GEPHE SUMMARY

Gephebase Gene
Kit (type III receptor protein-tyrosine kinase)

GephelD
GP00002219

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

Trait State in Taxon A
Thoroughbred

Trait State in Taxon B
Thoroughbred - white

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Equus caballus

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)

NCBI Taxonomy ID
9796

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Equus caballus

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)

NCBI Taxonomy 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

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
GO:0005524 : ATP binding
GO:0042803 : protein homodimerization activity
GO:0046872 : metal ion binding
GO:0002020 : protease binding
GO:0004714 : transmembrane receptor protein tyrosine kinase activity
GO:0004713 : protein tyrosine kinase activity
GO:0019955 : cytokine binding
GO:0005020 : stem cell factor receptor activity

UniProtKB Mus musculus
P05532

GenebankID or UniProtKB

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
GO:0030154 : cell differentiation
GO:0043473 : pigmentation
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
GO:0035234 : ectopic germ cell programmed cell death
GO:0035162 : embryonic hemopoiesis
GO:0008584 : male gonad development
GO:0001541 : ovarian follicle development
GO:0008284 : positive regulation of cell proliferation
GO:0043406 : positive regulation of MAP kinase activity
GO:0010628 : positive regulation of gene expression
GO:0043410 : positive regulation of MAPK cascade
GO:0007283 : spermatogenesis
GO:0008360 : regulation of cell shape
GO:0048070 : regulation of developmental pigmentation
GO:0006468 : protein phosphorylation
GO:0060326 : cell chemotaxis
GO:0006935 : chemotaxis
GO:0048565 : digestive tract development
GO:0006954 : inflammatory response
GO:0019221 : cytokine-mediated signaling pathway
GO:0048863 : stem cell differentiation
GO:0048066 : developmental pigmentation
GO:0030318 : melanocyte differentiation
GO:0009968 : negative regulation of signal transduction
GO:0046777 : protein autophosphorylation
GO:0030218 : erythrocyte differentiation
GO:0018108 : peptidyl-tyrosine phosphorylation
GO:0097067 : cellular response to thyroid hormone stimulus
GO:1904349 : positive regulation of small intestine smooth muscle contraction
GO:0000187 : activation of MAPK activity
GO:0046427 : positive regulation of JAK-STAT cascade
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
GO:0030335 : positive regulation of cell migration
GO:0046686 : response to cadmium ion
GO:0035556 : intracellular signal transduction
GO:0031532 : actin cytoskeleton reorganization
GO:0002371 : dendritic cell cytokine production
GO:0050910 : detection of mechanical stimulus involved in sensory perception of sound
GO:0050673 : epithelial cell proliferation
GO:0038162 : erythropoietin-mediated signaling pathway
GO:0038093 : Fc receptor signaling pathway
GO:0007281 : germ cell development
GO:0008354 : germ cell migration
GO:0006687 : glycosphingolipid metabolic process
GO:0035701 : hematopoietic stem cell migration
GO:0030097 : hemopoiesis
GO:0002327 : immature B cell differentiation
GO:0038109 : Kit signaling pathway
GO:0030032 : lamellipodium assembly
GO:0002320 : lymphoid progenitor cell differentiation
GO:0002551 : mast cell chemotaxis
GO:0032762 : mast cell cytokine production
GO:0043303 : mast cell degranulation
GO:0060374 : mast cell differentiation
GO:0035855 : megakaryocyte development
GO:0097326 : melanocyte adhesion
GO:0097324 : melanocyte migration
GO:0002573 : myeloid leukocyte differentiation
GO:0002318 : myeloid progenitor cell differentiation
GO:0043069 : negative regulation of programmed cell death
GO:1904343 : positive regulation of colon smooth muscle contraction
GO:0051091 : positive regulation of DNA-binding transcription factor activity
GO:0048170 : positive regulation of long-term neuronal synaptic plasticity
GO:0045747 : positive regulation of Notch signaling pathway
GO:0031274 : positive regulation of pseudopodium assembly
GO:0120072 : positive regulation of pyloric antrum smooth muscle contraction
GO:1905065 : positive regulation of vascular smooth muscle cell differentiation
GO:1904251 : regulation of bile acid metabolic process
GO:0009314 : response to radiation
GO:0048103 : somatic stem cell division
GO:0035019 : somatic stem cell population maintenance
GO:0007286 : spermatid development
GO:0030217 : T cell differentiation
GO:0043586 : tongue development
GO:0008542 : visual learning

GO - Cellular Component

GO:0005886 : plasma membrane
GO:0005737 : cytoplasm
GO:0005887 : integral component of plasma membrane
GO:0043235 : receptor complex
GO:0005615 : extracellular space

GO:0009986 : cell surface
GO:0009898 : cytoplasmic side of plasma membrane
GO:0009897 : external side of plasma membrane
GO:0001669 : acrosomal vesicle
GO:0005911 : cell-cell junction
GO:0042629 : mast cell granule

Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

Deletion Size
1-10 kb

Molecular Details of the Mutation
g.77;740;239_77;742;136del1898insTATAT ~1.9-kb deletion

Experimental Evidence
Candidate Gene

Main Reference
Whole genome sequencing reveals a novel deletion variant in the KIT gene in horses with white spotted coat colour phenotypes. (2017)

Authors
DÃ¾rig N; Jude R; Holl H; Brooks SA; Lafayette C; Jagannathan V; Leeb T

Abstract
White spotting phenotypes in horses can range in severity from the common white markings up to completely white horses. EDNRB, KIT, MITF, PAX3 and TRPM1 represent known candidate genes for such phenotypes in horses. For the present study, we re-investigated a large horse family segregating a variable white spotting phenotype, for which conventional Sanger sequencing of the candidate genes' individual exons had failed to reveal the causative variant. 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 ~1.9-kb deletion spanning exons 10-13 of the KIT gene (chr3:77,740,239_77,742,136del1898insTATAT). In continuity with previously named equine KIT variants we propose to designate the newly identified deletion variant W22. We had access to 21 horses carrying the W22 allele. Four of them were compound heterozygous W20/W22 and had a completely white phenotype. Our data suggest that W22 represents a true null allele of the KIT gene, whereas the previously identified W20 leads to a partial loss of function. These findings will enable more precise genetic testing for depigmentation phenotypes in horses.

Additional References

RELATED GEPHE

Related Genes
13 (Agouti, Endothelin receptor B, MC1R, MFSD12, Microphtalmia-associated transcription factor, Pax3, PMEL17, SLC24A, SLC36A1, SLC45A2=MATP, STX17, T-box transcription factor (TBX3), TRPM1)
Related Haplotypes
28

EXTERNAL LINKS

COMMENTS

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