

## GEPHE SUMMARY

BMP15 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase= <sup>^</sup> BMP15 <sup>^</sup> #gephebase-summary-title)	Gephebase Gene	GP00002159	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Physiology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Fertility (increased ovulation rate) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> = <sup>^</sup> Fertility (increased ovulation rate) <sup>^</sup> #gephebase-summary-title)	Trait		
Ovis aries	Trait State in Taxon A		
Ovis aries - Barbarine - Increased ovulation rate; Heterozygote shows phenotype; homozygote results in ovarian failure	Trait State in Taxon B		
	Ancestral State		
Taxon A	Taxonomic Status		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status= <sup>^</sup> Domesticated <sup>^</sup> #gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms</a> = <sup>^</sup> Ovis aries <sup>^</sup> #gephebase-summary-title)		Ovis aries ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms</a> = <sup>^</sup> Ovis aries <sup>^</sup> #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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )	NCBI Taxonomy ID	Ovis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9935</a> )	NCBI Taxonomy ID
9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )		9940 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9940</a> )	
	is Taxon A an Infrappecies?		is Taxon B an Infrappecies?
No		Yes	
			Taxon B Description
		Barbarine	

## GENOTYPIC CHANGE

Bmp15	Generic Gene Name	Q9Z0L4 ( <a href="http://www.uniprot.org/uniprot/Q9Z0L4">http://www.uniprot.org/uniprot/Q9Z0L4</a> )	UniProtKB Mus musculus
Bmp-15; C86824; C87336; GDF-9B; AU015375; AU018861; AU021453; Gdf9b	Synonyms	AHB23439 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AHB23439">https://www.ncbi.nlm.nih.gov/nuccore/AHB23439</a> )	GenebankID or UniProtKB
10090.ENSMUSP00000024049 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000024049">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000024049</a> )	String		
	Sequence Similarities		
Belongs to the TGF-beta family.			
	GO - Molecular Function		
GO:0005125 : cytokine activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005125">https://www.ebi.ac.uk/QuickGO/term/GO:0005125</a> )			
GO:0008083 : growth factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008083">https://www.ebi.ac.uk/QuickGO/term/GO:0008083</a> )			
GO:0005160 : transforming growth factor beta receptor binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0005160>)

GO - Biological Process

GO:0045893 : positive regulation of transcription, DNA-templated

(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)

GO:0001541 : ovarian follicle development

(<https://www.ebi.ac.uk/QuickGO/term/GO:0001541>)

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:0060016 : granulosa cell development

(<https://www.ebi.ac.uk/QuickGO/term/GO:0060016>)

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 - 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>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes\\*#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes*#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding\\*#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding*#gephebase-summary-title))

Aberration Type

Insertion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Insertion\\*#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Insertion*#gephebase-summary-title))

Insertion Size

1-9 bp

Molecular Details of the Mutation

C insertion (c.310insC) in the ovine BMP15 cDNA leading to a frame shift at protein position 101

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene\\*#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene*#gephebase-summary-title))

Main Reference

FecX a Novel BMP15 mutation responsible for prolificacy and female sterility in Tunisian Barbarine Sheep. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28506298>)

Authors

Lassoued N; Benkhilil Z; Woloszyn F; Rejeb A; Aouina M; Rekek M; Fabre S; Bedhiaf-Romdhani S

Abstract

Naturally occurring mutations in growth and differentiation factor 9 (GDF9) or bone morphogenetic protein 15 (BMP15) genes are associated with increased ovulation rate (OR) and litter size (LS) but also sterility. Observing the Tunisian Barbarine ewes of the "W" flock selected for improved prolificacy, we found prolific and infertile ewes with streaky ovaries. Blood genomic DNA was extracted from a subset of low-ovulating, prolific and infertile ewes of the "W" flock, and the entire coding sequences of GDF9 and BMP15 were sequenced.

We evidenced a novel polymorphism in the exon 1 of the BMP15 gene associated with increased prolificacy and sterility. This novel mutation called FecX is a composite polymorphism associating a single nucleotide substitution (c.301G>T), a 3Å bp deletion (c.302\_304delCTA) and a C insertion (c.310insC) in the ovine BMP15 cDNA leading to a frame shift at protein position 101. Calculated in the "W" flock, the FecX allele increased OR by 0.7 ova and LS by 0.3 lambs (p=0.08). As for already identified mutations, homozygous females carrying FecX exhibited streaky ovaries with a blockade at the primary stage of folliculogenesis as shown by histochemistry.

Our investigation demonstrates a new mutation in the BMP15 gene providing a valuable genetic tool to control fecundity in Tunisian Barbarine, usable for diffusion program into conventional flocks looking for prolificacy improvement.

Additional References

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RELATED GEPHE

Related Genes

3 (B4GALNT2, BMP receptor IB (BMPRII), GDF9) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9940\\*/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9940*/and+Trait=Fertility/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

9 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~BMP15\\*/and+Taxon ID=~9940\\*/or+Gene Gephebase=~BMP15\\*/and+Taxon ID=~9940\\*#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~BMP15*/and+Taxon+ID=~9940*/or+Gene+Gephebase=~BMP15*/and+Taxon+ID=~9940*#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @SexualTrait <https://omia.org/OMIA002107/9940/>

