

## GEPHE SUMMARY

### Gephebase Gene

Twist2

### Entry Status

Published

### GepheID

GP00002035

### Main curator

Courtier

## PHENOTYPIC CHANGE

### Trait Category

Morphology

### Trait

Coloration (coat; white belt)

### Trait State in Taxon A

Bos taurus

### Trait State in Taxon B

Bos taurus - white belt

### Ancestral State

Taxon A

### Taxonomic Status

Domesticated

### Taxon A

#### Latin Name

*Bos taurus*

#### 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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

#### Parent

Bos (oxen, cattle) - (Rank: genus)

#### NCBI Taxonomy ID

9913

#### is Taxon A an Intraspecies?

No

### Taxon B

#### Latin Name

*Bos taurus*

#### 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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

#### Parent

Bos (oxen, cattle) - (Rank: genus)

#### NCBI Taxonomy ID

9913

#### is Taxon B an Intraspecies?

No

## GENOTYPIC CHANGE

### Generic Gene Name

Twist2

### Synonyms

Dermo1; bHLHa39

### String

10090.ENSMUSP00000139531

### Sequence Similarities

-

### GO - Molecular Function

GO:0046983 : protein dimerization activity

GO:0003700 : DNA-binding transcription factor activity

GO:0008134 : transcription factor binding

GO:0003677 : DNA binding

GO:0003682 : chromatin binding

GO:0019904 : protein domain specific binding

### GO - Biological Process

GO:0043066 : negative regulation of apoptotic process

GO:0006357 : regulation of transcription by RNA polymerase II

### UniProtKB Mus musculus

Q9D030

### GenebankID or UniProtKB

GO:0000122 : negative regulation of transcription by RNA polymerase II  
GO:0045892 : negative regulation of transcription, DNA-templated  
GO:0008285 : negative regulation of cell proliferation  
GO:0001649 : osteoblast differentiation  
GO:0048701 : embryonic cranial skeleton morphogenesis  
GO:0030335 : positive regulation of cell migration  
GO:0043392 : negative regulation of DNA binding  
GO:0032720 : negative regulation of tumor necrosis factor production  
GO:0045668 : negative regulation of osteoblast differentiation  
GO:0044092 : negative regulation of molecular function  
GO:0061303 : cornea development in camera-type eye  
GO:0060325 : face morphogenesis  
GO:0045638 : negative regulation of myeloid cell differentiation  
GO:0010838 : positive regulation of keratinocyte proliferation

#### GO - Cellular Component

GO:0005737 : cytoplasm  
GO:0005654 : nucleoplasm  
GO:0005634 : nucleus  
GO:0005667 : transcription factor complex  
GO:0005730 : nucleolus

#### Presumptive Null

No

#### Molecular Type

Cis-regulatory

#### Aberration Type

Insertion

#### Insertion Size

10-100 kb

#### Molecular Details of the Mutation

quadruplication of a 6 kb non-coding sequence located approximately 16 kb upstream of the TWIST2 gene

#### Experimental Evidence

Linkage Mapping

#### Main Reference

A structural variant in the 5'-flanking region of the TWIST2 gene affects melanocyte development in belted cattle. (2017)

#### Authors

Awasthi Mishra N; DrÄqgemÄ¼ller C; Jagannathan V; Keller I; WÄ¼thrich D; Bruggmann R; Beck J; SchÄ¼tz E; Brenig B; Demmel S; Moser S; Signer-Hasler H; PieÄkowska-Schelling A; Schelling C; Sande M; Rongen R; Rieder S; Kelsh RN; Mercader N; Leeb T

#### Abstract

Belted cattle have a circular belt of unpigmented hair and skin around their midsection. The belt is inherited as a monogenic autosomal dominant trait. We mapped the causative variant to a 37 kb segment on bovine chromosome 3. Whole genome sequence data of 2 belted and 130 control cattle yielded only one private genetic variant in the critical interval in the two belted animals. The belt-associated variant was a copy number variant (CNV) involving the quadruplication of a 6 kb non-coding sequence located approximately 16 kb upstream of the TWIST2 gene. Increased copy numbers at this CNV were strongly associated with the belt phenotype in a cohort of 333 cases and 1322 controls. We hypothesized that the CNV causes aberrant expression of TWIST2 during neural crest development, which might negatively affect melanoblasts. Functional studies showed that ectopic expression of bovine TWIST2 in neural crest in transgenic zebrafish led to a decrease in melanocyte numbers. Our results thus implicate an unsuspected involvement of TWIST2 in regulating pigmentation and reveal a non-coding CNV underlying a captivating Mendelian character.

#### Additional References

## RELATED GEPHE

#### Related Genes

10 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PMEL17, SLC45A2=MATP, tyrosinase-related protein 1 (TYRP1))

#### Related Haplotypes

No matches found.

## COMMENTS

<https://omia.org/OMIA001469/9913/>