GEPHE SUMMARY

Gephebase Gene
BBS9 (+ BMPER) (https://recette.gephebase.org/search-criteria?/and+Gene Gephebase=^BBS9 (+ BMPER)^#gephebase-summary-title)

Entry Status
Published

GepheID
GP00002341

Main curator
Martin

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
WT

Trait State in Taxon B
Fast growth in heterozygotes ; recessive lethal due to loss of BMPR expression

Ancestral State
Taxon A

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

Taxon A

Latin Name
Sus scrofa domesticus (https://recette.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://recette.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
Large White breed

GENOTYPIC CHANGE

Generic Gene Name
Bbs9

Synonyms
E130103I17Rik; Pthb1

String
10090.ENSMUSP00000116629 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000116629)

Sequence Similarities
-

GO - Molecular Function
-

GO - Biological Process
GO:0060271 : cilium assembly (https://www.ebi.ac.uk/QuickGO/term/GO:0060271)
GO:0061512 : protein localization to cilium (https://www.ebi.ac.uk/QuickGO/term/GO:0061512)

UniProtKB Mus musculus
Q811G0 (http://www.uniprot.org/uniprot/Q811G0)

GenebankID or UniProtKB
()

GO:0045444 : fat cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045444)
GO:0015031 : protein transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015031)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0060170 : ciliary membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0060170)
GO:0005929 : cilium (https://www.ebi.ac.uk/QuickGO/term/GO:0005929)
GO:0034464 : BBSome (https://www.ebi.ac.uk/QuickGO/term/GO:0034464)
GO:0034451 : centriolar satellite (https://www.ebi.ac.uk/QuickGO/term/GO:0034451)
GO:0036064 : ciliary basal body (https://www.ebi.ac.uk/QuickGO/term/GO:0036064)
GO:0035869 : ciliary transition zone (https://www.ebi.ac.uk/QuickGO/term/GO:0035869)
GO:0000242 : pericentriolar material (https://www.ebi.ac.uk/QuickGO/term/GO:0000242)

Presumptive Null

Yes (https://recette.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

Deletion (https://recette.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

100-1000 kb

Molecular Details of the Mutation

212kb deletion resulting in truncated BBS9 protein and recessive loss of neighbor gene expression BMPER

Experimental Evidence

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

Main Reference

Balancing selection on a recessive lethal deletion with pleiotropic effects on two neighboring genes in the porcine genome. (2018) (https://pubmed.ncbi.nlm.nih.gov/30231021)

Authors

Derks MFL; Lopes MS; Bosse M; Madsen O; Dibbits B; Harlizius B; Groenen MAM; Megens HJ

Abstract

Livestock populations can be used to study recessive defects caused by deleterious alleles. The frequency of deleterious alleles including recessive lethal alleles can stay at high or moderate frequency within a population, especially if recessive lethal alleles exhibit an advantage for favourable traits in heterozygotes. In this study, we report such a recessive lethal deletion of 212kb (del) within the BBS9 gene in a breeding population of pigs. The deletion produces a truncated BBS9 protein expected to cause a complete loss-of-function, and we find a reduction of approximately 20% on the total number of piglets born from carrier by carrier matings. Homozygous del/del animals die mid- to late-gestation, as observed from high increase in numbers of mummified piglets resulting from carrier-by-carrier crosses. The moderate 10.8% carrier frequency (5.4% allele frequency) in this pig population suggests an advantage on a favourable trait in heterozygotes. Indeed, heterozygous carriers exhibit increased growth rate, an important selection trait in pig breeding. Increased growth and appetite together with a lower birth weight for carriers of the BBS9 null allele in pigs is analogous to the phenotype described in human and mouse for (naturally occurring) BBS9 null-mutants. We show that fetal death, however, is induced by reduced expression of the downstream BMPER gene, an essential gene for normal foetal development. In conclusion, this study describes a lethal 212kb deletion with pleiotropic effects on two different genes, one resulting in fetal death in homozygous state (BMPER), and the other increasing growth (BBS9) in heterozygous state. We provide strong evidence for balancing selection resulting in an unexpected high frequency of a lethal allele in the population. This study shows that the large amounts of genomic and phenotypic data routinely generated in modern commercial breeding programs deliver a powerful tool to monitor and control lethal alleles much more efficiently.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@BalancingSelection @HeterozygoteAdvantage