

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)+"#gephebase-summary-title)	Gephebase Gene	GepheID
	GP00000521	
	Martin	Main curator
Published	Entry Status	

PHENOTYPIC CHANGE

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+"#gephebase-summary-title)		Trait Category		
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)+"#gephebase-summary-title)		Trait		
Vulpes vulpes		Trait State in Taxon A		
Vulpes vulpes - platinum (farm-bred)		Trait State in Taxon B		
Taxon A		Ancestral State		
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+"#gephebase-summary-title)		Taxonomic Status		
Taxon A		Taxon B		
Vulpes vulpes (<a +vulpes+vulpes+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Vulpes+vulpes+"#gephebase-summary-title)		Vulpes vulpes (<a +vulpes+vulpes+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Vulpes+vulpes+"#gephebase-summary-title)		
red fox		red fox		
Canis vulpes; red fox; silver fox		Canis vulpes; red fox; silver fox		
species		species		
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Vulpes		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Vulpes		
Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9625)		Vulpes () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9625)		
9627 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9627)		9627 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9627)		
No		Yes		
		Taxon B Description		
		Vulpes vulpes - platinum (farm-bred)		

GENOTYPIC CHANGE

Kit		Generic Gene Name	UniProtKB Mus musculus
W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfsc03; SI		Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000005815 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815)		String	KM196092 (https://www.ncbi.nlm.nih.gov/nuccore/KM196092)
Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.		Sequence Similarities	
GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)		GO - Molecular Function	
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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)	Aberration Type
-	Deletion Size
splice mutation; G to A substitution in the first nucleotide of intron 17; leads to skipping of exon 17.	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
Platinum coat color in red fox (<i>Vulpes vulpes</i>) is caused by a mutation in an autosomal copy of KIT. (2015) (https://pubmed.ncbi.nlm.nih.gov/25662789)	Main Reference
Johnson JL; Kozysa A; Kharlamova AV; Gulevich RG; Perelman PL; Fong HW; Vladimirova AV; Oskina IN; Trut LN; Kukekova AV	Authors
The red fox (<i>Vulpes vulpes</i>) demonstrates a variety of coat colors including platinum, a common phenotype maintained in farm-bred fox populations. Foxes heterozygous for the platinum allele have a light silver coat and extensive white spotting, whereas homozygosity is embryonic lethal. Two KIT transcripts were identified in skin cDNA from platinum foxes. The long transcript was identical to the KIT transcript of silver foxes, whereas the short transcript, which lacks exon 17, was specific to platinum. The KIT gene has several copies in the fox genome: an autosomal copy on chromosome 2 and additional copies on the B chromosomes. To identify the platinum-specific KIT sequence, the genomes of one platinum and one silver fox were sequenced. A single nucleotide polymorphism (SNP) was identified at the first nucleotide of KIT intron 17 in the platinum fox. In platinum foxes, the A allele of the SNP disrupts the donor splice site and causes exon 17, which is part of a segment that encodes a conserved tyrosine kinase domain, to be skipped. Complete cosegregation of the A allele with the platinum phenotype was confirmed by linkage mapping (LOD 25.59). All genotyped farm-bred platinum foxes from Russia and the US were heterozygous for the SNP (A/G), whereas foxes with different coat colors were homozygous for the G allele. Identification of the platinum mutation suggests that other fox white-spotting phenotypes, which are allelic to platinum, would also be caused by mutations in the KIT gene.	Abstract
Â© 2015 Stichting International Foundation for Animal Genetics.	Additional References

RELATED GEPHE

2 (Agouti, MC1R) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9627^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9627^/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9627^#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Splicing @HeterozygoteAdvantage Dominance: All genotyped farm-bred platinum foxes from Russia and the US were heterozygous for the SNP (A/G); Parallelism: The loss of KIT exon 17 has been shown to cause the Sabino spotting pattern in horses and in combination with a duplication of the normal copy of the KIT gene to cause a dominant white phenotype in pigs. Interestingly; the mutation in pigs that leads to the skipping of exon 17 is the same as in the platinum fox