

GEPHE SUMMARY

	Gephebase Gene		GepheID
GDF9 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~GDF9~#gephebase-summary-title)		GP00000391	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~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 - Icelandic Thoka - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure			
	Ancestral State		
Data not curated			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated~#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
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)	
	Common Name		Common Name
sheep		sheep	
	Synonyms		Synonyms
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	
	Rank		Rank
species		species	
	Lineage		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		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		Parent
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)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
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)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		Yes	
			Taxon B Description
		Ovis aries - Icelandic Thoka	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
GDF9		O60383 (http://www.uniprot.org/uniprot/O60383)	
	Synonyms		GenebankID or UniProtKB
POF14		CCl87994 (https://www.ncbi.nlm.nih.gov/nuccore/CCl87994)	
	String		
9606.ENSPO00000296875 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO00000296875)			
	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 - 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.41841117A>C c.1279A>C p.S427R	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Linkage+Mapping~#gephebase-summary-title)	Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ser	Arg	427

Homozygosity for a single base-pair mutation in the oocyte-specific GDF9 gene results in sterility in Thoka sheep. (2009) (https://pubmed.ncbi.nlm.nih.gov/19713444)	Main Reference
Nicol L; Bishop SC; Pong-Wong R; Bendixen C; Holm LE; Rhind SM; McNeilly AS	Authors
The control of fecundity is critical in determining mammalian offspring survival. It is regulated principally by the ovulation rate, so that primates and large farm species commonly have a single offspring. Previously, several mutations have been identified in sheep which increase the naturally low ovulation rate; although in some cases homozygous ewes are infertile. In the present study we present a detailed characterization of a novel mutation in growth differentiation factor 9 (GDF9), found in Icelandic Thoka sheep. This mutation is a single base change (A1279C) resulting in a nonconservative amino acid change (S109R) in the C-terminus of the mature GDF9 protein, which is normally expressed in oocytes at all stages of development. Genotyping all animals for which reproductive records were available confirmed this mutation to be associated with increased fecundity in heterozygous ewes and infertility in homozygotes. Analysis of homozygote ovarian morphology and a number of genes normally activated in growing follicles showed that GDF9 was not involved in oocyte activation, but in subsequent development of the follicle. This study highlights the importance of oocyte factors in regulating fertility and provides new information for structural analysis and investigation of the potentially important sites of dimerization or translational modifications required to produce biologically active GDF9. It also provides the basis for the utilization of these animals to enhance sheep production.	Abstract
	Additional References

RELATED GEPHE

3 (B4GALNT2, BMP receptor IB (BMPRI1), 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

@HeterozygoteAdvantage ; <https://omia.org/OMIA000385/9940/>