GEPHE SUMMARY

Gephebase Gene
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Myostatin (MSTN = GDF8)^#gephebase-summary-title)
Entry Status
Published

GepheID
GP00000696
Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)
Trait
Muscular mass (double muscling) (https://www.gephebase.org/search-criteria?/and+Trait=^Muscular mass (double muscling)^#gephebase-summary-title)
Trait State in Taxon A
Sus scrofa
Trait State in Taxon B
Sus scrofa - Pietrain
Ancestral State
Data not curated
Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Sus scrofa
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa^#gephebase-summary-title)
Common Name
pig
Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus
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)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)
NCBI Taxonomy ID
9823
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)
is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Sus scrofa domesticus
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Sus scrofa domesticus^#gephebase-summary-title)
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)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)
NCBI Taxonomy ID
9825
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)
is Taxon B an Infraspecies?
Yes
Taxon B Description
Sus scrofa - Pietrain

GENOTYPIC CHANGE

Generic Gene Name
MSTN
Synonyms
GDF8; MSLHP
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)

UniProtKB Homo sapiens
O14793 (http://www.uniprot.org/uniprot/O14793)
GenebankID or UniProtKB
AAR18245 (https://www.ncbi.nlm.nih.gov/nuccore/AAR18245)

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

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

Molecular Details of the Mutation

Possible promoter disruption ; changes the G nucleotide at position 8 of the MEF3 binding site into an A nucleotide, thus disrupting the MEF3 binding site

Experimental Evidence

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

Main Reference

Characterization of the complete porcine MSTN gene and expression levels in pig breeds differing in muscularity. (2008) (https://pubmed.ncbi.nlm.nih.gov/18822098)

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 PiÃ©train, also shows a heavily muscled phenotype. The similarity of muscular phenotypes between the double-muscled cattle and PiÃ©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 PiÃ©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 PiÃ©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)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9823^/and+Trait=Muscular mass/or+Taxon ID=^9825^/and+Trait=Muscular mass/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism https://omia.org/OMIA000683/9823/