GEPHE SUMMARY

Gephebase Gene
GCNF (NR6A1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GCNF (NR6A1)^#gephebase-summary-title)

GepheID
GP00000386

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Vertebrae number (https://www.gephebase.org/search-criteria?/and+Trait=^Vertebrae number^#gephebase-summary-title)

Trait State in Taxon A
Sus scrofa

Trait State in Taxon B
Sus scrofa

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?
No

GENOTYPIC CHANGE

Generic Gene Name
Nr6a1

Synonyms
RTR; Gcnf; NCNF; 1700113M01Rik

String
10090.ENSMUSP00000075624 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000075624)

Sequence Similarities
Belongs to the nuclear hormone receptor family. NR6 subfamily.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
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:0004879 : nuclear receptor activity

UniProtKB Mus musculus
Q64249 (http://www.uniprot.org/uniprot/Q64249)

GenebankID or UniProtKB
BAF37829 (https://www.ncbi.nlm.nih.gov/nuccore/BAF37829)

(https://www.ebi.ac.uk/QuickGO/term/GO:0004879)
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:0003707 : steroid hormone receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003707)

GO - Biological Process

GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)
GO:0000122 : negative regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0007283 : spermatogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0007283)
GO:0007276 : gamete generation (https://www.ebi.ac.uk/QuickGO/term/GO:0007276)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0005667 : transcription factor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005667)

Presumptive Null

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

Molecular Type

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

Pro192Leu

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	-	-	-

Main Reference

Fine mapping of a swine quantitative trait locus for number of vertebrae and analysis of an orphan nuclear receptor, germ cell nuclear factor (NR6A1). (2007) (https://pubmed.ncbi.nlm.nih.gov/17416745)

Authors

Mikawa S; Morozumi T; Shimanuki S; Hayashi T; Uenishi H; Domukai M; Okumura N; Awata T

Abstract

The number of vertebrae in pigs varies and is associated with meat productivity. Wild boars, which are ancestors of domestic pigs, have 19 vertebrae. In comparison, European commercial breeds have 21-23 vertebrae, probably owing to selective breeding for enlargement of body size. We previously identified two quantitative trait loci (QTL) for the number of vertebrae on Sus scrofa chromosomes (SSC) 1 and 7. These QTL explained an increase of more than two vertebrae. Here, we performed a map-based study to define the QTL region on SSC1. By using three F2 experimental families, we performed interval mapping and recombination analyses and defined the QTL within a 1.9-cM interval. Then we analyzed the linkage disequilibrium of microsatellite markers in this interval and found that 10 adjacent markers in a 300-kb region were almost fixed in European commercial breeds. Genetic variation of the markers was observed in Asian local breeds or wild boars. This region encoded an orphan nuclear receptor, germ cell nuclear factor (NR6A1, formerly known as GCNF), which contained an amino acid substitution (Pro192Leu) coincident with the QTL. This substitution altered the binding activity of NR6A1 to its corepressors, nuclear receptor-associated protein 80 (RAP80) and nuclear receptor corepressor 1 (NCOR1). In addition, somites of mouse embryos demonstrated expression of NR6A1 protein. Together, these results suggest that NR6A1 is a strong candidate for one of the QTL that influence number of vertebrae in pigs.

Additional References

RELATED GEPHE

Related Genes

2 (Vertnin (VRTN), Vertnin (VRTN) [possibly a pseudo-replicate of other Pig entry]) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9823^/and+Trait=Vertebrae number/or+Taxon ID=^9825^/and+Trait=Vertebrae number/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS