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

GP00001340

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Donkey ; wild type

Trait State in Taxon B

Donkey - Dominant white ; pink skin and white coat (but dark eyes)

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 asinus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus asinus^#gephebase-summary-title)

Common Name

ass

Synonyms

ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Asinus

Parent

Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID

9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Equus asinus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Equus asinus^#gephebase-summary-title)

Common Name

ass

Synonyms

ass; African ass; African wild ass; Somali wild ass; domestic ass; donkey

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; Asinus

Parent

Asinus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 35508)

NCBI Taxonomy ID

9793 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9793)

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)

UniProtKB Mus musculus

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

GenebankID or UniProtKB

()

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.662A>C p.Tyr221Ser

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	Tyr	Ser	221

Main Reference

Two variants in the KIT gene as candidate causative mutations for a dominant white and a white spotting phenotype in the donkey. (2015) (https://pubmed.ncbi.nlm.nih.gov/25818843)

Authors

Haase B; Rieder S; Leeb T

Abstract

White spotting phenotypes have been intensively studied in horses, and although similar phenotypes occur in the donkey, little is known about the molecular genetics underlying these patterns in donkeys. White spotting in donkeys can range from only a few white areas to almost complete depigmentation and is characterised by a loss of pigmentation usually progressing from a white spot in the hip area. Completely white-born donkeys are rare, and the phenotype is characterised by the complete absence of pigment resulting in pink skin and a white coat. A dominant mode of inheritance has been demonstrated for spotting in donkeys. Although the mode of inheritance for the completely white phenotype in donkeys is not clear, the phenotype shows similarities to dominant white in horses. As variants in the KIT gene are known to cause a range of white phenotypes in the horse, we investigated the KIT gene as a potential candidate gene for two phenotypes in the donkey, white spotting and white. A mutation analysis of all 21 KIT exons identified a missense variant in exon 4 (c.662A>C; p.Tyr221Ser) present only in a white-born donkey. A second variant affecting a splice donor site (c.1978+2T>A) was found exclusively in donkeys with white spotting. Both variants were absent in 24 solid-coloured controls. To the authors' knowledge, this is the first study investigating genetic mechanisms underlying white phenotypes in donkeys. Our results suggest that two independent KIT alleles are probably responsible for white spotting and white in donkeys.

Additional References

RELATED GEPHE

Related Genes

3 (Agouti (ASIP), MC1R, tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9793^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9793^/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9793^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries @HeterozygousAdvantage ; https://omia.org/OMIA000209/9793/