## GEPHE SUMMARY

Gephebase Gene
MRC2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MRC2^#gephebase-summary-title)

GepheID
GP00002265

Entry Status
Published

Main curator
Martin

## PHENOTYPIC CHANGE

Trait #1

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

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

Trait State in Taxon A
Belgian Blue with average muscular mass for this breed

Trait State in Taxon B
Belgian Blue with enhanced muscular development in heterozygotes

Trait #2

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

Trait
Tail shape (crooked ; defect) (https://www.gephebase.org/search-criteria?/and+Trait=^Tail shape (crooked ; defect)^#gephebase-summary-title)

Trait State in Taxon A
WT tail

Trait State in Taxon B
Crooked tail in homozygotes (not a selected trait)

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

MRC2

Synonyms

CD280; UPARAP; CLEC13E; ENDO180; KIAA0709

String

9606.ENSP00000307513 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000307513 )

Sequence Similarities

-

GO - Molecular Function

GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)
GO:0038023 : signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0038023)
GO:0005518 : collagen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005518)

GO - Biological Process

GO:0001649 : osteoblast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0001649)
GO:0006897 : endocytosis (https://www.ebi.ac.uk/QuickGO/term/GO:0006897)
GO:0030574 : collagen catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0030574)

GO - Cellular Component

GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005925 : focal adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0005925)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)

UniProtKB Homo sapiens

Q9UBG0 (http://www.uniprot.org/uniprot/Q9UBG0)

GenebankID or UniProtKB

()

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

-

Molecular Details of the Mutation

c.1906T>G p.C636G

Experimental Evidence

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

| | Taxon A | Taxon B | Position |
|---|---|---|---|
| Codon | - | - | - |
| Amino-acid | Cys | Gly | 636 |

Main Reference

Balancing selection of a frame-shift mutation in the MRC2 gene accounts for the outbreak of the Crooked Tail Syndrome in Belgian Blue Cattle. (2009) (https://pubmed.ncbi.nlm.nih.gov/19779552)

Authors

Fasquelle C; Sartelet A; Li W; Dive M; Tamma N; Michaux C; Druet T; Huijbers IJ; Isacke CM; Coppieters W; Georges M; Charlier C

Abstract

We herein describe the positional identification of a 2-bp deletion in the open reading frame of the MRC2 receptor causing the recessive Crooked Tail Syndrome in cattle. The resulting frame-shift reveals a premature stop codon that causes nonsense-mediated decay of the mutant messenger RNA, and the virtual absence of functional Endo180 protein in affected animals. Cases exhibit skeletal anomalies thought to result from impaired extracellular matrix remodeling during ossification, and as of yet unexplained muscular symptoms. We demonstrate that carrier status is very significantly associated with desired characteristics in the general population, including enhanced muscular development, and that the resulting heterozygote advantage caused a selective sweep which explains the unexpectedly high frequency (25%) of carriers in the Belgian Blue Cattle Breed.

Additional References

Allelic heterogeneity of Crooked Tail Syndrome: result of balancing selection?. (2012) (https://pubmed.ncbi.nlm.nih.gov/22497452)
Selection in action: dissecting the molecular underpinnings of the increasing muscle mass of Belgian Blue Cattle. (2014) (https://pubmed.ncbi.nlm.nih.gov/25228463)

## RELATED GEPHE

Related Genes

1 (Myostatin (MSTN = GDF8)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Muscular mass/or+Taxon ID=^9913^/and+Trait=Tail shape/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^MRC2^/and+Taxon ID=^9913^/or+Gene Gephebase=^MRC2^/and+Taxon ID=^9913^#gephebase-summary-title)

## EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage @AllelicSeries @BalancingSelection compound heterozygotes of two alleles exist ; https://omia.org/OMIA001452/9913/