

GEPHE SUMMARY

	Gephebase Gene		GepheID
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Myostatin (MSTN = GDF8)"#gephebase-summary-title)		GP00000692	
	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 - Wagyu			
	Trait State in Taxon B		
Bos taurus - Limousin			
	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?
Yes		Yes	
	Taxon A Description		Taxon B Description
Bos taurus - Wagyu		Bos taurus - Limousin	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
MSTN		O14793 (http://www.uniprot.org/uniprot/O14793)	
	Synonyms		GenebankID or UniProtKB
GDF8; MSLHP		ACS70961 (https://www.ncbi.nlm.nih.gov/nuccore/ACS70961)	
	String		
9606.ENSPP00000260950 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000260950)			
	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
No (<a +no`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Presumptive Null=">https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No`#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
Nonsynonymous	
	Molecular Details of the Mutation
c.282C>A p.Phe94Leu	
	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Phe	Leu	94

A Limousin specific myostatin allele affects longissimus muscle area and fatty acid profiles in a Wagyu-Limousin F2 population. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19213716>)

Alexander LJ; Kuehn LA; Smith TP; Matukumalli LK; Mote B; Koltes JE; Reecy J; Geary TW; Rule DC; Macneil MD

A microsatellite-based genome scan of a Wagyu x Limousin F(2) cross population previously demonstrated QTL affecting LM area and fatty acid composition were present in regions near the centromere of BTA2. In this study, we used 70 SNP markers to examine the centromeric 24 megabases (Mb) of BTA2, including the Limousin-specific F94L myostatin allele (AB076403.1; 415C > A) located at approximately 6 Mb on the draft genome sequence of BTA2. A significant effect of the F94L marker was observed (F = 60.17) for LM area, which indicated that myostatin is most likely responsible for the effect. This is consistent with previous reports that the substitution of Leu for Phe at AA 94 of myostatin (caused by the 415C > A transversion) is associated with increased muscle growth. Surprisingly, several fatty acid trait QTL, which affected the amount of unsaturated fats, also mapped to or very near the myostatin marker, including the ratio of C16:1 MUFA to C16:0 saturated fat (F = 16.72), C18:1 to C18:0 (F = 18.88), and total content of MUFA (F = 17.12). In addition, QTL for extent of marbling (F = 14.73) approached significance (P = 0.05), and CLA concentration (F = 9.22) was marginally significant (P = 0.18). We also observed associations of SNP located at 16.3 Mb with KPH (F = 15.00) and for the amount of SFA (F = 12.01). These results provide insight into genetic differences between the Wagyu and Limousin breeds and may lead to a better tasting and healthier product for consumers through improved selection for lipid content of beef.

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/>