

Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?and+Gene=Gephebase=^Myostatin (MSTN = GDF8)^#gephebase-summary-title)	Gephebase Gene	GP00000690	GepheID
Published	Entry Status	Martin	Main curator

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 - Marchigiana		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Bos taurus (#gephebase-summary-title)		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 Taurus		Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	
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	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
Bos taurus - Marchigiana		Bos taurus - Marchigiana	

MSLN GDF8; MSLHP 9606.ENSP00000260950 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000260950) Belongs to the TGF-beta family. 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)	Generic Gene Name Synonyms String Sequence Similarities GO - Molecular Function	UniProtKB Homo sapiens O14793 (http://www.uniprot.org/uniprot/O14793) GenebankID or UniProtKB ACS70961 (https://www.ncbi.nlm.nih.gov/nuccore/ACS70961)
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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 (<a +yes+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive+Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+Yes" #gephebase-summary-title)	
	Molecular Type
Coding (<a +coding+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Molecular+Type=">https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding" #gephebase-summary-title)	
	Aberration Type
SNP (<a +snp+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Aberration+Type=">https://www.gephebase.org/search-criteria?/and+Aberration Type="+SNP" #gephebase-summary-title)	
	SNP Coding Change
Nonsense	
	Molecular Details of the Mutation
E291* caused by G>T	
	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gln	STP	291

Main Reference

Double muscling in Marchigiana beef breed is caused by a stop codon in the third exon of myostatin gene. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12879361>)

Authors

Marchitelli C; Savarese MC; Cris   A; Nardone A; Marsan PA; Valentini A

Abstract

Double muscling is a partially recessive trait present in some beef breeds. It shows a high frequency in some breeds, while in others the frequency is low, and double-muscled individuals are rare. The double muscling is caused by an allelic series of mutations that cause a loss of function of the myostatin gene (GDF8). We describe here a new mutation in the myostatin gene in Marchigiana breed, a typical beef breed of Central Italy, in which rare double-muscling individuals have been described. A PCR product of the third exon was sequenced in subjects phenotypically showing double muscling, and a G > T transversion was discovered that introduces a premature stop codon. The variant found adds to the large series of mutations present in cattle, and particularly to the only two causative of double muscling in the third exon. A PCR-RFLP test is described for the rapid and effective identification of both heterozygous and homozygous subjects. It was applied to a larger survey carried on the same and also in two other beef breeds, Chianina and Romagnola. Further individuals carrying the new variant were found in Marchigiana, but none in the other breeds. The results may be important for a better comprehension of the role of myostatin in muscular development, for commercial use and for the inference of phylogeny of this gene.

Additional References

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/>