GEPHE SUMMARY

Gephebase Gene
Twist1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Twist1^#gephebase-summary-title)

GepheID
GP00002034

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
Bos taurus

Trait State in Taxon B
Bos taurus - horn abnormalities similar to classical scurs phenotype and skull interfrontal suture synostosis

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
Twist1

Synonyms
Pde; pdt; Ska10; Twist; M-Twist; bHLHa38; Ska<m10Jus>

String
10090.ENSMUSP00000040089 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000040089)

Sequence Similarities
-

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001228)
GO:0003700 : DNA-binding transcription factor activity

UniProtKB Mus musculus
P26687 (http://www.uniprot.org/uniprot/P26687)

GenebankID or UniProtKB
()

(https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0046982 : protein heterodimerization activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0008134 : transcription factor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0008134)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0043425 : bHLH transcription factor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0043425)
GO:0070888 : E-box binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070888)
GO:0019904 : protein domain specific binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0019904)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)
GO:0030326 : embryonic limb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0030326)
GO:0048704 : embryonic skeletal system morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048704)
GO:0000122 : negative regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)
GO:0045892 : negative regulation of transcription, DNA-templated
(https://www.ebi.ac.uk/QuickGO/term/GO:0045892)
GO:0001843 : neural tube closure (https://www.ebi.ac.uk/QuickGO/term/GO:0001843)
GO:0010718 : positive regulation of epithelial to mesenchymal transition
(https://www.ebi.ac.uk/QuickGO/term/GO:0010718)
GO:0060021 : roof of mouth development
(https://www.ebi.ac.uk/QuickGO/term/GO:0060021)
GO:0001701 : in utero embryonic development
(https://www.ebi.ac.uk/QuickGO/term/GO:0001701)
GO:0010628 : positive regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010628)
GO:0001503 : ossification (https://www.ebi.ac.uk/QuickGO/term/GO:0001503)
GO:0001649 : osteoblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001649)
GO:0050679 : positive regulation of epithelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0050679)
GO:0071456 : cellular response to hypoxia
(https://www.ebi.ac.uk/QuickGO/term/GO:0071456)
GO:0043433 : negative regulation of DNA-binding transcription factor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0043433)
GO:0060363 : cranial suture morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060363)
GO:0035116 : embryonic hindlimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035116)
GO:0048701 : embryonic cranial skeleton morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0048701)
GO:0042476 : odontogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0042476)
GO:0035137 : hindlimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035137)
GO:2000773 : negative regulation of cellular senescence
(https://www.ebi.ac.uk/QuickGO/term/GO:2000773)
GO:2000679 : positive regulation of transcription regulatory region DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:2000679)
GO:0032720 : negative regulation of tumor necrosis factor production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032720)
GO:2000144 : positive regulation of DNA-templated transcription, initiation
(https://www.ebi.ac.uk/QuickGO/term/GO:2000144)
GO:0007517 : muscle organ development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007517)
GO:0032000 : positive regulation of fatty acid beta-oxidation
(https://www.ebi.ac.uk/QuickGO/term/GO:0032000)
GO:0048511 : rhythmic process (https://www.ebi.ac.uk/QuickGO/term/GO:0048511)
GO:0035115 : embryonic forelimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035115)
GO:0032760 : positive regulation of tumor necrosis factor production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032760)
GO:0045668 : negative regulation of osteoblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045668)
GO:2000778 : positive regulation of interleukin-6 secretion
(https://www.ebi.ac.uk/QuickGO/term/GO:2000778)
GO:0042733 : embryonic digit morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0042733)
GO:0061029 : eyelid development in camera-type eye
(https://www.ebi.ac.uk/QuickGO/term/GO:0061029)

GO:0045843 : negative regulation of striated muscle tissue development
(https://www.ebi.ac.uk/QuickGO/term/GO:0045843)
GO:0001764 : neuron migration (https://www.ebi.ac.uk/QuickGO/term/GO:0001764)
GO:0003180 : aortic valve morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003180)
GO:0061309 : cardiac neural crest cell development involved in outflow tract morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0061309)
GO:0003253 : cardiac neural crest cell migration involved in outflow tract morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003253)
GO:2000793 : cell proliferation involved in heart valve development
(https://www.ebi.ac.uk/QuickGO/term/GO:2000793)
GO:0071363 : cellular response to growth factor stimulus
(https://www.ebi.ac.uk/QuickGO/term/GO:0071363)
GO:0060900 : embryonic camera-type eye formation
(https://www.ebi.ac.uk/QuickGO/term/GO:0060900)
GO:0003203 : endocardial cushion morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003203)
GO:0003183 : mitral valve morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0003183)
GO:0045596 : negative regulation of cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0045596)
GO:0043518 : negative regulation of DNA damage response, signal transduction by p53
class mediator (https://www.ebi.ac.uk/QuickGO/term/GO:0043518)
GO:2000780 : negative regulation of double-strand break repair
(https://www.ebi.ac.uk/QuickGO/term/GO:2000780)
GO:0035067 : negative regulation of histone acetylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0035067)
GO:0033128 : negative regulation of histone phosphorylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0033128)
GO:0044092 : negative regulation of molecular function
(https://www.ebi.ac.uk/QuickGO/term/GO:0044092)
GO:2000276 : negative regulation of oxidative phosphorylation uncoupler activity
(https://www.ebi.ac.uk/QuickGO/term/GO:2000276)
GO:0035359 : negative regulation of peroxisome proliferator activated receptor signaling
pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0035359)
GO:0014067 : negative regulation of phosphatidylinositol 3-kinase signaling
(https://www.ebi.ac.uk/QuickGO/term/GO:0014067)
GO:0048642 : negative regulation of skeletal muscle tissue development
(https://www.ebi.ac.uk/QuickGO/term/GO:0048642)
GO:2000147 : positive regulation of cell motility
(https://www.ebi.ac.uk/QuickGO/term/GO:2000147)
GO:2000802 : positive regulation of endocardial cushion to mesenchymal transition
involved in heart valve formation (https://www.ebi.ac.uk/QuickGO/term/GO:2000802)
GO:0071639 : positive regulation of monocyte chemotactic protein-1 production
(https://www.ebi.ac.uk/QuickGO/term/GO:0071639)
GO:0030500 : regulation of bone mineralization
(https://www.ebi.ac.uk/QuickGO/term/GO:0030500)

GO - Cellular Component

GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Insertion Size

10-99 bp

Molecular Details of the Mutation

small duplication (c.148_157dup (p.A56RfsX87)) that causes a frameshift and inactivates the gene.

Experimental Evidence

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

Main Reference

A newly described bovine type 2 scurs syndrome segregates with a frame-shift mutation in TWIST1. (2011) (https://pubmed.ncbi.nlm.nih.gov/21814570)

Authors

Capitan A; Grohs C; Weiss B; Rossignol MN; ReversÃ© P; Eggen A

Abstract

The developmental pathways involved in horn development are complex and still poorly understood. Here we report the description of a new dominant inherited syndrome in the bovine Charolais breed that we have named type 2 scurs. Clinical examination revealed that, despite a strong phenotypic variability, all affected individuals show both horn abnormalities similar to classical scurs phenotype and skull interfrontal suture synostosis. Based on a genome-wide linkage analysis using Illumina BovineSNP50 BeadChip genotyping data from 57 half-sib and full-sib progeny, this locus was mapped to a 1.7 Mb interval on bovine chromosome 4. Within this region, the TWIST1 gene encoding a transcription factor was considered as a strong candidate gene since its haploinsufficiency is responsible for the human Saethre-Chotzen syndrome, characterized by skull coronal suture synostosis. Sequencing of the TWIST1 gene identified a c.148_157dup (p.A56RfsX87) frame-shift mutation predicted to completely inactivate this gene. Genotyping 17 scurred and 20 horned founders of our pedigree as well as 48 unrelated horned controls revealed a perfect association between this mutation and the type 2 scurs phenotype. Subsequent genotyping of 32 individuals born from heterozygous parents showed that homozygous mutated progeny are completely absent, which is consistent with the embryonic lethality reported in Drosophila and mouse suffering from TWIST1 complete insufficiency. Finally, data from previous studies on model species and a fine description of type 2 scurs symptoms allowed us to propose different mechanisms to explain the features of this syndrome. In conclusion, this first report on the identification of a potential causal mutation affecting horn development in cattle offers a unique opportunity to better understand horn ontogenesis.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Homozygous lethal. Dominant allele. https://omia.org/OMIA001593/9913/