

GEPHE SUMMARY

	Gephebase Gene		GepheID
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Myostatin (MSTN = GDF8)"#gephebase-summary-title)	GP00000685		
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category="Morphology"#gephebase-summary-title)			
	Trait		
Muscular mass (double muscling) (<a (double="" href="https://www.gephebase.org/search-criteria?/and+Trait=" mass="" muscling)"#gephebase-summary-title"="" muscular="">https://www.gephebase.org/search-criteria?/and+Trait="Muscular mass (double muscling)"#gephebase-summary-title)			
	Trait State in Taxon A		
Bos taurus			
	Trait State in Taxon B		
Bos taurus - Belgian Blue; Asturiana de los Valles; Rubia Gallega; Arthenaise			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Domesticated"#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Bos taurus"#gephebase-summary-title)		Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Bos taurus"#gephebase-summary-title)	
	Common Name		Common Name
cattle		cattle	
	Synonyms		Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus		Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	
	Rank		Rank
species		species	
	Lineage		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		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		Parent
Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)		Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)		9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
			Bos taurus - Belgian Blue; Asturiana de los Valles; Rubia Gallega; Arthenaise

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
MSTN		O14793 (http://www.uniprot.org/uniprot/O14793)	
	Synonyms		GenebankID or UniProtKB
GDF8; MSLHP		AAB81508 (https://www.ncbi.nlm.nih.gov/nuccore/AAB81508)	
	String		
9606.ENSPO0000260950 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000260950)			
	Sequence Similarities		
Belongs to the TGF-beta family.			
	GO - Molecular Function		
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)			
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			

GO:0008201 : heparin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0008201>)
GO:0005125 : cytokine activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0005125>)
GO:0008083 : growth factor activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0008083>)
GO:0005102 : signaling receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005102>)
GO:0005160 : transforming growth factor beta receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005160>)

GO - Biological Process

GO:0045893 : positive regulation of transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0048468 : cell development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048468>)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010862>)
GO:0042981 : regulation of apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042981>)
GO:0043408 : regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043408>)
GO:0060395 : SMAD protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060395>)
GO:0043627 : response to estrogen (<https://www.ebi.ac.uk/QuickGO/term/GO:0043627>)
GO:0046716 : muscle cell cellular homeostasis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046716>)
GO:0045471 : response to ethanol (<https://www.ebi.ac.uk/QuickGO/term/GO:0045471>)
GO:0033574 : response to testosterone
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033574>)
GO:0007179 : transforming growth factor beta receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007179>)
GO:0009408 : response to heat (<https://www.ebi.ac.uk/QuickGO/term/GO:0009408>)
GO:0071549 : cellular response to dexamethasone stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071549>)
GO:0007517 : muscle organ development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007517>)
GO:0014839 : myoblast migration involved in skeletal muscle regeneration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014839>)
GO:0046627 : negative regulation of insulin receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046627>)
GO:0033673 : negative regulation of kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033673>)
GO:0014741 : negative regulation of muscle hypertrophy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014741>)
GO:0045662 : negative regulation of myoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045662>)
GO:2000818 : negative regulation of myoblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000818>)
GO:0051898 : negative regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051898>)
GO:1902725 : negative regulation of satellite cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902725>)
GO:1902723 : negative regulation of skeletal muscle satellite cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902723>)
GO:0048632 : negative regulation of skeletal muscle tissue growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048632>)
GO:0022602 : ovulation cycle process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0022602>)
GO:0010592 : positive regulation of lamellipodium assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010592>)
GO:0010759 : positive regulation of macrophage chemotaxis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010759>)
GO:0051602 : response to electrical stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051602>)
GO:0009629 : response to gravity (<https://www.ebi.ac.uk/QuickGO/term/GO:0009629>)
GO:0014850 : response to muscle activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014850>)
GO:0014732 : skeletal muscle atrophy (<https://www.ebi.ac.uk/QuickGO/term/GO:0014732>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Deletion Size

10-99 bp

Molecular Details of the Mutation

nt821 11bp deletion c.821-831delTGAACTCCA p.Glu275ArgfsX14

Experimental Evidence

A deletion in the bovine myostatin gene causes the double-muscling phenotype in cattle. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9288100>)

Grobet L; Martin LJ; Poncelet D; Pirottin D; Brouwers B; Riquet J; Schoeberlein A; Dunner S; MÃ©nissier F; Massabanda J; Fries R; Hanset R; Georges M

An exceptional muscle development commonly referred to as ‘double-muscling’ (Fig. 1) has been seen in several cattle breeds and has attracted considerable attention from beef producers. Double-muscling animals are characterized by an increase in muscle mass of about 20%, due to general skeletal-muscle hyperplasia-that is, an increase in the number of muscle fibers rather than in their individual diameter. Although the hereditary nature of the double-muscling condition was recognized early on, the precise mode of inheritance has remained controversial; monogenic (dominant and recessive), oligogenic and polygenic models have been proposed. In the Belgian Blue cattle breed (BBCB), segregation analysis performed both in experimental crosses and in the outbred population suggested an autosomal recessive inheritance. This was confirmed when the muscular hypertrophy (mh) locus was mapped 3.1 cM from microsatellite TGLA44 on the centromeric end of bovine chromosome 2 (ref. 5). We used a positional candidate approach to demonstrate that a mutation in bovine MSTN, which encodes myostatin, a member of the TGF beta superfamily, is responsible for the double-muscling phenotype. We report an 11-bp deletion in the coding sequence for the bioactive carboxy-terminal domain of the protein causing the muscular hypertrophy observed in Belgian Blue cattle.

Double muscling in cattle due to mutations in the myostatin gene. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9356471>)
Mutations in myostatin (GDF8) in double-muscling Belgian Blue and Piedmontese cattle. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9314496>)

RELATED GEPHE

1 (MRC2) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9913~/and+Trait=Muscular+mass/and+groupHaplotypes=true#gephebase-summary-title>)

7 ([https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~Myostatin+\(MSTN+=GDF8\)~/and+Taxon+ID=~9913~/or+Gene+Gephebase=~Myostatin+\(MSTN+=GDF8\)~/and+Taxon+ID=~9913~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~Myostatin+(MSTN+=GDF8)~/and+Taxon+ID=~9913~/or+Gene+Gephebase=~Myostatin+(MSTN+=GDF8)~/and+Taxon+ID=~9913~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@Parallelism @AllelicSeries <https://omia.org/OMIA000683/9913/>