

GEPHE SUMMARY

	Gephebase Gene	GephelD
GDF9 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GDF9^#gephebase-summary-title)	GP00002188	
	Entry Status	Main curator
Published	Martin	

PHENOTYPIC CHANGE

	Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait

	Trait State in Taxon A
Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria?/and+Trait criteria?/and+Trait=^Fertility (increased ovulation rate)^#gephebase-summary-title)	

	Ancestral State
Ovis aries	Trait State in Taxon B

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

	Taxon A	Taxon B	Latin Name
Ovis aries			

	Taxon A	Taxon B	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title)			

	Taxon A	Taxon B	Latin Name
Ovis aries			

	Taxon A	Taxon B	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Ovis+aries^#gephebase-summary-title)			

	Taxon A	Taxon B	Latin Name
Ovis aries			

	Taxon A	Taxon B	Latin Name
sheep			

	Taxon A	Taxon B	Latin Name
Synonyms			

	Taxon A	Taxon B	Latin Name
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758			

	Taxon A	Taxon B	Latin Name
Rank			

	Taxon A	Taxon B	Latin Name
species			

	Taxon A	Taxon B	Latin Name
Lineage			

	Taxon A	Taxon B	Latin Name
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			

	Taxon A	Taxon B	Latin Name
Parent			

	Taxon A	Taxon B	Latin Name
Ovis () - (Rank: genus)			

	Taxon A	Taxon B	Latin Name
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)			

	Taxon A	Taxon B	Latin Name
NCBI Taxonomy ID			

	Taxon A	Taxon B	Latin Name
9940			

	Taxon A	Taxon B	Latin Name
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)			

	Taxon A	Taxon B	Latin Name
is Taxon A an Infraspecies?			

	Taxon A	Taxon B	Latin Name
No			

	Taxon A	Taxon B	Latin Name
Yes			

	Taxon A	Taxon B	Latin Name
Ovis aries - Ile de France			

	Taxon A	Taxon B	Latin Name
Taxon B Description			

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
GDF9		O60383 (http://www.uniprot.org/uniprot/O60383)

	Synonyms	GenebankID or UniProtKB
POF14		CC187994 (https://www.ncbi.nlm.nih.gov/nuccore/CC187994)

	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 - Molecular Function	
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)		

	GO - Molecular Function	
GO:0005160 : transforming growth factor beta receptor binding		

(<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 (P < 0.001), averaging 2.1 ± 0.1 when compared to 1.2 ± 0.1 in wild-type ewes. The OR was also influenced by age, increasing in the second and third breeding seasons (P < 0.001). In flocks segregating this allele, the LS was higher in mutant sheep (P < 0.001), averaging 1.61 ± 0.07 in heterozygotes and 1.29 ± 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 1B (BMPR1B), 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/>