

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

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

(<https://www.ebi.ac.uk/QuickGO/term/GO:0061630>)

GO - Biological Process

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 - Cellular Component

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

Molecular Type

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

Aberration Type

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

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

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; Carrière 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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

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