

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology~#gephebase-summary-title)		Trait Category	
Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria?/and+Trait=~Fertility (increased ovulation rate)~#gephebase-summary-title)		Trait	
Ovis aries		Trait State in Taxon A	
Ovis aries - Vacaria, FecG(V) - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Ovis aries~#gephebase-summary-title)		Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Ovis aries~#gephebase-summary-title)	
sheep		sheep	
Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758		Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	
species		species	
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		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	
Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)		Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	
9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)		9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	
No		Yes	
		Ovis aries - Ile de France	

GENOTYPIC CHANGE

GDF9	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	O60383 (http://www.uniprot.org/uniprot/O60383)
	String	CCl87994 (https://www.ncbi.nlm.nih.gov/nuccore/CCl87994)
9606.ENSPO0000296875 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000296875)		
Belongs to the TGF-beta family.		
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		

(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)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title)	Aberration Type
Nonsynonymous	SNP Coding Change
g.41841453C>T c.943C>T p.R315C	Molecular Details of the Mutation
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	Experimental Evidence

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

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)	Main Reference
Souza CJ; McNeilly AS; Benavides MV; Melo EO; Moraes JC	Authors

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 ($P \leq 0.001$), 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 ($P \leq 0.001$). In flocks segregating this allele, the LS was higher in mutant sheep ($P \leq 0.001$), 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

3 (B4GALNT2, BMP receptor IB (BMPRII), BMP15) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~9940~/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
4 (https://www.gephebase.org/search-criteria?/or+Gene Gephabase=~GDF9~/and+Taxon ID=~9940~/or+Gene Gephabase=~GDF9~/and+Taxon ID=~9940~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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