

GEPHE SUMMARY

	Gephebase Gene		GepheID
HMGA2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~HMGA2~#gephebase-summary-title)		GP00000487	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology~#gephebase-summary-title)		Trait	
Ear size (https://www.gephebase.org/search-criteria?/and+Trait=~Ear size~#gephebase-summary-title)			
	Trait State in Taxon A		
Sus scrofa - White Duroc		Trait State in Taxon B	
Sus scrofa- Erhualian (large ears; wrinkly skin)			
	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
Sus scrofa (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Sus scrofa~#gephebase-summary-title)		Sus scrofa domesticus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Sus scrofa domesticus~#gephebase-summary-title)	
	Common Name		Common Name
pig		domestic pig	
	Synonyms		Synonyms
pig; pigs; swine; wild boar; Sus scrofa Linnaeus, 1758; Sus scrofus		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
	Rank		Rank
species		subspecies	
	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; Cetartiodactyla; Suina; Suidae; Sus		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		Parent
Sus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9822)		Sus scrofa (pig) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9823 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9823)		9825 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9825)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
Yes		Yes	
	Taxon A Description		Taxon B Description
Sus scrofa - White Duroc		Sus scrofa- Erhualian (large ears; wrinkly skin)	

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		AEO79122 (https://www.ncbi.nlm.nih.gov/nuccore/AEO79122)	
	String		
9606.ENSP00000384026 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier= 9606.ENSP00000384026)			
	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

Linkage Mapping (<https://www.gephebase.org/search-criteria/?and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title>)

Main Reference

Fine mapping of a QTL for ear size on porcine chromosome 5 and identification of high mobility group AT-hook 2 (HMGA2) as a positional candidate gene. (2012)
(<https://pubmed.ncbi.nlm.nih.gov/22420340>)

Authors

Li P; Xiao S; Wei N; Zhang Z; Huang R; Gu Y; Guo Y; Ren J; Huang L; Chen C

Abstract

Ear size and shape are distinct conformation characteristics of pig breeds. Previously, we identified a significant quantitative trait locus (QTL) influencing ear surface on pig chromosome 5 in a White Duroc—Erhualian F2 resource population. This QTL explained more than 17% of the phenotypic variance.

Four new markers on pig chromosome 5 were genotyped across this F2 population. RT-PCR was performed to obtain expression profiles of different candidate genes in ear tissue. Standard association test, marker-assisted association test and F-drop test were applied to determine the effects of single nucleotide polymorphisms (SNP) on ear size. Three synthetic commercial lines were also used for the association test.

We refined the QTL to an 8.7-cM interval and identified three positional candidate genes i.e. HMGA2, SOX5 and PTHLH that are expressed in ear tissue. Seven SNP within these three candidate genes were selected and genotyped in the F2 population. Of the seven SNP, HMGA2 SNP (JF748727: g.2836 A>G) showed the strongest association with ear size in the standard association test and marker-assisted association test. With the F-drop test, F value decreased by more than 97% only when the genotypes of HMGA2 g.2836 A>G were included as a fixed effect. Furthermore, the significant association between g.2836 A>G and ear size was also demonstrated in the synthetic commercial Suta pig line. The haplotype-based association test showed that the phenotypic variance explained by HMGA2 was similar to that explained by the QTL and at a much higher level than by SOX5. More interestingly, HMGA2 is also located within the dog orthologous chromosome region, which has been shown to be associated with ear type and size.

HMGA2 was the closest gene with a potential functional effect to the QTL or marker for ear size on chromosome 5. This study will contribute to identify the causative gene and mutation underlying this QTL.

Additional References

RELATED GEPHE

2 (WIF1, PPAR-delta) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9823~/and+Trait=Ear size/or+Taxon ID=~9825~/and+Trait=Ear size/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9823~/and+Trait=Ear+size/or+Taxon+ID=~9825~/and+Trait=Ear+size/and+groupHaplotypes=true#gephebase-summary-title))

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS