

GEPHE SUMMARY

	Gephebase Gene		GepheID
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="Myostatin (MSTN = GDF8)"#gephebase-summary-title)		GP00002248	
	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 - Gelbvieh			
	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 - Gelbvieh	

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.191T>C p.L64P	
	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Leu	Pro	64

	Main Reference
A novel myostatin mutation in double-muscled German Gelbvieh. (2015) (https://pubmed.ncbi.nlm.nih.gov/25515003)	
	Authors
Dierks C; Eder J; Glatzer S; Lehner S; Distl O	
	Abstract
-	
	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 Gephabase=~Myostatin (MSTN = GDF8)~/and+Taxon ID=~9913~/or+Gene Gephabase=~Myostatin (MSTN = GDF8)~/and+Taxon ID=~9913~#gephebase-summary-title)	

EXTERNAL LINKS

COMMENTS

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