

GEPHE SUMMARY

BBS9 (+ BMPER) (https://www.gephebase.org/search-criteria?/and+Gene)	Gephebase Gene	GP00002341	GepheID
Gephebase="BBS9 (+ BMPER)"#gephebase-summary-title)			Main curator
Published	Entry Status	Martin	

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait)	Trait Category		
Category="Physiology"#gephebase-summary-title)			
Growth rate (<a ^growth"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="^Growth)	Trait		
rate"#gephebase-summary-title)			
WT	Trait State in Taxon A		
Fast growth in heterozygotes ; recessive lethal due to loss of BMPR expression	Trait State in Taxon B		
	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic)			
Status="Domesticated"#gephebase-summary-title)			
	Taxon A	Taxon B	
	Latin Name		Latin Name
Sus scrofa domesticus		Sus scrofa domesticus	
(<a ^sus+scrofa"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="^Sus+scrofa)		(<a ^sus+scrofa"="" 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	domesticus"#gephebase-summary-title)	Common Name
domestic pig		domestic pig	
	Synonyms		Synonyms
Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig		Sus domestica; Sus domesticus; Sus scrofa domestica; domestic pig	
	Rank		Rank
subspecies		subspecies	
	Lineage		Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;	
Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;		Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;	
Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;		Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;	
Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; Sus scrofa		Laurasiatheria; Cetartiodactyla; Suina; Suidae; Sus; Sus scrofa	
	Parent		Parent
Sus scrofa (pig) - (Rank: species)		Sus scrofa (pig) - (Rank: species)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	NCBI Taxonomy ID	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	NCBI Taxonomy ID
9825		9825	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	is Taxon A an Intraspecies?	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9825)	is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Large White breed	

GENOTYPIC CHANGE

Bbs9	Generic Gene Name	Q811G0 (http://www.uniprot.org/uniprot/Q811G0)	UniProtKB Mus musculus
E130103117Rik; Pthb1	Synonyms	0	GenebankID or UniProtKB
	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)			

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

Molecular Type

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

Aberration Type

Deletion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title](https://www.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://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping^#gephebase-summary-title](https://www.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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@BalancingSelection @HeterozygoteAdvantage