive regulation of pseudopodium assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0031274)
GO:0120072 : positive regulation of pyloric antrum smooth muscle contraction
(https://www.ebi.ac.uk/QuickGO/term/GO:0120072)
GO:1905065 : positive regulation of vascular smooth muscle cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:1905065)
GO:1904251 : regulation of bile acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:1904251)
GO:0009314 : response to radiation (https://www.ebi.ac.uk/QuickGO/term/GO:0009314)
GO:0048103 : somatic stem cell division
(https://www.ebi.ac.uk/QuickGO/term/GO:0048103)
GO:0035019 : somatic stem cell population maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:0035019)
GO:0007286 : spermatid development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007286)
GO:0030217 : T cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030217)
GO:0043586 : tongue development (https://www.ebi.ac.uk/QuickGO/term/GO:0043586)
GO:0008542 : visual learning (https://www.ebi.ac.uk/QuickGO/term/GO:0008542)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)

GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)
GO:0009898 : cytoplasmic side of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0009898)
GO:0009897 : external side of plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0009897)
GO:0001669 : acrosomal vesicle (https://www.ebi.ac.uk/QuickGO/term/GO:0001669)
GO:0005911 : cell-cell junction (https://www.ebi.ac.uk/QuickGO/term/GO:0005911)
GO:0042629 : mast cell granule (https://www.ebi.ac.uk/QuickGO/term/GO:0042629)

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.2045G>A p.R682H

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	Arg	His	682

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.

Â© 2013 The Authors, Animal Genetics Â© 2013 Stichting International Foundation for Animal Genetics.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, MC1R, MFSD12, Microphtalmia-associated transcription factor, 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

28 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9796^/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9796^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA000209/9796/