

## GEPHE SUMMARY

<p>Kit (type III receptor protein-tyrosine kinase) (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+(type+III+receptor+protein-tyrosine+kinase)^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002227</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Morphology (<a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a>)</p> <p>Coloration (coat) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title</a>)</p> <p>WT coat</p> <p>Roan coat ; black phenotype of the Korean Native pig</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title</a>)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p><i>Sus scrofa domesticus</i> (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic pig</p> <p>Synonyms</p> <p><i>Sus domestica</i>; <i>Sus domesticus</i>; <i>Sus scrofa domestica</i>; domestic pig</p> <p>Rank</p> <p>subspecies</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; <i>Sus scrofa</i></p> <p>Parent</p> <p><i>Sus scrofa</i> (pig) - (Rank: species) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823</a>)</p> <p>NCBI Taxonomy ID</p> <p>9825 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825</a>)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p><i>Sus scrofa domesticus</i> (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title</a>)</p> <p>Common Name</p> <p>domestic pig</p> <p>Synonyms</p> <p><i>Sus domestica</i>; <i>Sus domesticus</i>; <i>Sus scrofa domestica</i>; domestic pig</p> <p>Rank</p> <p>subspecies</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; <i>Sus scrofa</i></p> <p>Parent</p> <p><i>Sus scrofa</i> (pig) - (Rank: species) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823</a>)</p> <p>NCBI Taxonomy ID</p> <p>9825 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825</a>)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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## GENOTYPIC CHANGE

<p>Kit</p> <p>W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfscow3; SI</p> <p>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>)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P05532 (<a href="http://www.uniprot.org/uniprot/P05532">http://www.uniprot.org/uniprot/P05532</a>)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>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>)</p> <p>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>)</p> <p>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>)</p>		

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 ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null+No))

Molecular Type

Cis-regulatory ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type+Cis-regulatory))

Aberration Type

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

Deletion Size

-

Molecular Details of the Mutation

U(26) repeat in intron 5 of the KIT gene with a single G interruption likely to mediate skipping of exon 5 of the gene in some tissues including skin ; the WT allele has an U(27) repeat interrupted by 5 G

Experimental Evidence

Linkage Mapping ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence+Linkage+Mapping))

Main Reference

Novel alternative splicing by exon skipping in KIT associated with whole-body roan in an intercrossed population of Landrace and Korean Native pigs. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21749430>)

Authors

Lim HT; Zhong T; Cho IC; Seo BY; Kim JH; Lee SS; Ko MS; Park HB; Kim BW; Lee JH; Jeon JT

Abstract

The KIT locus has been suggested to be a strong candidate region linked with whole-body roan in the F(2) population produced by intercrosses between Landrace and Korean Native pigs. In this manuscript, we report the finding of a novel alternative splicing event in the porcine KIT gene that results in the skipping of exon 5 in the I(Rn) allele. KIT mRNAs that lack exon 5 were identified in the large intestine and skin, suggesting that the mechanism responsible for the skipping of exon 5 may be tissue specific. A U(26) repeat in intron 5 showed complete linkage (LOD = 11.8) with the roan phenotype and absolute association with the black phenotype of the Korean Native pig (KNP) population samples, inferring that the repeat pattern may alter the complementary base-pairing-mediated looping-out of introns 4 and 5, which may mediate the exon 5-skipping event. Although the sample size in our study was relatively small, we speculate that the R3 allele containing the U(26) repeat is a causative element for the roan phenotype via alternative control of the exon skipping in our roan pedigree.

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Additional References

A sensitive method for detecting variation in copy numbers of duplicated genes. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12952884>)

## RELATED GEPHE

Related Genes

3 (Agouti, MC1R, tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID+9825#/and+Trait+Coloration/and+groupHaplotypes=true#gpepbase-summary-title>)

Related Haplotypes

3 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase+Kit+\(type+III+receptor+protein-tyrosine+kinase\)/and+Taxon+ID+9825/or+Gene+Gephebase+Kit+\(type+III+receptor+protein-tyrosine+kinase\)/and+Taxon+ID+9825#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase+Kit+(type+III+receptor+protein-tyrosine+kinase)/and+Taxon+ID+9825/or+Gene+Gephebase+Kit+(type+III+receptor+protein-tyrosine+kinase)/and+Taxon+ID+9825#gpepbase-summary-title))

## EXTERNAL LINKS

## COMMENTS

@Splicing <https://omia.org/OMIA001216/9823/>