

Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene)	Gephebase Gene	GP00000688	GepheID
Gephebase="Myostatin (MSTN = GDF8)"#gephebase-summary-title)	Entry Status	Martin	Main curator
Published			

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^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 - Maine-Anjou	Trait State in Taxon B
Taxon A	Ancestral State
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)	Taxonomic Status

GENOTYPIC CHANGE			
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:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		

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

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

E226* caused by G676T

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gln	STP	226

Main Reference

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>)

Authors

Grobet L; Poncelet D; Royo LJ; Brouwers B; Pirottin D; Michaux C; MÃ©nissier F; Zanotti M; Dunner S; Georges M

Abstract

We have determined the entire myostatin coding sequence for 32 double-muscled cattle sampled from ten European cattle breeds. Seven DNA sequence polymorphisms were identified, of which five would be predicted to disrupt the function of the protein, one is a conservative amino acid substitution, and one a silent DNA sequence variant. Four additional DNA sequence polymorphisms were identified in myostatin intronic sequences. In all but two breeds, all double-muscled animals were either homozygous or compound heterozygotes for one of the five loss-of-function mutations. The absence of obvious loss-of-function mutations in the coding sequence of the two remaining breeds points either towards additional mutations in unexplored segments of the gene, or towards locus heterogeneity of double-muscling.

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/>