GEPHE SUMMARY

Gephebase Gene

Microphtalmia-associated transcription factor (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Microphtalmia-associated transcription factor^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002326

Main curator

Martin

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 - spotted and white (Brown Swiss bred)

Ancestral State

Taxon A

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^#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

Rank

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 A an Infraspecies?

No

Taxon B

Latin Name

Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#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

Rank

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?

No

GENOTYPIC CHANGE

Generic Gene Name

Mitf

Synonyms

Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit

String

10090.ENSMUSP00000044938 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000044938)

Sequence Similarities

Belongs to the MiT/TFE family.

GO - Molecular Function

GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0043565 : sequence-specific DNA binding

UniProtKB Mus musculus

Q08874 (http://www.uniprot.org/uniprot/Q08874)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0003682 : chromatin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003682)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000978)
GO:0070888 : E-box binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070888)
GO:0003705 : transcription factor activity, RNA polymerase II distal enhancer sequence-specific binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003705)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006357 : regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0006355 : regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006355)
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:0000122 : negative regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0010628 : positive regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0006351 : transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0006351)
GO:0045165 : cell fate commitment (https://www.ebi.ac.uk/QuickGO/term/GO:0045165)
GO:0010468 : regulation of gene expression (https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0030318 : melanocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0043010 : camera-type eye development (https://www.ebi.ac.uk/QuickGO/term/GO:0043010)
GO:0030316 : osteoclast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030316)
GO:0042127 : regulation of cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0042127)
GO:0046849 : bone remodeling (https://www.ebi.ac.uk/QuickGO/term/GO:0046849)
GO:0044336 : canonical Wnt signaling pathway involved in negative regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0044336)
GO:0030336 : negative regulation of cell migration (https://www.ebi.ac.uk/QuickGO/term/GO:0030336)
GO:2000144 : positive regulation of DNA-templated transcription, initiation (https://www.ebi.ac.uk/QuickGO/term/GO:2000144)
GO:0065003 : protein-containing complex assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0065003)
GO:0045670 : regulation of osteoclast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045670)
GO:2001141 : regulation of RNA biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:2001141)
GO:0016055 : Wnt signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0016055)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0032991 : protein-containing complex (https://www.ebi.ac.uk/QuickGO/term/GO:0032991)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

unknown based SNP association

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

A non-coding regulatory variant in the 5′-region of the MITF gene is associated with white-spotted coat in Brown Swiss cattle. (2019) (https://pubmed.ncbi.nlm.nih.gov/30506810)

Authors

Hofstetter S; Seefried F; HÃ¤fliger IM; Jagannathan V; Leeb T; DrÃ¶gemÃ¼ller C

Abstract

Recently, the Swiss breeding association reported an increasing number of white-spotted cattle in the Brown Swiss breed, which is normally solid brown coloured. A total of 60 Brown Swiss cattle with variably sized white abdominal spots, facial markings and depigmented claws were collected for this study. A genome-wide association study using 40k SNP genotypes of 20 cases

and 1619 controls enabled us to identify an associated genome region on chromosome 22 containing the MITF gene, encoding the melanogenesis associated transcription factor. Variants at the MITF locus have been reported before to be associated with white or white-spotted phenotypes in other species such as horses, dogs and mice. Whole-genome sequencing of a single white-spotted cow and subsequent genotyping of 172 Brown Swiss cattle revealed two significantly associated completely linked single nucleotide variants (rs722765315 and rs719139527). Both variants are located in the 5′-regulatory region of the bovine MITF gene, and comparative sequence analysis showed that the variant rs722765315, located 139Â kb upstream of the transcription start site of the bovine melanocyte-specific MITF transcript, is situated in a multi-species conserved sequence element which is supposed to be regulatory important. Therefore, we hypothesize that rs722765315 represents the most likely causative variant for the white-spotting phenotype observed in Brown Swiss cattle. Presence of the mutant allele in a heterozygous or homozygous state supports a dominant mode of inheritance with incomplete penetrance and results in a variable extent of coat colour depigmentation.

Additional References

RELATED GEPHE

Related Genes

11 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), 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

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Microphtalmia-associated transcription factor^/and+Taxon ID=^9913^/or+Gene Gephebase=^Microphtalmia-associated transcription factor^/and+Taxon ID=^9913^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism @AllelicSeries https://omia.org/OMIA000214/9913/