GEPHE SUMMARY

Gephebase Gene
Myostatin (MSTN = GDF8)

GephelD
GP00000693

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Muscular mass (double muscling)

Trait State in Taxon A
Ovis aries

Trait State in Taxon B
Ovis aries; Norwegian White

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Ovis aries

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus)

NCBI Taxonomy ID
9940

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Ovis aries

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

Rank
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Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus)

NCBI Taxonomy ID
9940

is Taxon B an Infraspecies?
Yes

Taxon B Description
Ovis aries; Norwegian White

GENOTYPIC CHANGE

Generic Gene Name
MSTN

Synonyms
GDF8; MSLHP

String
9606.ENSP00000260950

Sequence Similarities
Belongs to the TGF-beta family.

GO - Molecular Function
GO:0042802 : identical protein binding
GO:0042803 : protein homodimerization activity
GO:0008201 : heparin binding
GO:0005125 : cytokine activity
GO:0008083 : growth factor activity
GO:0005102 : signaling receptor binding
GO:0005160 : transforming growth factor beta receptor binding

UniProtKB Homo sapiens
O14793

GenebankID or UniProtKB
CCD67081

GO - Biological Process
GO:0045893 : positive regulation of transcription, DNA-templated
GO:0048468 : cell development
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
GO:0042981 : regulation of apoptotic process
GO:0043408 : regulation of MAPK cascade
GO:0060395 : SMAD protein signal transduction
GO:0043627 : response to estrogen
GO:0046716 : muscle cell cellular homeostasis
GO:0045471 : response to ethanol
GO:0033574 : response to testosterone
GO:0007179 : transforming growth factor beta receptor signaling pathway
GO:0009408 : response to heat
GO:0071549 : cellular response to dexamethasone stimulus
GO:0007517 : muscle organ development
GO:0014839 : myoblast migration involved in skeletal muscle regeneration
GO:0046627 : negative regulation of insulin receptor signaling pathway
GO:0033673 : negative regulation of kinase activity
GO:0014741 : negative regulation of muscle hypertrophy
GO:0045662 : negative regulation of myoblast differentiation
GO:2000818 : negative regulation of myoblast proliferation
GO:0051898 : negative regulation of protein kinase B signaling
GO:1902725 : negative regulation of satellite cell differentiation
GO:1902723 : negative regulation of skeletal muscle satellite cell proliferation
GO:0048632 : negative regulation of skeletal muscle tissue growth
GO:0022602 : ovulation cycle process
GO:0010592 : positive regulation of lamellipodium assembly
GO:0010759 : positive regulation of macrophage chemotaxis
GO:0051602 : response to electrical stimulus
GO:0009629 : response to gravity
GO:0014850 : response to muscle activity
GO:0014732 : skeletal muscle atrophy

GO - Cellular Component
GO:0005737 : cytoplasm
GO:0005615 : extracellular space

Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
Deletion

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

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)

Authors
Boman IA; Klemetsdal G; Blichfeldt T; Nafstad O; VÃ¥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))
Related Haplotypes
2

COMMENTS