

GEPHE SUMMARY

MRC2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=MRC2#gephebase-summary-title)	Gephebase Gene	GP00002264	GepheID
Published	Entry Status	Martin	Main curator

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)	

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

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 Taurus	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 Intraspecies?
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 Taurus	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 Intraspecies?
No	

GENOTYPIC CHANGE

MRC2	Generic Gene Name	Q9UBG0 (http://www.uniprot.org/uniprot/Q9UBG0)	UniProtKB Homo sapiens
CD280; UPARAP; CLEC13E; ENDO180; KIAA0709	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000307513 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000307513)	String		
-	Sequence Similarities		
GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)	GO - Molecular Function		
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:0001649 : osteoblast differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0001649)	GO - Biological Process		
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:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)	GO - Cellular Component		
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)			
Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)			Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)			Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)			Aberration Type
1-9 bp			Deletion Size
c.2904-2905delAG p.Gly934X			Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)			Experimental Evidence
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)			Main Reference
Fasquelle C; Sartelet A; Li W; Dive M; Tamma N; Michaux C; Druet T; Huijbers IJ; Isacke CM; Coppieters W; Georges M; Charlier C			Authors
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.			Abstract
Allelic heterogeneity of Crooked Tail Syndrome: result of balancing selection?. (2012) (https://pubmed.ncbi.nlm.nih.gov/22497452)			Additional References
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

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 Genes
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)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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

