

GEPHE SUMMARY

MRC2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase="MRC2">#gephebase-summary-title)	Gephebase Gene GP00002264	GephelD
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	Taxon B	Latin Name
Bos taurus (#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Bos taurus">#gephebase-summary-title)	Common Name	Bos taurus (#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=Bos taurus">#gephebase-summary-title)	Common Name
cattle	Synonyms	cattle	Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taururus	Rank	Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taururus	Rank
species	Lineage	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	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	Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)	NCBI Taxonomy ID 9913
	is Taxon A an Infraspecies?		is Taxon B an Infraspecies?
No	No		

GENOTYPIC CHANGE

MRC2	Generic Gene Name	UniProt Homo sapiens
CD280; UPARAP; CLEC13E; ENDO180; KIAA0709	Synonyms	GenebankID or UniProtKB
9606.ENSP00000307513 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000307513)	String	
	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)		
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	
All 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/>

