

GEPHE SUMMARY

Gephebase Gene
BMP15

Entry Status
Published

GepheID
GP00002159

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 - Barbarine - 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
Barbarine

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

Insertion

Insertion Size

1-9 bp

Molecular Details of the Mutation

C insertion (c.310insC) in the ovine BMP15 cDNA leading to a frame shift at protein position 101

Experimental Evidence

Candidate Gene

Main Reference

FecX a Novel BMP15 mutation responsible for prolificacy and female sterility in Tunisian Barbarine Sheep. (2017)

Authors

Lassoued N; Benkhilil Z; Woloszyn F; Rejeb A; Aouina M; Rekik M; Fabre S; Bedhiaf-Romdhani S

Abstract

Naturally occurring mutations in growth and differentiation factor 9 (GDF9) or bone morphogenetic protein 15 (BMP15) genes are associated with increased ovulation rate (OR) and litter size (LS) but also sterility. Observing the Tunisian Barbarine ewes of the "W" flock selected for improved prolificacy, we found prolific and infertile ewes with streaky ovaries. Blood genomic DNA was extracted from a subset of low-ovulating, prolific and infertile ewes of the "W" flock, and the entire coding sequences of GDF9 and BMP15 were sequenced.

We evidenced a novel polymorphism in the exon 1 of the BMP15 gene associated with increased prolificacy and sterility. This novel mutation called FecX is a composite polymorphism associating a single nucleotide substitution (c.301G>T), a 3 bp deletion (c.302_304delCTA) and a C insertion (c.310insC) in the ovine BMP15 cDNA leading to a frame shift at protein position 101. Calculated in the "W" flock, the FecX allele increased OR by 0.7 ova and LS by 0.3 lambs ($p = 0.08$). As for already identified mutations, homozygous females carrying FecX exhibited streaky ovaries with a blockade at the primary stage of folliculogenesis as shown by histochemistry.

Our investigation demonstrates a new mutation in the BMP15 gene providing a valuable genetic tool to control fecundity in Tunisian Barbarine, usable for diffusion program into conventional flocks looking for prolificacy improvement.

Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

9

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @SexualTrait <https://omia.org/OMIA002107/9940/>