

GEPHE SUMMARY

Gephebase Gene
BMP15

Entry Status
Published

GepheID
GP00002158

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Fertility (increased ovulation rate)

Trait State in Taxon A
Ovis aries

Trait State in Taxon B
Ovis aries - Rasa Aragonesa - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Ovis aries

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)

NCBI Taxonomy ID
9940

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Ovis aries

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)

NCBI Taxonomy ID
9940

is Taxon B an Intraspecies?
Yes

Taxon B Description
Rasa Aragonesa

GENOTYPIC CHANGE

Generic Gene Name
Bmp15

Synonyms
Bmp-15; C86824; C87336; GDF-9B; AU015375; AU018861; AU021453; Gdf9b

String
10090.ENSMUSP00000024049

Sequence Similarities
Belongs to the TGF-beta family.

GO - Molecular Function
GO:0005125 : cytokine activity
GO:0008083 : growth factor activity
GO:0005160 : transforming growth factor beta receptor binding

GO - Biological Process
GO:0045893 : positive regulation of transcription, DNA-templated
GO:0001541 : ovarian follicle development

UniProtKB Mus musculus
Q9Z0L4

GenebankID or UniProtKB
AHB23439

GO:0030509 : BMP signaling pathway
GO:0048468 : cell development
GO:0060016 : granulosa cell development
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
GO:0042981 : regulation of apoptotic process
GO:0043408 : regulation of MAPK cascade
GO:0060395 : SMAD protein signal transduction

GO - Cellular Component

GO:0005737 : cytoplasm
GO:0005615 : extracellular space

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

Deletion

Deletion Size

10-99 bp

Molecular Details of the Mutation

c.525_541del17 p.Pro45Asnfs*54

Experimental Evidence

Candidate Gene

Main Reference

A deletion in the bone morphogenetic protein 15 gene causes sterility and increased prolificacy in Rasa Aragonesa sheep. (2008)

Authors

Martinez-Royo A; Jurado JJ; Smulders JP; Mart   JI; Alabart JL; Roche A; Fantova E; Bodin L; Mulsant P; Serrano M; Folch J; Calvo JH

Abstract

Bone morphogenetic protein 15 (BMP15) is a member of the transforming growth factor beta superfamily, is specifically expressed in oocytes and is essential for sheep prolificacy. Reported mutations in this gene cause increased ovulation rate and infertility in a dosage-sensitive manner. In this work, a new naturally occurring mutation in the BMP15 gene from the ovine Rasa Aragonesa breed is described. This mutation is a deletion of 17 bp that leads to an altered amino acid sequence and introduces a premature stop codon in the protein. Highly significant associations ($P < 0.0001$) were found between the estimated breeding value for prolificacy and the genotype of BMP15 in Rasa Aragonesa animals with high and low breeding values for this trait. As for other mutations in BMP15, this new mutation is associated with increased prolificacy and sterility in heterozygous and homozygous ewes respectively.

Additional References

A 17 bp deletion in the Bone Morphogenetic Protein 15 (BMP15) gene is associated to increased prolificacy in the Rasa Aragonesa sheep breed. (2009)

RELATED GEPHE

Related Genes

3 (B4GALNT2, BMP receptor IB (BMPRII), GDF9)

Related Haplotypes

9

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @SexualTrait <https://omia.org/OMIA001800/9940/>