

	Gephebase Gene	GepheID
Myostatin (MSTN = GDF8) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Myostatin (MSTN = GDF8)^#gephebase-summary-title)	GP00000698	Main curator
Published	Entry Status: Martin	

Taxon A		Taxon B	
Latin Name	Common Name	Latin Name	Common Name
<i>Equus caballus</i> (<a +equus+caballus`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus`#gephebase-summary-title)	horse	<i>Equus caballus</i> (<a +equus+caballus`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Equus+caballus`#gephebase-summary-title)	horse
Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758		Equus przewalskii f. caballus; Equus przewalskii forma caballus; horse; domestic horse; equine; Equus caballus Linnaeus, 1758	
species		species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Perissodactyla; Equidae; Equus; Equus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Perissodactyla; Equidae; Equus; Equus	
Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)		Equus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=35510)	
9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)		9796 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9796)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

MSTN	Generic Gene Name	O14793 (http://www.uniprot.org/uniprot/O14793)	UniProtKB Homo sapiens
GDF8; MSLHP	Synonyms	BAB16046 (https://www.ncbi.nlm.nih.gov/nuccore/BAB16046)	GenebankID or UniProtKB
9606.ENSPP00000260950 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000260950)	String		
	Sequence Similarities		
Belongs to the TGF-beta family.			
GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)	GO - Molecular Function		
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
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
Insertion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Insertion^#gephebase-summary-title)	
	Insertion Size
100-999 bp	
	Molecular Details of the Mutation
227-bp SINE insertion in promoter	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)	
	Main Reference

Identification of the myostatin locus (MSTN) as having a major effect on optimum racing distance in the Thoroughbred horse in the USA. (2010) (<https://pubmed.ncbi.nlm.nih.gov/21070290>)

Authors

Binns MM; Boehler DA; Lambert DH

Abstract

One hundred and eighty-nine Thoroughbred horses that had won Graded Stakes races in North America were genotyped with the Illumina Equine SNP50 bead chip. Association tests using PLINK to determine whether any SNPs were associated with optimum racing distance (7 furlongs and under compared to 8-10 furlongs) identified a locus on ECA18 that was statistically significant ($-\log_{10} \text{EMP2}=1.63$) at the genome-wide level following permutation analysis (10,000 permutations). Bioinformatic analysis revealed that the two ECA18 SNPs with the highest statistical significance spanned the MSTN (myostatin) locus. Mutations in myostatin in several mammalian species have been associated with increased muscling, with a preferential increase in fast glycolytic type IIB fibres, which would increase power potential. Thoroughbred horses that race over sprint distances, which are 5-7 furlongs, are often characterized by impressive hind quarter musculature, strongly suggesting that the association observed between the ECA18 SNPs and optimum race distance is mediated through MSTN.

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Additional References

Genome-wide analysis reveals selection for important traits in domestic horse breeds. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23349635>)

A cohort study of racing performance in Japanese Thoroughbred racehorses using genome information on ECA18. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22221024>)

The “speed gene” effect of myostatin arises in Thoroughbred horses due to a promoter proximal SINE insertion. (2018) (<https://pubmed.ncbi.nlm.nih.gov/30379863>)

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