Gephebase Gene
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)
Entry Status
Published

## PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait
Category=^Morphology^\#gephebase-summary-title)
Trait
Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (coat)^\#gephebase-summary-title)

Trait State in Taxon A
Bos bovis
Trait State in Taxon B
Bos bovis - side-colored

Data not curated
Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic
Status=^Domesticated^\#gephebase-summary-title)

## Taxon A

Latin Name
Bos taurus
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus ${ }^{\wedge}$ \#gephebase-summary-title)

Common Name
cattle
Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus
species Rank
ellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; _aurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Bos (oxen, cattle) - (Rank: genus)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)
NCBI Taxonomy ID
9913
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)
is Taxon A an Infraspecies?
No

## GENOTYPIC CHANGE

Kit $\quad$ Generic Gene Name
Synonyms Sy
W; Bs; Fdc; Ssm; SCO1; SCO5; SOW3; CD117; c-KIT; Tr-kit; Gsfsco1; Gsfsco5; Gsfsow3; SI 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

P05532 (http://www.uniprot.org/uniprot/P05532)
UniProtKB Mus musculus

## Taxon B

Latin Name
Bos taurus
(https://www.gephebase.org/search-criteria?/and + Taxon and Synonyms=^Bos taurus^\#gephebase-summary-title)
cattle
Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus
species
Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent
Bos (oxen, cattle) - (Rank: genus)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)
NCBI Taxonomy ID
9913
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)
is Taxon B an Infraspecies?
Yes

Taxon B Description
Belgian Blue Brown Swiss with Cs6 allele
"

GenebankID or UniProtKB

0

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
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GO:0001541 : ovarian follicle development
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GO:0008284 : positive regulation of cell proliferation
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GO:0043406 : positive regulation of MAP kinase activity
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(https://www.ebi.ac.uk/QuickGO/term/GO:0043410)
GO:0007283 : spermatogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0007283)
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GO:0048066 : developmental pigmentation
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GO:0009968 : negative regulation of signal transduction
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GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein
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(https://www.ebi.ac.uk/QuickGO/term/GO:0051091)
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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
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(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)
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(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
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(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)
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Molecular Type
Gene Amplification (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Gene Amplification^\#gephebase-summary-title)

Complex Change (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Complex Change^\#gephebase-summary-title)
Aberration Type
Molecular Details of the Mutation
Copy Number Variation of <600kb segments encompassing KIT
Experimental Evidence
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^\#gephebase-summary-title)
Serial translocation by means of circular intermediates underlies colour sidedness in cattle. (2012) (https://pubmed.ncbi.nlm.nih.gov/22297974)
Durkin K; Coppieters W; DrÃ $q$ gemẽ¹⁄4ller C; Ahariz N; Cambisano N; Druet T; Fasquelle C; Haile A; Horin P; Huang L; Kamatani Y; Karim L; Lathrop M; Moser S; Oldenbroek K; Rieder S; Sartelet A; SÃqlkner J; StÃ Allhammar H; Zelenika D; Zhang Z; Leeb T; Georges M; Charlier C

Colour sidedness is a dominantly inherited phenotype of cattle characterized by the polarization of pigmented sectors on the flanks, snout and ear tips. It is also referred to as 'lineback' or 'witrik' (which means white back), as colour-sided animals typically display a white band along their spine. Colour sidedness is documented at least since the Middle Ages and is presently segregating in several cattle breeds around the globe, including in Belgian blue and brown Swiss. Here we report that colour sidedness is determined by a first allele on chromosome 29 ( $\mathrm{Cs}(29)$ ), which results from the translocation of a 492-kilobase chromosome 6 segment encompassing KIT to chromosome 29, and a second allele on chromosome 6 ( $\mathrm{Cs}(6)$ ), derived from the first by repatriation of fused 575 -kilobase chromosome 6 and 29 sequences to the KIT locus. We provide evidence that both translocation events involved circular intermediates. This is the first example, to our knowledge, of a phenotype determined by homologous yet non-syntenic alleles that result from a novel copy-number-variant-generating mechanism.

Additional References

## RELATED GEPHE

## Related Genes

11 (Agouti, coatomer protein complex subunit alpha (COPA), Kit ligand, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PMEL17, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Coloration/and+groupHaplotypes=true\#gephebase-summary-title)

Related Haplotypes
2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9913^/or+Gene Gephebase=^Kit (type III receptor protein-tyrosine kinase)^/and+Taxon ID=^9913^\#gephebase-summary-title)

## EXTERNAL LINKS

## COMMENTS

@CNV @AllelicSeries ; the colour-sided variant in yaks is due to introgression into yaks from Mongolian Turano cattle that have been herded with Mongolian yaks for more than 1500 years enabling the backcrossing of female yak-cattle hybrids to male yaks. https://omia.org/OMIA001576/9913/

