

## GEPHE SUMMARY

**Gephebase Gene**  
Myostatin (MSTN = GDF8)

**Entry Status**  
Published

**GepheID**  
GP00000696

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Muscular mass (double muscling)

**Trait State in Taxon A**  
Sus scrofa

**Trait State in Taxon B**  
Sus scrofa - Pietrain

**Ancestral State**  
Data not curated

**Taxonomic Status**  
Domesticated

### Taxon A

**Latin Name**  
*Sus scrofa*

**Common Name**  
pig

**Synonyms**  
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofa

**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; Suina; Suidae; Sus

**Parent**  
Sus () - (Rank: genus)

**NCBI Taxonomy ID**  
9823

**is Taxon A an Intraspecies?**  
No

### Taxon B

**Latin Name**  
*Sus scrofa domesticus*

**Common Name**  
domestic pig

**Synonyms**  
Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig

**Rank**  
subspecies

**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; Suina; Suidae; Sus; Sus scrofa

**Parent**  
Sus scrofa (pig) - (Rank: species)

**NCBI Taxonomy ID**  
9825

**is Taxon B an Intraspecies?**  
Yes

**Taxon B Description**  
Sus scrofa - Pietrain

## 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

**GO - Biological Process**

**UniProtKB Homo sapiens**  
O14793

**GenebankID or UniProtKB**  
AAR18245

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

Unknown

#### Molecular Type

Cis-regulatory

#### Aberration Type

Unknown

#### Molecular Details of the Mutation

Possible promoter disruption

#### Experimental Evidence

Candidate Gene

#### Main Reference

Characterization of the complete porcine MSTN gene and expression levels in pig breeds differing in muscularity. (2008)

#### Authors

Stinckens A; Luyten T; Bijttebier J; Van den Maagdenberg K; Dieltiens D; Janssens S; De Smet S; Georges M; Buys N

#### Abstract

Myostatin (MSTN), a transforming growth factor beta superfamily member, is an essential factor for the growth and development of muscle mass. The protein functions as a negative regulator of muscle growth and is related to the so-called double-muscling phenotype in cattle, where a series of mutations renders the gene inactive. One particular breed of pigs, the Belgian Pi $\bar{A}$ @train, also shows a heavily muscled phenotype. The similarity of muscular phenotypes between the double-muscled cattle and Pi $\bar{A}$ @train pigs indicated that MSTN may be a candidate gene for muscular hypertrophy in pigs. In this study, we sequenced and analysed the complete MSTN gene from 45 pigs of five different breeds, including the heavily muscled Pi $\bar{A}$ @train breed at one extreme and the Meishan and Wild boar breeds at the other extreme. In total, 7626 bp of the porcine MSTN gene were sequenced, including the 5' and 3' UTR. Fifteen polymorphic loci were found, three of which were located in the promoter region, five in intron 1 and seven in intron 2. Most mutations were found when comparing the obtained MSTN sequence with porcine MSTN sequences already published. However, one polymorphism located at position 447 of the porcine MSTN promoter had a very high allele frequency in the Pi $\bar{A}$ @train pig breed and disrupted a putative myocyte enhancer factor 3 binding site. Real-time PCR using Sybr Green showed that this mutation was associated with expression levels of the MSTN gene in m. longissimus dorsi at an age of 4 weeks.

#### Additional References

## RELATED GEPHE

#### Related Genes

1 (Insulin-like growth factor 2 (IGF2))

#### Related Haplotypes

No matches found.

## COMMENTS

