

GEPHE SUMMARY

<p>Microphthalmia-associated transcription factor (#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00002327</p> <p>Martin</p> <p>Entry Status</p>	<p>GepheID</p> <p>Main curator</p>
--	---	------------------------------------

PHENOTYPIC CHANGE

<p>Morphology (#gephebase-summary-title)</p> <p>Coloration (coat) (#gephebase-summary-title)</p> <p>WT</p> <p>white-spotted</p> <p>Taxon A</p> <p>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>Latin Name</p> <p>Bubalus bubalis (#gephebase-summary-title)</p> <p>Common Name</p> <p>water buffalo</p> <p>Synonyms</p> <p>Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus</p> <p>Rank</p> <p>species</p> <p>Lineage</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; Bubalus</p> <p>Parent</p> <p>Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9918)</p> <p>NCBI Taxonomy ID</p> <p>89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=89462)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Bubalus bubalis (#gephebase-summary-title)</p> <p>Common Name</p> <p>water buffalo</p> <p>Synonyms</p> <p>Bubalus arnee; Bubalus arnee bubalis; water buffalo; domestic water buffalo; river buffalo; Bubalis arnee bubalis; Bubalis bubalis; Bubalus bubalus</p> <p>Rank</p> <p>species</p> <p>Lineage</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; Bubalus</p> <p>Parent</p> <p>Bubalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9918)</p> <p>NCBI Taxonomy ID</p> <p>89462 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=89462)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
--	---	--	--

GENOTYPIC CHANGE

<p>Mitf</p> <p>Wh; bw; mi; vit; BCC2; Bhlhe32; Gsfbcc2; Vitiligo; Bw; Mi; Vit</p> <p>10090.ENSMUSP00000044938 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000044938)</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>Q08874 (http://www.uniprot.org/uniprot/Q08874)</p> <p>0</p> <p>GenebankID or UniProtKB</p>
<p>GO:0046983 : protein dimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046983)</p> <p>GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)</p> <p>GO:0043565 : sequence-specific DNA binding</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>)

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

Presumptive Null

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

Molecular Type

Aberration Type

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

c.328C>T p.R110*

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=~Candidate+Gene^#gephebase-summary-title))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	STP	110

Main Reference

Microphthalmia-associated transcription factor mutations are associated with white-spotted coat color in swamp buffalo. (2015) (<https://pubmed.ncbi.nlm.nih.gov/26417640>)

Authors

Yusnizar Y; Wilbe M; Herlino AO; Sumantri C; Noor RR; Boediono A; Andersson L; Andersson G

Abstract

A candidate gene analysis of the microphthalmia-associated transcription factor (MITF) gene was used in an attempt to identify the genetic basis for a white-spotted coat color phenotype in the Asian swamp buffalo (*Bubalus bubalis carabanensis*). Ninety-three buffaloes-32 solid, 38 spotted and 23 white individuals-were Sanger-sequenced for all MITF exons as well as highly conserved intronic and flanking regions. MITF cDNA representing skin and iris tissue from six spotted, nine solid and one white buffaloes was also Sanger-sequenced to confirm detected mutations. Two independent loss-of-function mutations, a premature stop codon (c.328C>T, p.Arg110*) and a donor splice-site mutation (c.840+2T>A, p.Glu281_Leu282Ins8), both of which cause white-spotted coat color in swamp buffaloes, were identified. The nonsense mutation leads to a premature stop codon in exon 3, and likely removal of the resulting mRNA via nonsense-mediated decay pathway, whereas the donor splice-site mutation leads to aberrant splicing of exon 8 that encodes part of a highly conserved region of MITF. The resulting insertion of eight amino acid residues is expected to perturb the leucine zipper part in the basic helix-loop-helix leucine zipper (bHLH-Zip) domain and will most likely influence dimerization and DNA binding capacity. Electrophoretic mobility shift assay was performed using mutant and wild-type MITF proteins and showed that the mutant MITF protein resulting from the splice-site mutation decreased in vitro DNA binding capacity compared to wild-type MITF. White-spotted buffalo bulls are sacrificed in funeral ceremonies in Tana Toraja, Indonesia, because they are considered holy, and our results show that genetic variation causes a tie to the cultural use of these buffaloes.

© 2015 The Authors. Animal Genetics published by John Wiley & Sons Ltd on behalf of Stichting International Foundation for Animal Genetics.

Additional References

RELATED GEPHE

Related Genes

2 (Agouti (ASIP), tyrosinase (TYR)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^89462^/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=^89462^/or+Gene Gephebase=^Microphthalmia-associated transcription factor^/and+Taxon ID=^89462^#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

@Parallelism @AllelicSeries @HeterozygoteAdvantage <https://omia.org/OMIA000214/89462/>