

GEPHE SUMMARY

	Gephebase Gene		GepheID
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene)		GP00000693	
Gephebase="Myostatin (MSTN = GDF8)"#gephebase-summary-title)			Main curator
Published	Entry Status	Martin	

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)			
Ovis aries	Trait State in Taxon A		
Ovis aries; Norwegian White	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic)	Taxonomic Status		
Status="Domesticated"#gephebase-summary-title)			
	Taxon A	Taxon B	
	Latin Name		Latin Name
Ovis aries		Ovis aries	
(<a aries"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=" ovis="">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Ovis aries"#gephebase-summary-title)		(<a aries"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=" ovis="">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Ovis aries"#gephebase-summary-title)	
	Common Name		Common Name
sheep		sheep	
	Synonyms		Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758		Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	
	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; Caprinae; Ovis		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; Caprinae; Ovis	
	Parent		Parent
Ovis () - (Rank: genus)		Ovis () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9940		9940	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Ovis aries; Norwegian White	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
MSTN		O14793 (http://www.uniprot.org/uniprot/O14793)	
	Synonyms		GenebankID or UniProtKB
GDF8; MSLHP		CCD67081 (https://www.ncbi.nlm.nih.gov/nuccore/CCD67081)	
	String		
9606.ENSP00000260950			
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000260950)			
	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

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion resulting in a premature stop codon at position 320

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title>)

Main Reference

A frameshift mutation in the coding region of the myostatin gene (MSTN) affects carcass conformation and fatness in Norwegian White Sheep (*Ovis aries*). (2009) (<https://pubmed.ncbi.nlm.nih.gov/19392824>)

Authors

Boman IA; Klemetsdal G; Blichfeldt T; Nafstad O; VÅge DI

Abstract

Mutations in the coding region of the myostatin gene (MSTN) are known to cause an increased muscle mass (IMM) phenotype in several mammals, including mice, dogs, cattle and humans. In sheep, a mutation in the 3'-UTR region introducing a microRNA target site has been reported to cause an IMM-like phenotype because of downregulation of translation. Here we report a novel single base deletion in the coding region of the myostatin gene causing an IMM phenotype in Norwegian White Sheep, characterized by a high carcass conformation class and low fat class (EUROP classification system). The deletion disrupts the reading frame from amino acid (aa) position 320, ending in a premature stop codon in aa position 359. In our material, these MSTN mutations segregated in a pattern showing that they reside in two different haplotypes. The phenotypic effect of the single base deletion is more profound than that of the 3'-UTR mutation.

Additional References

RELATED GEPHE

Related Genes

1 (Callipyge (CLPG1)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9940~/and+Trait=Muscular mass/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

2 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Myostatin \(MSTN = GDF8\)~/and+Taxon ID=~9940~/or+Gene Gephebase=~Myostatin \(MSTN = GDF8\)~/and+Taxon ID=~9940~#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Myostatin (MSTN = GDF8)~/and+Taxon ID=~9940~/or+Gene Gephebase=~Myostatin (MSTN = GDF8)~/and+Taxon ID=~9940~#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS