

GEPHE SUMMARY

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

<p>Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=Physiology#gephebase-summary-title)</p> <p>Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria?/and+Trait=Fertility (increased ovulation rate)#gephebase-summary-title)</p> <p>WT</p> <p>Lacaune breed with increased litter size</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Domesticated#gephebase-summary-title)</p>		<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<table border="0" style="width: 100%;"> <tr> <td style="width: 50%; text-align: center;">Taxon A</td> <td style="width: 50%; text-align: center;">Taxon B</td> </tr> <tr> <td style="vertical-align: top;"> <p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p> </td> <td style="vertical-align: top;"> <p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p> </td> </tr> </table>		Taxon A	Taxon B	<p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>B4galnt2</p> <p>Dlb1; Ggm3; Dlb-1; Galg2; Galgt2; Igf2bp1; AL593864</p> <p>10090.ENSMUSP00000037239 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000037239)</p> <p>Belongs to the glycosyltransferase 2 family.</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>Q09199 (http://www.uniprot.org/uniprot/Q09199)</p> <p>()</p> <p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
<p>GO:0008376 : acetylglucosaminyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008376)</p> <p>GO:0030259 : lipid glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0030259)</p> <p>GO:0006486 : protein glycosylation (https://www.ebi.ac.uk/QuickGO/term/GO:0006486)</p> <p>GO:0022408 : negative regulation of cell-cell adhesion</p>		

(<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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

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

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

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; Vigui  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 (BMPRI3), BMP15, GDF9) ([https://www.gephebase.org/search-criteria?/or+Taxon ID="+9940^/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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