GEPHE SUMMARY

Gephebase Gene
BMP15

GephelD
GP00002155

Entry Status
Published

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

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 Infraspecies?
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 Infraspecies?
Yes

Taxon B Description
Ovis aries - Galway FecX(G) - High fecundity in heterozygotes

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
GO:0030509 : BMP signaling pathway

UniProtKB Mus musculus
Q9Z0L4

GenebankID or UniProtKB
AHB23439

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
SNP

SNP Coding Change
Nonsense

Molecular Details of the Mutation
c.718C>T ; p.Q239*

Experimental Evidence
Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gln	STP	239

Main Reference
Mutations in the genes for oocyte-derived growth factors GDF9 and BMP15 are associated with both increased ovulation rate and sterility in Cambridge and Belclare sheep (Ovis aries). (2004)

Authors
Hanrahan JP; Gregan SM; Mulsant P; Mullen M; Davis GH; Powell R; Galloway SM

Abstract
Belclare and Cambridge are prolific sheep breeds, the origins of which involved selecting ewes with exceptionally high litter size records from commercial flocks. The variation in ovulation rate in both breeds is consistent with segregation of a gene (or genes) with a large effect on this trait. Sterile ewes, due to a failure of normal ovarian follicle development, occur in both breeds. New naturally occurring mutations in genes for the oocyte-derived growth factors growth differentiation factor 9 (GDF9) and bone morphogenetic protein 15 (BMP15) are described. These mutations are associated with increased ovulation rate in heterozygous carriers and sterility in homozygous carriers in both breeds. This is the first time that a mutation in the gene for GDF9 has been found that causes increased ovulation rate and infertility in a manner similar to inactivating mutations in BMP15, and shows that GDF9 is essential for normal folliculogenesis in sheep. Furthermore, it is shown, for the first time in any species, that individuals with mutations in both GDF9 and BMP15 have a greater ovulation rate than sheep with either of the mutations separately.

Additional References

RELATED GEPHE

Related Genes
3 (B4GALNT2, BMP receptor IB (BMPRIB), GDF9)
Related Haplotypes
9

EXTERNAL LINKS

COMMENTS

Heterozygote shows phenotype; homozygote results in ovarian failure ; @HeterozygoteAdvantage @SexualTrait https://omia.org/OMIA000384/9940/