

GEPHE SUMMARY

| | | |
|---|---|------------------------------------|
| <p>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)</p> <p>Published</p> | <p>Gephebase Gene</p> <p>GP00002196</p> <p>Martin</p> <p>Entry Status</p> | <p>GepheID</p> <p>Main curator</p> |
|---|---|------------------------------------|

PHENOTYPIC CHANGE

| | | | |
|---|---|---|---|
| <p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat)^#gephebase-summary-title)</p> <p>Wild Boar</p> <p>Domestic pig - Patch phenotype</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)</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>Taxon B</p> | |
| <p><i>Sus scrofa</i> (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa^#gephebase-summary-title)</p> <p>pig</p> <p>pig; pigs; swine; wild boar; <i>Sus scrofa</i> Linnaeus, 1758; <i>Sus scrofa</i> Linnaeus, 1758; <i>Sus scrofa</i> Linnaeus, 1758</p> <p>species</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</p> <p><i>Sus</i> (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9822)</p> <p>9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> | <p><i>Sus scrofa domesticus</i> (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Sus+scrofa+domesticus^#gephebase-summary-title)</p> <p>domestic pig</p> <p><i>Sus domestica</i>; <i>Sus domestica</i>; <i>Sus scrofa domestica</i>; domestic pig</p> <p>subspecies</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><i>Sus scrofa</i> (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9823)</p> <p>9825 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)</p> <p>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Taxon B Description</p> <p>Domestic pig - Belt phenotype</p> | <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> |

GENOTYPIC CHANGE

| | | | |
|--|--|---|--|
| <p>Kit</p> <p>W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsc01; Gsfsc05; Gsfsc03; SI</p> <p>10090.ENSMUSP00000005815 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000005815)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. CSF-1/PDGF receptor subfamily.</p> <p>GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)</p> <p>GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)</p> | <p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> | <p>P05532 (http://www.uniprot.org/uniprot/P05532)</p> <p>()</p> | <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p> |
|--|--|---|--|

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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

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

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 Size

1-10 kb

Molecular Details of the Mutation

the patch allele comprises a 450kb duplication that includes KIT (roughly in the middle)

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=))

Main Reference

Strong signatures of selection in the domestic pig genome. (2012) (<https://pubmed.ncbi.nlm.nih.gov/23151514>)

Authors

Rubin CJ; Megens HJ; Martinez Barrio A; Maqbool K; Sayyab S; Schwochow D; Wang C; Carlborg Å-; Jern P; JÃ_rgensen CB; Archibald AL; Fredholm M; Groenen MA; Andersson L

Abstract

Domestication of wild boar (*Sus scrofa*) and subsequent selection have resulted in dramatic phenotypic changes in domestic pigs for a number of traits, including behavior, body composition, reproduction, and coat color. Here we have used whole-genome resequencing to reveal some of the loci that underlie phenotypic evolution in European domestic pigs. Selective sweep analyses revealed strong signatures of selection at three loci harboring quantitative trait loci that explain a considerable part of one of the most characteristic morphological changes in the domestic pig--the elongation of the back and an increased number of vertebrae. The three loci were associated with the NR6A1, PLAG1, and LCORL genes. The latter two have repeatedly been associated with loci controlling stature in other domestic animals and in humans. Most European domestic pigs are homozygous for the same haplotype at these three loci. We found an excess of derived nonsynonymous substitutions in domestic pigs, most likely reflecting both positive selection and relaxed purifying selection after domestication. Our analysis of structural variation revealed four duplications at the KIT locus that were exclusively present in white or white-spotted pigs, carrying the Dominant white, Patch, or Belt alleles. This discovery illustrates how structural changes have contributed to rapid phenotypic evolution in domestic animals and how alleles in domestic animals may evolve by the accumulation of multiple causative mutations as a response to strong directional selection.

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="+9823"/and+Trait=Coloration/or+Taxon ID="+9825"/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

Related Haplotypes

3 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase="+Kit \(type III receptor protein-tyrosine kinase\)"/and+Taxon ID="+9823"/or+Gene Gephebase="+Kit \(type III receptor protein-tyrosine kinase\)"/and+Taxon ID="+9825"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=))

EXTERNAL LINKS

COMMENTS

@AllelicSeries ; <https://omia.org/OMIA001743/9823/>