GEPHE SUMMARY

Gephebase Gene

GDF9 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GDF9^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002188

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

Ovis aries

Trait State in Taxon B

Ovis aries - Vacaria, FecG(V) - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure

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?

Yes

Taxon B Description

Ovis aries - Ile de France

GENOTYPIC CHANGE

Generic Gene Name

GDF9

Synonyms

POF14

String

9606.ENSP00000296875 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296875)

Sequence Similarities

Belongs to the TGF-beta family.

GO - Molecular Function

GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)
GO:0005160 : transforming growth factor beta receptor binding

UniProtKB Homo sapiens

O60383 (http://www.uniprot.org/uniprot/O60383)

GenebankID or UniProtKB

CCI87994 (https://www.ncbi.nlm.nih.gov/nuccore/CCI87994)

(https://www.ebi.ac.uk/QuickGO/term/GO:0005160)

GO - Biological Process

GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0030509 : BMP signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0030509)
GO:0048468 : cell development (https://www.ebi.ac.uk/QuickGO/term/GO:0048468)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010862)
GO:0042981 : regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042981)
GO:0043408 : regulation of MAPK cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0043408)
GO:0060395 : SMAD protein signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0060395)
GO:0030308 : negative regulation of cell growth
(https://www.ebi.ac.uk/QuickGO/term/GO:0030308)
GO:0007179 : transforming growth factor beta receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0007179)
GO:0007292 : female gamete generation
(https://www.ebi.ac.uk/QuickGO/term/GO:0007292)
GO:0001555 : oocyte growth (https://www.ebi.ac.uk/QuickGO/term/GO:0001555)
GO:2000870 : regulation of progesterone secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:2000870)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

g.41841453C>T c.943C>T p.R315C

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Cys	315

Main Reference

Mutation in the protease cleavage site of GDF9 increases ovulation rate and litter size in heterozygous ewes and causes infertility in homozygous ewes. (2014)
(https://pubmed.ncbi.nlm.nih.gov/25039891)

Authors

Souza CJ; McNeilly AS; Benavides MV; Melo EO; Moraes JC

Abstract

Litter size (LS) in sheep is determined mainly by ovulation rate (OR). Several polymorphisms have been identified in the growth differentiation factor 9 (GDF9) gene that result in an increase in OR and prolificacy of sheep. Screening the databank of the Brazilian Sheep Breeders Association for triplet delivery, we identified flocks of prolific Ile de France ewes. After resequencing of GDF9, a point mutation (c.943C>T) was identified, resulting in a non-conservative amino acid change (p.Arg315Cys) in the cleavage site of the propeptide. This new allele was called Vacaria (FecG(v)). A flock of half-sib ewes was evaluated for OR in the first three breeding seasons, and Vacaria heterozygotes had higher OR (PÂ <Â 0.001), averaging 2.1Â Â±Â 0.1 when compared to 1.2Â Â±Â 0.1 in wild-type ewes. The OR was also influenced by age, increasing in the second and third breeding seasons (PÂ <Â 0.001). In flocks segregating this allele, the LS was higher in mutant sheep (PÂ <Â 0.001), averaging 1.61Â Â±Â 0.07 in heterozygotes and 1.29Â Â±Â 0.03 in wild-type ewes. Analysis of homozygote reproductive tract morphology revealed uterine and ovarian hypoplasia. Ovarian follicles continue to develop up to small antral stages, although with abnormal oocyte morphology and altered arrangement of granulosa cells. After the collapse of the oocyte in most follicles, the remaining cells formed clusters that persisted in the ovary. This SNP is useful to improve selection for dam prolificacy and also as a model to investigate GDF9 post-translation processing and the fate of the follicular cells that remain after the oocyte demise.

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Additional References

RELATED GEPHE

Related Genes

3 (B4GALNT2, BMP receptor IB (BMPRIB), BMP15) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9940^/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

4 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^GDF9^/and+Taxon ID=^9940^/or+Gene Gephebase=^GDF9^/and+Taxon ID=^9940^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage ; https://omia.org/OMIA001961/9940/