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

GP00002223

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

German Riding Pony

Trait State in Taxon B

German Riding 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)
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)
GO:0042803 : protein homodimerization activity

UniProtKB Mus musculus

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

GenebankID or UniProtKB

()

(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

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

1-10 kb

Molecular Details of the Mutation

g.79;579;925-79;581;197del 1273bp deletion

Experimental Evidence

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

Main Reference

A novel KIT deletion variant in a German Riding Pony with white-spotting coat colour phenotype. (2019) (https://pubmed.ncbi.nlm.nih.gov/31463981)

Authors

Hug P; Jude R; Henkel J; Jagannathan V; Leeb T

Abstract

White spotting phenotypes in horses may be caused by developmental alterations impairing melanoblast differentiation, survival, migration and/or proliferation. Candidate genes for white-spotting phenotypes in horses include EDNRB, KIT, MITF, PAX3 and TRPM1. We investigated a German Riding Pony with a sabino-like phenotype involving extensive white spots on the body together with large white markings on the head and almost completely white legs. We obtained whole genome sequence data from this horse. The analysis revealed a heterozygous 1273-bp deletion spanning parts of intron 2 and exon 3 of the equine KIT gene (Chr3: 79 579 925-79 581 197). We confirmed the breakpoints of the deletion by PCR and Sanger sequencing. Knowledge of the functional impact of similar KIT variants in horses and other species suggests that this deletion represents a plausible candidate causative variant for the white-spotting phenotype. We propose the designation W28 for the mutant allele.

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