

GEPHE SUMMARY

<p>RNF11 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+RNF11+Gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002275</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title)</p> <p>Body size (dwarfism) (https://www.gephebase.org/search-criteria?/and+Trait+Body+size+(dwarfism)+Gephebase-summary-title)</p> <p>WT</p> <p>Dwarfism and inflammatory lesions in Belgian Blue homozygotes ; those traits are not bred but the heterozygote carriers exist at high frequency</p> <p>Taxon A</p> <p>Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title)</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><i>Bos taurus</i> (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Bos+taurus+Gephebase-summary-title)</p> <p>Common Name</p> <p>cattle</p> <p>Synonyms</p> <p><i>Bos bovis</i>; <i>Bos primigenius taurus</i>; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; <i>Bos taurus</i> Linnaeus, 1758; <i>Bos Taurus</i></p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p><i>Bos</i> (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)</p> <p>NCBI Taxonomy ID</p> <p>9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)</p> <p>is Taxon A an Infrapopulation?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p><i>Bos taurus</i> (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Bos+taurus+Gephebase-summary-title)</p> <p>Common Name</p> <p>cattle</p> <p>Synonyms</p> <p><i>Bos bovis</i>; <i>Bos primigenius taurus</i>; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; <i>Bos taurus</i> Linnaeus, 1758; <i>Bos Taurus</i></p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p><i>Bos</i> (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)</p> <p>NCBI Taxonomy ID</p> <p>9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)</p> <p>is Taxon B an Infrapopulation?</p> <p>No</p>
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GENOTYPIC CHANGE

<p>RNF11</p> <p>CGI-123; SID1669</p> <p>9606.ENSP00000242719 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000242719)</p> <p>-</p> <p>GO - Molecular Function</p> <p>GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)</p> <p>GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)</p> <p>GO:0061630 : ubiquitin protein ligase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0061630)</p> <p>GO - Biological Process</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p>	<p>Q9Y3C5 (http://www.uniprot.org/uniprot/Q9Y3C5)</p> <p>0</p>	<p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

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

Aberration Type

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

Deletion Size

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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](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

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](https://www.gephebase.org/search-criteria?/or+Taxon+ID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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