

## GEPHE SUMMARY

<p>Microphthalmia-associated transcription factor (<a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^Microphthalmia-associated+transcription+factor+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+^Microphthalmia-associated+transcription+factor+^#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00002326</p> <p>Martin</p> <p>Entry Status</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>Bos bovis</p> <p>Bos bovis - spotted and white (Brown Swiss bred)</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>Taxon B</p>	
<p><b>Bos taurus</b> (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Bos+taurus+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Bos+taurus+^#gephebase-summary-title</a>)</p> <p>cattle</p> <p>Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus</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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos</p> <p>Bos (oxen, cattle) - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a>)</p> <p>9913 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913</a>)</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>is Taxon A an Intraspecies?</p>	<p><b>Bos taurus</b> (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Bos+taurus+^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+^Bos+taurus+^#gephebase-summary-title</a>)</p> <p>cattle</p> <p>Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus</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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos</p> <p>Bos (oxen, cattle) - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a>)</p> <p>9913 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913</a>)</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>is Taxon B an Intraspecies?</p>

## GENOTYPIC CHANGE

<p>Mitf</p> <p>Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit</p> <p>10090.ENSMUSP00000044938 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938</a>)</p> <p>Belongs to the MiT/TFE family.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>GO:0046983 : protein dimerization activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046983">https://www.ebi.ac.uk/QuickGO/term/GO:0046983</a>)</p> <p>GO:0003700 : DNA-binding transcription factor activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003700">https://www.ebi.ac.uk/QuickGO/term/GO:0003700</a>)</p> <p>GO:0043565 : sequence-specific DNA binding</p>	<p>Q08874 (<a href="http://www.uniprot.org/uniprot/Q08874">http://www.uniprot.org/uniprot/Q08874</a>)</p> <p>0</p>	<p>0</p>

(<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>)

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

Presumptive Null

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

Molecular Type

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

Aberration Type

unknown based SNP association

Molecular Details of the Mutation

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

Experimental Evidence

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>)

Main Reference

Hofstetter S; Seefried F; HÄßfliger IM; Jagannathan V; Leeb T; DrÄgemÄller C

Authors

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

Abstract

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 139 Å 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.

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Additional References

## RELATED GEPHE

Related Genes

11 (Agouti, coatmer 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=^Microphthalmia-associated transcription factor^/and+Taxon ID=^9913^/or+Gene Gephebase=^Microphthalmia-associated transcription factor^/and+Taxon ID=^9913^#gephebase-summary-title>)

## EXTERNAL LINKS

## COMMENTS

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