

GEPHE SUMMARY

B4GALNT2 (https://www.gephebase.org/search-criteria/?and+Gene Gephebase=B4GALNT2#gephebase-summary-title)	Gephebase Gene	GP00001979	GephelD
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Trait Category			
Physiology (https://www.gephebase.org/search-criteria/?and+Trait Category=Physiology#gephebase-summary-title)	Trait		
Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria/?and+Trait=^Fertility+(increased+ovulation+rate)^#gephebase-summary-title)	Trait State in Taxon A		
WT	Trait State in Taxon B		
Lacaune breed with increased litter size	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic Status=Domesticated#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Ovis aries (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Ovis aries#gephebase-summary-title)	Ovis aries (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Ovis aries#gephebase-summary-title)		
sheep	Common Name		Common Name
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	Synonyms
species	Rank		Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis	Lineage
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	Parent	Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	Parent
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	NCBI Taxonomy ID	9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

B4galnt2	Generic Gene Name	UniProtKB Mus musculus
Dlb1; Ggm3; Dlb-1; Galg2; Galgt2; Igf2bp1; Al593864	Synonyms	GenebankID or UniProtKB
10090.ENSMUSP00000037239 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000037239)	String	
Belongs to the glycosyltransferase 2 family.	Sequence Similarities	
GO:0008376 : acetylgalactosaminyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008376)	GO - Molecular Function	
GO:0030259 : lipid glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0030259)	GO - Biological Process	
GO:0006486 : protein glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0006486)		
GO:0022408 : negative regulation of cell-cell adhesion		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0022408>)
GO:0019276 : UDP-N-acetylgalactosamine metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019276>)
GO:0006047 : UDP-N-acetylglucosamine metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006047>)

GO - Cellular Component

GO:0030173 : integral component of Golgi membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030173>)

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

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

Molecular Details of the Mutation

candidate region to 197kb : putative mutation (g.36938224T>A) in intron 7 or (g.37034573A>G) 96 kb away in an intergenic region

Experimental Evidence

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

Main Reference

The highly prolific phenotype of Lacaune sheep is associated with an ectopic expression of the B4GALNT2 gene within the ovary. (2013) (<https://pubmed.ncbi.nlm.nih.gov/24086150>)

Authors

Drouilhet L; Mansanet C; Sarry J; Tabet K; Bardou P; Woloszyn F; Lluch J; Harichaux G; Viguié C; Monniaux D; Bodin L; Mulsant P; Fabre S

Abstract

Prolific sheep have proven to be a valuable model to identify genes and mutations implicated in female fertility. In the Lacaune sheep breed, large variation in litter size is genetically determined by the segregation of a fecundity major gene influencing ovulation rate, named FecL and its prolific allele FecL(L). Our previous work localized FecL on sheep chromosome 11 within a locus of 1.1 Mb encompassing 20 genes. With the aim to identify the FecL gene, we developed a high throughput sequencing strategy of long-range PCR fragments spanning the locus of FecL(L) carrier and non-carrier ewes. Resulting informative markers defined a new 194.6 kb minimal interval. The reduced FecL locus contained only two genes, insulin-like growth factor 2 mRNA binding protein 1 (IGF2BP1) and beta-1,4-N-acetyl-galactosaminyl transferase 2 (B4GALNT2), and we identified two SNP in complete linkage disequilibrium with FecL(L). B4GALNT2 appeared as the best positional and expressional candidate for FecL, since it showed an ectopic expression in the ovarian follicles of FecL(L)/FecL(L) ewes at mRNA and protein levels. In FecL(L) carrier ewes only, B4GALNT2 transferase activity was localized in granulosa cells and specifically glycosylated proteins were detected in granulosa cell extracts and follicular fluids. The identification of these glycoproteins by mass spectrometry revealed at least 10 proteins, including inhibin alpha and betaA subunits, as potential targets of B4GALNT2 activity. Specific ovarian protein glycosylation by B4GALNT2 is proposed as a new mechanism of ovulation rate regulation in sheep, and could contribute to open new fields of investigation to understand female infertility pathogenesis.

Additional References

RELATED GEPHE

3 (BMP receptor IB (BMPRIB), BMP15, GDF9) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=^9940^/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title>)

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001885/9940/>