GEPHE SUMMARY

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

Entry Status
Published

GepheID
GP00002218

Main curator
Martin

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
Italian Trotter

Trait State in Taxon B
Italian Trotter - white

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://recette.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Equus caballus (https://recette.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://recette.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

No (https://recette.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://recette.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

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

Deletion Size

-

Molecular Details of the Mutation

g.77736559C>T (putative splicing mutation)

Experimental Evidence

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

Main Reference

Splicing site disruption in the KIT gene as strong candidate for white dominant phenotype in an Italian Trotter. (2017) (https://pubmed.ncbi.nlm.nih.gov/28856698)

Authors

Capomaccio S; Milanesi M; Nocelli C; Giontella A; Verini-Supplizi A; Branca M; Silvestrelli M; Cappelli K

Abstract

-

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) (https://recette.gephebase.org/search-criteria?/or+Taxon ID=^9796^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

28 (https://recette.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

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