

## GEPHE SUMMARY

|   |   |                                    |
|---|---|------------------------------------|
| <p>Kit (type III receptor protein-tyrosine kinase) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit (type III receptor protein-tyrosine kinase)^#gephebase-summary-title</a>)</p> <p>Published</p> | <p>Gephebase Gene</p> <p>GP00002210</p> <p>Martin</p> <p>Entry Status</p> | <p>GepheID</p> <p>Main curator</p> |
|---|---|------------------------------------|

## PHENOTYPIC CHANGE

|   |   |   |  |   |  |
|---|---|---|--|---|--|
| <p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Coloration (coat) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^#gephebase-summary-title</a>)</p> <p>Miniature Horse; Quarter Horse</p> <p>Miniature Horse; Quarter Horse - white</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a>)</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>Taxon A</p> <p>Equus caballus<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+caballus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus caballus^#gephebase-summary-title</a>)</p> <p>horse</p> <p>Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758</p> <p>species</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>Equus () - (Rank: subgenus)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a>)</p> <p>9796<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a>)</p> <p>No</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Infrappecies?</p> | <p>Taxon B</p> <p>Equus caballus<br/>(<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus+caballus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Equus caballus^#gephebase-summary-title</a>)</p> <p>horse</p> <p>Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758</p> <p>species</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>Equus () - (Rank: subgenus)<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510</a>)</p> <p>9796<br/>(<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796</a>)</p> <p>No</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Infrappecies?</p> |
|---|---|---|--|---|--|

## GENOTYPIC CHANGE

|   |  |  |
|---|--|--|
| <p>Kit</p> <p>W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; SI</p> <p>10090.ENSMUSP00000005815<br/>(<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815</a>)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.</p> | <p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> | <p>P05532 (<a href="http://www.uniprot.org/uniprot/P05532">http://www.uniprot.org/uniprot/P05532</a>)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p> |
| <p>GO:0004888 : transmembrane signaling receptor activity<br/>(<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004888">https://www.ebi.ac.uk/QuickGO/term/GO:0004888</a>)</p> <p>GO:0005524 : ATP binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005524">https://www.ebi.ac.uk/QuickGO/term/GO:0005524</a>)</p> <p>GO:0042803 : protein homodimerization activity</p>   |  |  |

(<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

-

Molecular Details of the Mutation

c.2472+5G>C (putative splicing mutation)

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 | -       | -       | -        |

Main Reference

Five novel KIT mutations in horses with white coat colour phenotypes. (2011) (https://pubmed.ncbi.nlm.nih.gov/21554354)

Authors

Haase B; Rieder S; Tozaki T; Hasegawa T; Penedo MC; Jude R; Leeb T

Abstract

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