

## GEPHE SUMMARY

Kit ( <a +kit+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Kit+"#gephebase-summary-title</a> )	Gephebase Gene	GP00002228	GepheID
Published	Entry Status	Martin	Main curator

## 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</a> )	Trait Category		
Coloration (coat ; white-spotting) ( <a +coloration+(coat+;+white-spotting)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat+;+white-spotting)+"#gephebase-summary-title</a> )	Trait		
WT coat	Trait State in Taxon A		
white spotting in heterozygotes ; homozygous lethal	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</a> )	Taxonomic Status		
	Taxon A		Taxon B
Canis lupus ( <a +canis+lupus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+"#gephebase-summary-title</a> )	Latin Name	Canis lupus familiaris ( <a +canis+lupus+familiaris+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+familiaris+"#gephebase-summary-title</a> )	Latin Name
gray wolf	Common Name	dog	Common Name
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758;	Synonyms
species	Rank	Canis lupus familiaris Linnaeus, 1758	Rank
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; Canis	Lineage	subspecies	Lineage
Canis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611</a> )	Parent	Canis lupus (gray wolf) - (Rank: species) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )	Parent
9612 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612</a> )	NCBI Taxonomy ID	9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

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

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

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Yes^#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=^Coding^#gephebase-summary-title))

Aberration Type

Insertion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=^Insertion^#gephebase-summary-title))

Insertion Size

1-9 bp

Molecular Details of the Mutation

a 1-bp insertion of an adenine 70 bases downstream of the beginning of exon 2 resulting in frameshift

Experimental Evidence

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

Main Reference

A de novo mutation in KIT causes white spotting in a subpopulation of German Shepherd dogs. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23134432>)

Authors

Wong AK; Ruhe AL; Robertson KR; Loew ER; Williams DC; Neff MW

Abstract

Although variation in the KIT gene is a common cause of white spotting among domesticated animals, KIT has not been implicated in the diverse white spotting observed in the dog. Here, we show that a loss-of-function mutation in KIT recapitulates the coat color phenotypes observed in other species. A spontaneous white spotting observed in a pedigree of German Shepherd dogs was mapped by linkage analysis to a single locus on CFA13 containing KIT (pairwise LOD = 15). DNA sequence analysis identified a novel 1-bp insertion in the second exon that co-segregated with the phenotype. The expected frameshift and resulting premature stop codons predicted a severely truncated c-Kit receptor with presumably abolished activity. No dogs homozygous for the mutation were recovered from multiple intercrosses ( $P = 0.01$ ), suggesting the mutation is recessively embryonic lethal. These observations are consistent with the effects of null alleles of KIT in other species.

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Additional References

## RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103), RALY (hnRNP associated with lethal yellow)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=^9612^/and+Trait=Coloration/or+Taxon ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9612^/and+Trait=Coloration/or+Taxon+ID=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

<https://omia.org/OMIA001737/9615/> @HeterozygoteAdvantage homozygous lethal