

GEPHE SUMMARY

	Gephebase Gene		GepheID
HMGA2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~HMGA2~#gephebase-summary-title)		GP00000485	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)			
	Trait		
Body size (https://www.gephebase.org/search-criteria?/and+Trait=~Body size~#gephebase-summary-title)			
	Trait State in Taxon A		
Equus caballus			
	Trait State in Taxon B		
Equus caballus (QTL study : used German Warmblood)			
	Ancestral State		
Data not curated			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus~#gephebase-summary-title)		Equus caballus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Equus caballus~#gephebase-summary-title)	
	Common Name		Common Name
horse		horse	
	Synonyms		Synonyms
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	
	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; 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	
	Parent		Parent
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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
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		Yes	
			Taxon B Description
		Equus caballus (QTL study : used German Warmblood)	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
HMGA2		P52926 (http://www.uniprot.org/uniprot/P52926)	
	Synonyms		GenebankID or UniProtKB
BABL; LIPO; HMGIC; HMGI-C; STQTL9		CRK77045 (https://www.ncbi.nlm.nih.gov/nuccore/CRK77045)	
	String		
9606.ENSPP00000384026 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000384026)			
	Sequence Similarities		
Belongs to the HMGA family.			
	GO - Molecular Function		
GO:0008134 : transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008134)			
GO:0044212 : transcription regulatory region DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0044212)			

GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001077>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0003680 : AT DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003680>)
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)
GO:0003712 : transcription coregulator activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0003712>)
GO:0051575 : 5'-deoxyribose-5-phosphate lyase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0051575>)
GO:0070742 : C2H2 zinc finger domain binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0070742>)
GO:0035497 : cAMP response element binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0035497>)
GO:0008301 : DNA binding, bending (<https://www.ebi.ac.uk/QuickGO/term/GO:0008301>)
GO:0003906 : DNA-(apurinic or apyrimidinic site) endonuclease activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0003906>)
GO:0004677 : DNA-dependent protein kinase activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0004677>)
GO:0035501 : MH1 domain binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0035501>)
GO:0035500 : MH2 domain binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0035500>)
GO:0031492 : nucleosomal DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0031492>)
GO:0046332 : SMAD binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046332>)

GO - Biological Process

GO:0007275 : multicellular organism development (<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
GO:0009615 : response to virus (<https://www.ebi.ac.uk/QuickGO/term/GO:0009615>)
GO:0043066 : negative regulation of apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)
GO:0045944 : positive regulation of transcription by RNA polymerase II (<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006355 : regulation of transcription, DNA-templated (<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)
GO:0000122 : negative regulation of transcription by RNA polymerase II (<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)
GO:0045892 : negative regulation of transcription, DNA-templated (<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0045893 : positive regulation of transcription, DNA-templated (<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
GO:0051301 : cell division (<https://www.ebi.ac.uk/QuickGO/term/GO:0051301>)
GO:0010628 : positive regulation of gene expression (<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:2000774 : positive regulation of cellular senescence (<https://www.ebi.ac.uk/QuickGO/term/GO:2000774>)
GO:0035986 : senescence-associated heterochromatin focus assembly (<https://www.ebi.ac.uk/QuickGO/term/GO:0035986>)
GO:0006325 : chromatin organization (<https://www.ebi.ac.uk/QuickGO/term/GO:0006325>)
GO:0048863 : stem cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048863>)
GO:0001837 : epithelial to mesenchymal transition (<https://www.ebi.ac.uk/QuickGO/term/GO:0001837>)
GO:0048762 : mesenchymal cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048762>)
GO:0048333 : mesodermal cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0048333>)
GO:2000648 : positive regulation of stem cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:2000648>)
GO:0043065 : positive regulation of apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0043065>)
GO:0002062 : chondrocyte differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0002062>)
GO:0045444 : fat cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0045444>)
GO:0040008 : regulation of growth (<https://www.ebi.ac.uk/QuickGO/term/GO:0040008>)
GO:0006284 : base-excision repair (<https://www.ebi.ac.uk/QuickGO/term/GO:0006284>)
GO:0035988 : chondrocyte proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0035988>)
GO:0031052 : chromosome breakage (<https://www.ebi.ac.uk/QuickGO/term/GO:0031052>)
GO:0030261 : chromosome condensation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030261>)
GO:0042769 : DNA damage response, detection of DNA damage

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042769>)
GO:0035987 : endodermal cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035987>)
GO:0031507 : heterochromatin assembly
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031507>)
GO:0035978 : histone H2A-S139 phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035978>)
GO:0003131 : mesodermal-endodermal cell signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003131>)
GO:0007095 : mitotic G2 DNA damage checkpoint
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007095>)
GO:0043922 : negative regulation by host of viral transcription
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043922>)
GO:2000773 : negative regulation of cellular senescence
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000773>)
GO:0043392 : negative regulation of DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043392>)
GO:2001033 : negative regulation of double-strand break repair via nonhomologous end joining (<https://www.ebi.ac.uk/QuickGO/term/GO:2001033>)
GO:0045869 : negative regulation of single stranded viral RNA replication via double stranded DNA intermediate (<https://www.ebi.ac.uk/QuickGO/term/GO:0045869>)
GO:0090402 : oncogene-induced cell senescence
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090402>)
GO:0045766 : positive regulation of angiogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045766>)
GO:0071158 : positive regulation of cell cycle arrest
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071158>)
GO:0071864 : positive regulation of cell proliferation in bone marrow
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071864>)
GO:2000685 : positive regulation of cellular response to X-ray
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000685>)
GO:2001022 : positive regulation of response to DNA damage stimulus
(<https://www.ebi.ac.uk/QuickGO/term/GO:2001022>)
GO:2000679 : positive regulation of transcription regulatory region DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000679>)
GO:0010564 : regulation of cell cycle process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010564>)
GO:2001038 : regulation of cellular response to drug
(<https://www.ebi.ac.uk/QuickGO/term/GO:2001038>)
GO:2000036 : regulation of stem cell population maintenance
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000036>)

GO - Cellular Component

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0035985 : senescence-associated heterochromatin focus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035985>)
GO:0000228 : nuclear chromosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0000228>)
GO:0032993 : protein-DNA complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032993>)
GO:0071141 : SMAD protein complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0071141>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

unknown

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and*Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

Four loci explain 83% of size variation in the horse. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22808074>)

Authors

Makvandi-Nejad S; Hoffman GE; Allen JJ; Chu E; Gu E; Chandler AM; Loredó AI; Bellone RR; Mezey JG; Brooks SA; Sutter NB

Abstract

Horse body size varies greatly due to intense selection within each breed. American Miniatures are less than one meter tall at the withers while Shires and Percherons can exceed two meters. The genetic basis for this variation is not known. We hypothesize that the breed population structure of the horse should simplify efforts to identify genes controlling size. In support of this, here we show with genome-wide association scans (GWAS) that genetic variation at just four loci can explain the great majority of horse size variation. Unlike humans, which are naturally reproducing and possess many genetic variants with weak effects on size, we show that horses, like other domestic mammals, carry just a small number of size loci with alleles of large effect. Furthermore, three of our horse size loci contain the LCORL, HMGA2 and ZFAT genes that have previously been found to control human height. The LCORL/NCAPG locus is also implicated in cattle growth and HMGA2 is associated with dog size. Extreme size diversification is a hallmark of domestication. Our results in the horse, complemented by the prior work in cattle and dog, serve to pinpoint those very few genes that have played major roles in the rapid evolution of size during domestication.

Additional References

No matches found.

EXTERNAL LINKS

COMMENTS