

GEPHE SUMMARY

GDF9 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~GDF9~#gephebase-summary-title)	Gephebase Gene	GP00000390	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 - Brazilian Santa		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		Ovis aries	
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Ovis aries~#gephebase-summary-title)		(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)		Ovis () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	
9940		9940	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Ovis aries - Brazilian Santa	

GENOTYPIC CHANGE

GDF9		Generic Gene Name	UniProtKB Homo sapiens
POF14		Synonyms	GenebankID or UniProtKB
9606.ENSP00000296875		String	CCl87994 (https://www.ncbi.nlm.nih.gov/nuccore/CCl87994)
(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 (https://www.ebi.ac.uk/QuickGO/term/GO:0005160)			

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
F345C	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	Phe	Cys	345

A new polymorphism in the Growth and Differentiation Factor 9 (GDF9) gene is associated with increased ovulation rate and prolificacy in homozygous sheep. (2011) (https://pubmed.ncbi.nlm.nih.gov/20528846)	Main Reference
Silva BD; Castro EA; Souza CJ; Paiva SR; Sartori R; Franco MM; Azevedo HC; Silva TA; Vieira AM; Neves JP; Melo EO	Authors
Brazilian Santa In��s (SI) sheep are very well-adapted to the tropical conditions of Brazil and are an important source of animal protein. A high rate of twin births was reported in some SI flocks. Growth and Differentiation Factor 9 (GDF9) and Bone Morphogenetic Protein 15 (BMP15) are the first two genes expressed by the oocyte to be associated with an increased ovulation rate in sheep. All GDF9 and BMP15 variants characterized, until now, present the same phenotype: the heterozygote ewes have an increased ovulation rate and the mutated homozygotes are sterile. In this study, we have found a new allele of GDF9, named FecG(E) (Embrapa), which leads to a substitution of a phenylalanine with a cysteine in a conservative position of the mature peptide. Homozygote ewes presenting the FecG(E) allele have shown an increase in their ovulation rate (82%) and prolificacy (58%). This new phenotype can be very useful in better understanding the genetic control of follicular development; the mechanisms involved in the control of ovulation rate in mammals; and for the improvement of sheep production.	Abstract
�� 2010 The Authors, Animal Genetics �� 2010 Stichting International Foundation for Animal Genetics.	

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 Gephebase=~GDF9~/and+Taxon ID=~9940~/or+Gene Gephebase=~GDF9~/and+Taxon ID=~9940~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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