GEPHE SUMMARY

Gephebase Gene
B4GALNT2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^B4GALNT2^#gephebase-summary-title)

GepheID
GP00001979

Entry Status
Published

Main curator
Martin

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
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name
sheep

Synonyms
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Artiodactyla; Ruminantia; Pecora; Bovidae; Caprinae; Ovis

Parent
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
B4galnt2

Synonyms
Dlb1; Ggm3; Dlb-1; Galg2; Galgt2; Igf2bp1; AI593864

String
10090.ENSMUSP00000037239 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000037239)

Sequence Similarities
Belongs to the glycosyltransferase 2 family.

GO - Molecular Function
GO:0008376 : acetylgalactosaminyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008376)

GO - Biological Process
GO:0030259 : lipid glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0030259)
GO:0006486 : protein glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0006486)
GO:0022408 : negative regulation of cell-cell adhesion

UniProtKB Mus musculus
Q09199 (http://www.uniprot.org/uniprot/Q09199)

GenebankID or UniProtKB
()

(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 7or (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; ViguiÃ© 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

Related Genes

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 Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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