GEPHE SUMMARY

Gephebase Gene

RNF11 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^RNF11^#gephebase-summary-title)

GepheID

GP00002275

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Body size (dwarfism) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (dwarfism)^#gephebase-summary-title)

Trait State in Taxon A

WT

Trait State in Taxon B

Dwarfism and inflammatory lesions in Belgian Blue homozygotes ; those traits are not bred but the heterozygote carriers exist at high frequency

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank

species

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; Bovinae; Bos

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)

Common Name

cattle

Synonyms

Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus

Rank

species

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; Bovinae; Bos

Parent

Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9913)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

RNF11

Synonyms

CGI-123; SID1669

String

9606.ENSP00000242719 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000242719)

Sequence Similarities

-

GO - Molecular Function

GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)
GO:0061630 : ubiquitin protein ligase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0061630)

GO - Biological Process

UniProtKB Homo sapiens

Q9Y3C5 (http://www.uniprot.org/uniprot/Q9Y3C5)

GenebankID or UniProtKB

()

GO:0006511 : ubiquitin-dependent protein catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006511)
GO:0051865 : protein autoubiquitination (https://www.ebi.ac.uk/QuickGO/term/GO:0051865)

GO - Cellular Component

GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0005769 : early endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005769)
GO:0000151 : ubiquitin ligase complex (https://www.ebi.ac.uk/QuickGO/term/GO:0000151)
GO:0055037 : recycling endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0055037)

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

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

Deletion Size

-

Molecular Details of the Mutation

g.95601696A>G c.124-2A>G affecting splicing site

Experimental Evidence

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

Main Reference

A splice site variant in the bovine RNF11 gene compromises growth and regulation of the inflammatory response. (2012) (https://pubmed.ncbi.nlm.nih.gov/22438830)

Authors

Sartelet A; Druet T; Michaux C; Fasquelle C; GÃ©ron S; Tamma N; Zhang Z; Coppieters W; Georges M; Charlier C

Abstract

We report association mapping of a locus on bovine chromosome 3 that underlies a Mendelian form of stunted growth in Belgian Blue Cattle (BBC). By resequencing positional candidates, we identify the causative c124-2A>G splice variant in intron 1 of the RNF11 gene, for which all affected animals are homozygous. We make the remarkable observation that 26% of healthy Belgian Blue animals carry the corresponding variant. We demonstrate in a prospective study design that approximately one third of homozygous mutants die prematurely with major inflammatory lesions, hence explaining the rarity of growth-stunted animals despite the high frequency of carriers. We provide preliminary evidence that heterozygous advantage for an as of yet unidentified phenotype may have caused a selective sweep accounting for the high frequency of the RNF11 c124-2A>G mutation in Belgian Blue Cattle.

Additional References

RELATED GEPHE

Related Genes

5 (aggrecan, GH, LCORL, PLAG1, PRKG2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Splicing @HeterozygoteAdvantage but the selected phenotype in heterozygotes is unknown