

GEPHE SUMMARY

IRF2BP2 (https://www.gephebase.org/search-criteria/?and+Gene+Gephebase=^IRF2BP2^#gephebase-summary-title)	Gephebase Gene	GP00002195	GephelD
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

Trait Category			
Morphology (https://www.gephebase.org/search-criteria/?and+Trait+Category=^Morphology^#gephebase-summary-title)	Trait		
Hair type (woolly) (https://www.gephebase.org/search-criteria/?and+Trait=^Hair+type+(woolly)^#gephebase-summary-title)	Trait State in Taxon A		
Ancestral sheep with long and hairy fleece ; Rustic Romanov breed	Trait State in Taxon B		
Modern breeds with short and woolly fleece	Ancestral State		
Taxon A	Taxonomic Status		
Ovis aries	Domesticated (https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=^Domesticated^#gephebase-summary-title)		
Taxon A	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)	
species	Common Name	Common Name	
sheep	Synonyms	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	Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758	
species	Rank	Rank	
	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	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	
species	Parent	Parent	
Ovis () - (Rank: genus)	Ovis () - (Rank: genus)	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)	NCBI Taxonomy ID 9940	NCBI Taxonomy ID 9940	
is Taxon A an Infraspecies?		is Taxon B an Infraspecies?	
Yes	Taxon A Description	No	
-			

GENOTYPIC CHANGE

Generic Gene Name			
IRF2BP2	Synonyms	Q7Z5L9 (http://www.uniprot.org/uniprot/Q7Z5L9)	UniProtKB Homo sapiens
CVID14	String	0	GenebankID or UniProtKB
9606.ENSP00000355568 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000355568)	Sequence Similarities		
Belongs to the IRF2BP family.	GO - Molecular Function		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)			
GO:0003714 : transcription corepressor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003714)			
GO:0061630 : ubiquitin protein ligase activity			

GO:0006357 : regulation of transcription by RNA polymerase II

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)

GO:0000122 : negative regulation of transcription by RNA polymerase II

(<https://www.ebi.ac.uk/QuickGO/term/GO:0000122>)

GO:0002327 : immature B cell differentiation

(<https://www.ebi.ac.uk/QuickGO/term/GO:0002327>)

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

Presumptive Null

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No))

Molecular Type

Cis-regulatory ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory))

Aberration Type

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

Insertion Size

-

Molecular Details of the Mutation

insertion of an antisense EIF2S2 retrogene (asEIF2S2) into the 3' UTR of the IRF2BP2 gene ; RNA-RNA hybrid creates a long endogenous double-stranded RNA which alters the expression of both EIF2S2 and IRF2BP2 mRNA

Experimental Evidence

Linkage Mapping ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping))

Main Reference

Genome-Wide Identification of the Mutation Underlying Fleece Variation and Discriminating Ancestral Hairy Species from Modern Woolly Sheep. (2017)

(<https://pubmed.ncbi.nlm.nih.gov/28379502>)

Authors

Demars J; Cano M; Drouilhet L; Plisson-Petit F; Bardou P; Fabre S; Servin B; Sarry J; Woloszyn F; Mulsant P; Foulquier D; Carriére F; Aletru M; Rodde N; Cauet S; Bouchez O; Pirson M; Tosser-Klopp G; Allain D

Abstract

The composition and structure of fleece variation observed in mammals is a consequence of a strong selective pressure for fiber production after domestication. In sheep, fleece variation discriminates ancestral species carrying a long and hairy fleece from modern domestic sheep (*Ovis aries*) owning a short and woolly fleece. Here, we report that the "woolly" allele results from the insertion of an antisense EIF2S2 retrogene (called asEIF2S2) into the 3' UTR of the IRF2BP2 gene leading to an abnormal IRF2BP2 transcript. We provide evidence that this chimeric IRF2BP2/asEIF2S2 messenger 1) targets the genuine sense EIF2S2 RNA and 2) creates a long endogenous double-stranded RNA which alters the expression of both EIF2S2 and IRF2BP2 mRNA. This represents a unique example of a phenotype arising via a RNA-RNA hybrid, itself generated through a retroposition mechanism. Our results bring new insights on the sheep population history thanks to the identification of the molecular origin of an evolutionary phenotypic variation.

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Retroposition ; <https://omia.org/OMIA001528/9940/>