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

GP00000656

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 (Holstein and Simmental breeds)

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 but association with intronic variant

Experimental Evidence

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

Main Reference

Haplotype variability in the bovine MITF gene and association with piebaldism in Holstein and Simmental cattle breeds. (2012) (https://pubmed.ncbi.nlm.nih.gov/22486495)

Authors

Fontanesi L; Scotti E; Russo V

Abstract

Candidate gene analysis, quantitative trait locus mapping in outbreed and experimental cross-populations and a genomewide association study in Holstein have reported that a few chromosome regions contribute to great variability in the degree of white/black spotting in cattle. In particular, an important region affecting this trait was localized on bovine chromosome 22

in the region containing the microphthalmia-associated transcription factor (MITF) gene. We sequenced a total of 7258 bp of the MITF gene in 40 cattle of different breeds, including 20 animals from spotted breeds (10 Italian Holstein and 10 Italian Simmental) and 20 animals from solid coloured breeds (10 Italian Brown and 10 Reggiana), and identified 17 single nucleotide polymorphisms (SNPs). The allele frequencies of one polymorphism (g.32386957A>T) were clearly different between spotted (A = 0.875; T = 0.125) and non-spotted breeds (A = 0.125; T = 0.875) (P = 8.2E-12). This result was confirmed by genotyping additional animals of these four breeds (P < 1.0E-20). A total of 21 different haplotypes were inferred from the sequenced animals. Considering similarities among haplotypes, spotted and non-spotted groups of cattle showed significant differences in their haplotype distribution (P = 0.001), which was further supported by the analysis of molecular variance (amova) of two genotyped SNPs in an enlarged sample of cattle. Variability in the MITF gene clearly explained the differences between spotted and non-spotted phenotypes but, at the same time, it is evident that this gene is not the only genetic factor determining piebaldism in Italian Holstein and Italian Simmental cattle breeds.

Additional References

Genetic architecture of complex traits and accuracy of genomic prediction: coat colour, milk-fat percentage, and type in Holstein cattle as contrasting model traits. (2010) (https://pubmed.ncbi.nlm.nih.gov/20927186)
Genome-wide association analysis reveals QTL and candidate mutations involved in white spotting in cattle. (2019) (https://pubmed.ncbi.nlm.nih.gov/31703548)

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/