

GEPHE SUMMARY

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

<p>Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=Physiology#gephebase-summary-title)</p> <p>Fertility (increased ovulation rate) (https://www.gephebase.org/search-criteria?/and+Trait=Fertility+increased+ovulation+rate#gephebase-summary-title)</p> <p>Ovis aries</p> <p>Ovis aries - Icelandic Thoka - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure</p> <p>Data not curated</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Domesticated#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Ovis aries</p> <p>(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis+aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>No is Taxon A an Intraspecies?</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Ovis aries</p> <p>(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Ovis+aries#gephebase-summary-title)</p> <p>sheep</p> <p>Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935)</p> <p>9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940)</p> <p>Yes is Taxon B an Intraspecies?</p> <p>Ovis aries - Icelandic Thoka</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
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GENOTYPIC CHANGE

<p>GDF9</p> <p>POF14</p> <p>9606.ENSPO0000296875 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000296875)</p> <p>Belongs to the TGF-beta family.</p> <p>GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)</p> <p>GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)</p> <p>GO:0005160 : transforming growth factor beta receptor binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>O60383 (http://www.uniprot.org/uniprot/O60383)</p> <p>CCl87994 (https://www.ncbi.nlm.nih.gov/nucore/CCl87994)</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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(<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

Main Reference

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>)

Authors

Nicol L; Bishop SC; Pong-Wong R; Bendixen C; Holm LE; Rhind SM; McNeilly AS

Abstract

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.

Additional References

RELATED GEPHE

Related Genes

3 (B4GALNT2, BMP receptor IB (BMPRI), 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/OMIA000385/9940/>