

GEPHE SUMMARY

<p>FGFR3 (<a +fgfr3+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+FGFR3+"#gephebase-summary-title</a>)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002184</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title</a>)</p> <p>Body size (bone length) (<a +body+size+(bone+length)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Body+size+(bone+length)+"#gephebase-summary-title</a>)</p> <p>Lambs with normal size and skeletal frame</p> <p>Lambs with increased long bone in heterozygotes ; will eventually result in greater muscularity and leanness ; Spider Lamb abnormality in homozygous lambs</p> <p>Taxon A</p> <p>Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title</a>)</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>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>Taxon A Description</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>
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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=))

sheep

Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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

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>)

Yes

Suffolk breed

GENOTYPIC CHANGE

<p>FGFR3</p> <p>ACH; CEK2; JTK4; CD333; HSFGR3EX</p> <p>9606.ENSPO0000339824 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000339824">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000339824</a>)</p> <p>Belongs to the protein kinase superfamily. Tyr protein kinase family. Fibroblast growth factor receptor subfamily.</p> <p>GO:0005524 : ATP binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005524">https://www.ebi.ac.uk/QuickGO/term/GO:0005524</a>)</p> <p>GO:0042802 : identical protein binding</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P22607 (<a href="http://www.uniprot.org/uniprot/P22607">http://www.uniprot.org/uniprot/P22607</a>)</p> <p>()</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0042802>)  
GO:0004714 : transmembrane receptor protein tyrosine kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004714>)  
GO:0004713 : protein tyrosine kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004713>)  
GO:0017134 : fibroblast growth factor binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0017134>)  
GO:0005007 : fibroblast growth factor-activated receptor activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005007>)

#### GO - Biological Process

GO:0007275 : multicellular organism development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)  
GO:0001501 : skeletal system development  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)  
GO:0070374 : positive regulation of ERK1 and ERK2 cascade  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070374>)  
GO:0000165 : MAPK cascade (<https://www.ebi.ac.uk/QuickGO/term/GO:0000165>)  
GO:0008284 : positive regulation of cell proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)  
GO:0051897 : positive regulation of protein kinase B signaling  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)  
GO:0007267 : cell-cell signaling (<https://www.ebi.ac.uk/QuickGO/term/GO:0007267>)  
GO:0043410 : positive regulation of MAPK cascade  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)  
GO:0001958 : endochondral ossification  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001958>)  
GO:0030282 : bone mineralization (<https://www.ebi.ac.uk/QuickGO/term/GO:0030282>)  
GO:0046777 : protein autophosphorylation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046777>)  
GO:0018108 : peptidyl-tyrosine phosphorylation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0018108>)  
GO:0008543 : fibroblast growth factor receptor signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008543>)  
GO:0060349 : bone morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060349>)  
GO:0003416 : endochondral bone growth  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003416>)  
GO:0010518 : positive regulation of phospholipase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010518>)  
GO:0002062 : chondrocyte differentiation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002062>)  
GO:0042531 : positive regulation of tyrosine phosphorylation of STAT protein  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042531>)  
GO:0035988 : chondrocyte proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035988>)  
GO:0070977 : bone maturation (<https://www.ebi.ac.uk/QuickGO/term/GO:0070977>)  
GO:0007169 : transmembrane receptor protein tyrosine kinase signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007169>)  
GO:1902178 : fibroblast growth factor receptor apoptotic signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902178>)  
GO:0048640 : negative regulation of developmental growth  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048640>)  
GO:0033674 : positive regulation of kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033674>)  
GO:0043552 : positive regulation of phosphatidylinositol 3-kinase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043552>)

#### GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)  
GO:0005887 : integral component of plasma membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)  
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)  
GO:0043235 : receptor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0043235>)  
GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)  
GO:0005794 : Golgi apparatus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005794>)  
GO:0005783 : endoplasmic reticulum  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005783>)  
GO:0009986 : cell surface (<https://www.ebi.ac.uk/QuickGO/term/GO:0009986>)  
GO:0030133 : transport vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0030133>)

No ([https://www.gephebase.org/search-criteria?/and+Presumptive+Null="+No+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))  
Coding ([https://www.gephebase.org/search-criteria?/and+Molecular+Type="+Coding+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))  
SNP ([https://www.gephebase.org/search-criteria?/and+Aberration+Type="+SNP+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))  
Nonsynonymous

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

c.1719T>A p.V700E

Experimental Evidence

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Val	Glu	700

Main Reference

A single-base change in the tyrosine kinase II domain of ovine FGFR3 causes hereditary chondrodysplasia in sheep. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16441300>)

Authors

Beever JE; Smit MA; Meyers SN; Hadfield TS; Bottema C; Albretsen J; Cockett NE

Abstract

Ovine hereditary chondrodysplasia, or spider lamb syndrome (SLS), is a genetic disorder that is characterized by severe skeletal abnormalities and has resulted in substantial economic losses for sheep producers. Here we demonstrate that a non-synonymous T>A transversion in the highly conserved tyrosine kinase II domain of a positional candidate gene, fibroblast growth factor receptor 3 (FGFR3), is responsible for SLS. We also demonstrate that the mutant FGFR3 allele has an additive effect on long-bone length, calling into question the long-standing belief that SLS is inherited as a strict monogenic, Mendelian recessive trait. Instead, we suggest that SLS manifestation is determined primarily by the presence of the mutant FGFR3 allele, but it is also influenced by an animal's genetic background. In contrast to FGFR3 mutations causing dwarfism in humans, this single-base change is the only known natural mutation of FGFR3 that results in a skeletal overgrowth phenotype in any species.

Additional References

Enhanced skeletal growth of sheep heterozygous for an inactivated fibroblast growth factor receptor 3. (2006) (<https://pubmed.ncbi.nlm.nih.gov/17032787>)

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage <https://omia.org/OMIA001703/9940/>