

GEPHE SUMMARY

Gephebase Gene		GepheID
PPAR-delta (<a +ppar-delta^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+PPAR-delta^#gephebase-summary-title)		GP00000919
Entry Status		Main curator
Published		Martin

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology (<a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title)	
Trait	
Ear size (<a +ear+size^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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)	

Trait #2	Trait Category
Morphology (<a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title)	
Trait	
Skin aspect (<a +skin+aspect^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Skin+aspect^#gephebase-summary-title)	
Trait State in Taxon A	
-	
Trait State in Taxon B	
-	

Ancestral State	
Data not curated	
Taxonomic Status	
Domesticated (<a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated^#gephebase-summary-title)	
Taxon A	Taxon B
Latin Name	
Sus scrofa (<a +sus+scrofa^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">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 scrofulus	
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 Intraspecies?	
Yes	
Taxon A Description	
Sus scrofa - White Duroc	
Latin Name	
Sus scrofa domesticus (<a +sus+scrofa+domesticus^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">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 Intraspecies?	
Yes	
Taxon B Description	
Sus scrofa- Erhualian (large ears; wrinkly skin)	

GENOTYPIC CHANGE

PPARD	Generic Gene Name	Q03181 (http://www.uniprot.org/uniprot/Q03181)	UniProtKB Homo sapiens
FAAR; NUC1; NUCI; NR1C2; NUCII; PPARB	Synonyms	NP_999317 (https://www.ncbi.nlm.nih.gov/nuccore/NP_999317)	GenebankID or UniProtKB
9606.ENSPP00000310928 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000310928)	String		
	Sequence Similarities		
Belongs to the nuclear hormone receptor family. NR1 subfamily.			
	GO - Molecular Function		
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)			
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)			
GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)			
GO:0008134 : transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008134)			
GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)			
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)			
GO:0008289 : lipid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008289)			
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)			
GO:0004879 : nuclear receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004879)			
GO:0001227 : DNA-binding transcription repressor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001227)			
GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001078)			
GO:0051059 : NF-kappaB binding (https://www.ebi.ac.uk/QuickGO/term/GO:0051059)			
GO:0008144 : drug binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008144)			
GO:0005504 : fatty acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005504)			
GO:0038023 : signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0038023)			
GO:0003707 : steroid hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003707)			
GO:0000976 : transcription regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000976)			
GO:0030374 : nuclear receptor transcription coactivator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0030374)			
GO:0001103 : RNA polymerase II repressing transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0001103)			
GO:0070539 : linoleic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070539)			
	GO - Biological Process		
GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)			
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:0006357 : regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006357)			
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)			
GO:0006091 : generation of precursor metabolites and energy (https://www.ebi.ac.uk/QuickGO/term/GO:0006091)			
GO:0009755 : hormone-mediated signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0009755)			
GO:0007507 : heart development (https://www.ebi.ac.uk/QuickGO/term/GO:0007507)			
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:0008203 : cholesterol metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0008203)			
GO:0050728 : negative regulation of inflammatory response (https://www.ebi.ac.uk/QuickGO/term/GO:0050728)			
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)			
GO:0050680 : negative regulation of epithelial cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0050680)			
GO:0006367 : transcription initiation from RNA polymerase II promoter (https://www.ebi.ac.uk/QuickGO/term/GO:0006367)			
GO:0014823 : response to activity (https://www.ebi.ac.uk/QuickGO/term/GO:0014823)			
GO:0048662 : negative regulation of smooth muscle cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0048662)			

GO:0030308 : negative regulation of cell growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030308>)

GO:0071456 : cellular response to hypoxia
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071456>)

GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)

GO:0071222 : cellular response to lipopolysaccharide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071222>)

GO:0009062 : fatty acid catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009062>)

GO:0006631 : fatty acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006631>)

GO:0006629 : lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006629>)

GO:0042060 : wound healing (<https://www.ebi.ac.uk/QuickGO/term/GO:0042060>)

GO:0060612 : adipose tissue development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060612>)

GO:0046697 : decidualization (<https://www.ebi.ac.uk/QuickGO/term/GO:0046697>)

GO:0006006 : glucose metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006006>)

GO:0032024 : positive regulation of insulin secretion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032024>)

GO:0009749 : response to glucose (<https://www.ebi.ac.uk/QuickGO/term/GO:0009749>)

GO:0033993 : response to lipid (<https://www.ebi.ac.uk/QuickGO/term/GO:0033993>)

GO:0014068 : positive regulation of phosphatidylinositol 3-kinase signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014068>)

GO:0045600 : positive regulation of fat cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045600>)

GO:2000288 : positive regulation of myoblast proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:2000288>)

GO:0045662 : negative regulation of myoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045662>)

GO:0032966 : negative regulation of collagen biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032966>)

GO:0097755 : positive regulation of blood vessel diameter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097755>)

GO:0015908 : fatty acid transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015908>)

GO:0010887 : negative regulation of cholesterol storage
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010887>)

GO:1902894 : negative regulation of pri-miRNA transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902894>)

GO:0019216 : regulation of lipid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019216>)

GO:0097190 : apoptotic signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097190>)

GO:0008366 : axon ensheathment (<https://www.ebi.ac.uk/QuickGO/term/GO:0008366>)

GO:0031589 : cell-substrate adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0031589>)

GO:0007566 : embryo implantation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007566>)

GO:0006635 : fatty acid beta-oxidation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006635>)

GO:1904659 : glucose transmembrane transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:1904659>)

GO:0051546 : keratinocyte migration (<https://www.ebi.ac.uk/QuickGO/term/GO:0051546>)

GO:0043616 : keratinocyte proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043616>)

GO:0009299 : mRNA transcription (<https://www.ebi.ac.uk/QuickGO/term/GO:0009299>)

GO:0014912 : negative regulation of smooth muscle cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014912>)

GO:0008654 : phospholipid biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008654>)

GO:0045684 : positive regulation of epidermis development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045684>)

GO:0043415 : positive regulation of skeletal muscle tissue regeneration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043415>)

GO:0006029 : proteoglycan metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006029>)

GO:0014842 : regulation of skeletal muscle satellite cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014842>)

GO:0033189 : response to vitamin A (<https://www.ebi.ac.uk/QuickGO/term/GO:0033189>)

GO:0006776 : vitamin A metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006776>)

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:0090575 : RNA polymerase II transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090575>)

GO:0000790 : nuclear chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000790>)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and*Presumptive Null=~No~#gephebase-summary-title)

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title>)

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

G32E

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Asp	32

Main Reference

A missense mutation in PPARD causes a major QTL effect on ear size in pigs. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21573137>)

Authors

Ren J; Duan Y; Qiao R; Yao F; Zhang Z; Yang B; Guo Y; Xiao S; Wei R; Ouyang Z; Ding N; Ai H; Huang L

Abstract

Chinese Erhualian is the most prolific pig breed in the world. The breed exhibits exceptionally large and floppy ears. To identify genes underlying this typical feature, we previously performed a genome scan in a large scale White Duroc × Erhualian cross and mapped a major QTL for ear size to a 2-cM region on chromosome 7. We herein performed an identical-by-descent analysis that defined the QTL within a 750-kb region. Historically, the large-ear feature has been selected for the ancient sacrificial culture in Erhualian pigs. By using a selective sweep analysis, we then refined the critical region to a 630-kb interval containing 9 annotated genes. Four of the 9 genes are expressed in ear tissues of piglets. Of the 4 genes, PPARD stood out as the strongest candidate gene for its established role in skin homeostasis, cartilage development, and fat metabolism. No differential expression of PPARD was found in ear tissues at different growth stages between large-eared Erhualian and small-eared Duroc pigs. We further screened coding sequence variants in the PPARD gene and identified only one missense mutation (G32E) in a conserved functionally important domain. The protein-altering mutation showed perfect concordance (100%) with the QTL genotypes of all 19 founder animals segregating in the White Duroc × Erhualian cross and occurred at high frequencies exclusively in Chinese large-eared breeds. Moreover, the mutation is of functional significance; it mediates down-regulation of β -catenin and its target gene expression that is crucial for fat deposition in skin. Furthermore, the mutation was significantly associated with ear size across the experimental cross and diverse outbred populations. A worldwide survey of haplotype diversity revealed that the mutation event is of Chinese origin, likely after domestication. Taken together, we provide evidence that PPARD G32E is the variation underlying this major QTL.

Additional References

RELATED GEPHE

Related Genes

2 (HMGA2, WIF1) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9823^/and+Trait=Ear+size/or+Taxon+ID=~9823^/and+Trait=Skin+aspect/or+Taxon+ID=~9825^/and+Trait=Ear+size/or+Taxon+ID=~9825^/and+Trait=Skin+aspect/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001579/9823/>