

	Gephebase Gene	GepheID
Myostatin (MSTN = GDF8) (<a +myostatin+(mstn='GDF8)^#gephebase-summary-title"' href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Myostatin+(MSTN=GDF8)^#gephebase-summary-title)	GP00000689	
	Entry Status	Main curator
Published	Martin	

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)	Trait Category
Muscular mass (double muscling) (https://www.gephebase.org/search-criteria?/and+Trait=~Muscular mass (double muscling)~#gephebase-summary-title)	Trait
Bos taurus	Trait State in Taxon A
Bos taurus - Piedmontese - Gasconne	Trait State in Taxon B
Taxon A	Ancestral State
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)	Taxonomic Status
Taxon A	Taxon B
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)
cattle	cattle
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus
species	species
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
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)
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)
No	Yes
	Taxon B Description
	Bos taurus - Piedmontese - Gasconne

MSTN	Generic Gene Name	O14793 (http://www.uniprot.org/uniprot/O14793)	UniProtKB Homo sapiens
GDF8; MSLHP	Synonyms	AFN44374 (https://www.ncbi.nlm.nih.gov/nucore/AFN44374)	GenebankID or UniProtKB
9606.ENSPP00000260950 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000260950)	String		
Belongs to the TGF-beta family.	Sequence Similarities		
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)	GO - Molecular Function		
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
No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No^#gephebase-summary-title)	
	Molecular Type
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title)	
	Aberration Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP^#gephebase-summary-title)	
	SNP Coding Change
Nonsynonymous	
	Molecular Details of the Mutation
C313Y caused by G938A; disrupts disulphide bond	
	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Cys	Tyr	313

Main Reference

Double muscling in cattle due to mutations in the myostatin gene. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9356471>)

Authors

McPherron AC; Lee SJ

Abstract

Myostatin (GDF-8) is a member of the transforming growth factor beta superfamily of secreted growth and differentiation factors that is essential for proper regulation of skeletal muscle mass in mice. Here we report the myostatin sequences of nine other vertebrate species and the identification of mutations in the coding sequence of bovine myostatin in two breeds of double-muscled cattle, Belgian Blue and Piedmontese, which are known to have an increase in muscle mass relative to conventional cattle. The Belgian Blue myostatin sequence contains an 11-nucleotide deletion in the third exon which causes a frameshift that eliminates virtually all of the mature, active region of the molecule. The Piedmontese myostatin sequence contains a missense mutation in exon 3, resulting in a substitution of tyrosine for an invariant cysteine in the mature region of the protein. The similarity in phenotypes of double-muscled cattle and myostatin null mice suggests that myostatin performs the same biological function in these two species and is a potentially useful target for genetic manipulation in other farm animals.

Additional References

Molecular definition of an allelic series of mutations disrupting the myostatin function and causing double-muscling in cattle. (1998) (<https://pubmed.ncbi.nlm.nih.gov/9501304>)

Mutations in myostatin (GDF8) in double-muscled Belgian Blue and Piedmontese cattle. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9314496>)

RELATED GEPHE

Related Genes

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

Related Haplotypes

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/>