GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00002339

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

WT

Trait State in Taxon B

High fertility in heterozygous females ; sperm defect in homozygous males

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

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 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?

Yes

Taxon B Description

Finnish Yorkshire breed

GENOTYPIC CHANGE

Generic Gene Name

PRLR

Synonyms

HPRL; MFAB; hPRLrI; RI-PRLR

String

9606.ENSP00000371432 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000371432)

Sequence Similarities

Belongs to the type I cytokine receptor family. Type 1 subfamily.

GO - Molecular Function

GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0017046 : peptide hormone binding (https://www.ebi.ac.uk/QuickGO/term/GO:0017046)

UniProtKB Homo sapiens

P16471 (http://www.uniprot.org/uniprot/P16471)

GenebankID or UniProtKB

()

GO:0019955 : cytokine binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019955)
GO:0004896 : cytokine receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004896)
GO:0042978 : ornithine decarboxylase activator activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042978)
GO:0004925 : prolactin receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004925)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0007171 : activation of transmembrane receptor protein tyrosine kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0007171)
GO:0007166 : cell surface receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007166)
GO:0120162 : positive regulation of cold-induced thermogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0120162)
GO:0042976 : activation of Janus kinase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042976)
GO:0060397 : JAK-STAT cascade involved in growth hormone signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0060397)
GO:0042110 : T cell activation (https://www.ebi.ac.uk/QuickGO/term/GO:0042110)
GO:0007566 : embryo implantation (https://www.ebi.ac.uk/QuickGO/term/GO:0007566)
GO:0007595 : lactation (https://www.ebi.ac.uk/QuickGO/term/GO:0007595)
GO:0006694 : steroid biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006694)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0043235 : receptor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0043235)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)
GO:0009897 : external side of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0009897)
GO:0031904 : endosome lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0031904)

Presumptive Null

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

1-10 kb

Molecular Details of the Mutation

L1 insertion in sperm-factor SPEF2 gene causes sperm defect but also downregulation of PRLR

Experimental Evidence

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

Main Reference

A recent L1 insertion within SPEF2 gene is associated with changes in PRLR expression in sow reproductive organs. (2014) (https://pubmed.ncbi.nlm.nih.gov/24712415)

Authors

Sironen A; Fischer D; Laiho A; Gyenesei A; Vilkki J

Abstract

The immotile short tail sperm (ISTS) defect was recognized in the Finnish Yorkshire population at the end of the 1990s when several affected boars were identified. The causal mutation for this defect is a recent L1 insertion within the SPEF2 gene. In 2001, the insertion frequency was already 0.23. Even though all homozygous boars are eliminated from the population due to infertility, the amount of affected boars increased rapidly until marker-assisted selection against the defect was established. Previously we identified an association between the L1 insertion and litter size in the first parity. In this study, we analyzed the expression of the genomic region adjacent to the L1 insertion on porcine chromosome 16. Based on the RNA-seq data analysis, prolactin receptor (PRLR) was identified as down-regulated in the oviduct of ISTS homozygous sows. Quantitative PCR (qPCR) analysis confirmed the significant down-regulation of PRLR in the ovary, oviduct, and uterus of ISTS homozygous and carrier sows compared with controls. In addition, three unannotated loci between PRLR and SPEF2 showed some transcription activity in the analyzed samples. We further investigated the possible mechanisms of the L1 influence on the decrease in the identified gene expression. The methylation pattern of the PRLR gene region appeared unaffected. However, reads mapping to the L1 sequence indicated an increase in L1 antisense promoter expression in the ISTS homozygous animals. The current data suggest that the presence of the L1 affects by some mechanism the expression patterns upstream of the insertion site.

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Additional References

L1 insertion within SPEF2 gene is associated with increased litter size in the Finnish Yorkshire population. (2012) (https://pubmed.ncbi.nlm.nih.gov/22394230)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage