

GEPHE SUMMARY

Kit (type III receptor protein-tyrosine kinase) (<a +kit+(type+iii+receptor+protein-tyrosine+kinase)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase=" Kit (type III receptor protein-tyrosine kinase) <a +kit+(type+iii+receptor+protein-tyrosine+kinase)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase=">^ <a +kit+(type+iii+receptor+protein-tyrosine+kinase)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene Gephebase=">#gephebase-summary-title)	Gephebase Gene	GepheID
	GP00000515	
	Martin	Main curator
Published	Entry Status	

PHENOTYPIC CHANGE

		Trait Category	
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category=" Morphology [^] <a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">#gephebase-summary-title)		Trait	
Coloration (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait=" Coloration (coat) <a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">#gephebase-summary-title)		Trait State in Taxon A	
Equus caballus		Trait State in Taxon B	
Equus caballus - Sabino and others		Ancestral State	
Taxon A		Taxonomic Status	
Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status=" Domesticated [^] <a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">#gephebase-summary-title)			
Taxon A		Taxon B	
Equus caballus (<a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=" Equus caballus [^] <a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#gephebase-summary-title)		Equus caballus (<a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=" Equus caballus [^] <a +equus+caballus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">#gephebase-summary-title)	
Common Name		Common Name	
horse		horse	
Synonyms		Synonyms	
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
Rank		Rank	
species		species	
Lineage		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		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		Parent	
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)		Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)		9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Appaloosa; Haflinger; Lipizzan; Noriker; Quarter Horse	

GENOTYPIC CHANGE

		Generic Gene Name	
Kit		P05532 (http://www.uniprot.org/uniprot/P05532)	UniProtKB Mus musculus
Synonyms		GenebankID or UniProtKB	
W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; SI		AEU10902 (https://www.ncbi.nlm.nih.gov/nuccore/AEU10902)	
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>)
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

T->A in intron 16; leads to skipping of exon 17

Experimental Evidence

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Exon skipping in the KIT gene causes a Sabino spotting pattern in horses. (2005) (<https://pubmed.ncbi.nlm.nih.gov/16284805>)

Authors

Brooks SA; Bailey E

Abstract

Sabino (SB) is a white spotting pattern in the horse characterized by white patches on the face, lower legs, or belly, and interspersed white hairs on the midsection. Based on comparable phenotypes in humans and pigs, the KIT gene was investigated as the origin of the Sabino phenotype. In this article we report the genetic basis of one type of Sabino spotting pattern in horses that we call Sabino 1, with the alleles represented by the symbols SB1 and sb1. Transcripts of KIT were characterized by reverse transcriptase polymerase chain reaction (RT-PCR) and sequencing cDNA from horses with the genotypes SB1/SB1, SB1/sb1, and sb1/sb1. Horses with the Sabino 1 trait produced a splice variant of KIT that did not possess exon 17. Genomic DNA sequencing of KIT revealed a single nucleotide polymorphism (SNP) caused by a base substitution for T with A in intron 16, 1037 bases following exon 16. The SNP associated with SB1 was designated KI16+1037A. This substitution eliminated a MnlI restriction site and allowed the use of PCR-RFLP to characterize individuals for this base change. Complete linkage was observed between this SNP and Sabino 1 in the Tennessee Walking Horse families (LOD = 9.02 for Theta = 0). Individual horses from other breeds were also tested. All five horses homozygous for this SNP were white, and all 68 horses with one copy of this SNP either exhibited the Sabino 1 phenotype or were multipatterned. Some multipatterned individuals appeared white due to the additive effect of white spotting patterns. However, 13 horses with other Sabino-type patterns did not have this SNP. Based on these results we propose the following: (1) this SNP, found within intron 16, is responsible for skipping of exon 17 and the SB1 phenotype, (2) the White and Sabino phenotypes are heterogeneous and this mechanism is not the only way to produce the pattern described as “Sabino” or “White,” and (3) homozygosity for SB1 results in a complete or nearly completely white phenotype.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti, Endothelin receptor B, MC1R, MFSD12, Microphthalmia-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](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

@Splicing @AllelicSeries <https://omia.org/OMIA000209/9796/>

