

GEPHE SUMMARY

GDF9 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=GDF9^#gephebase-summary-title)	Gephebase Gene	GP00002187	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 - High fertility, FecG(H) - 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		No	

GENOTYPIC CHANGE

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

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

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

Nonsynonymous

g.41841212C>T c.1184C>T p.S395F

Linkage Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Linkage Mapping~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ser	Phe	395

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) (<https://pubmed.ncbi.nlm.nih.gov/14627550>)

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

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.

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>)

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/OMIA001801/9940/>