

GEPHE SUMMARY

| | | | |
|--|----------------|------------|--------------|
| Twist2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+Twist2+Gephebase-summary-title) | Gephebase Gene | GP00002035 | GepheID |
| Published | Entry Status | Courtier | Main curator |

PHENOTYPIC CHANGE

| | | | |
|---|-----------------------------|---|-----------------------------|
| Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category+Morphology+Gephebase-summary-title) | Trait Category | | |
| Coloration (coat; white belt) (https://www.gephebase.org/search-criteria?/and+Trait+Coloration+coat+white+belt+Gephebase-summary-title) | Trait | | |
| Bos taurus | Trait State in Taxon A | | |
| Bos taurus - white belt | Trait State in Taxon B | | |
| Taxon A | Ancestral State | | |
| Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+Gephebase-summary-title) | Taxonomic Status | | |
| | Taxon A | | Taxon B |
| Bos taurus | Latin Name | Bos taurus | Latin Name |
| (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Bos+taurus+Gephebase-summary-title) | Common Name | (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 Taurus | Rank | Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus | 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) | NCBI Taxonomy ID | Bos (oxen, cattle) - (Rank: genus) | NCBI Taxonomy ID |
| (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903) | 9913 | (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903) | 9913 |
| | is Taxon A an Intraspecies? | | is Taxon B an Intraspecies? |
| No | | No | |

GENOTYPIC CHANGE

| | | | |
|---|-------------------------|--|-------------------------|
| Twist2 | Generic Gene Name | Q9D030 (http://www.uniprot.org/uniprot/Q9D030) | UniProtKB Mus musculus |
| Dermo1; bHLHa39 | Synonyms | Q | GenebankID or UniProtKB |
| 10090.ENSMUSP00000139531 | String | | |
| (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000139531) | Sequence Similarities | | |
| - | GO - Molecular Function | | |
| GO:0046983 : protein dimerization activity | | | |
| (https://www.ebi.ac.uk/QuickGO/term/GO:0046983) | | | |
| GO:0003700 : DNA-binding transcription factor activity | | | |
| (https://www.ebi.ac.uk/QuickGO/term/GO:0003700) | | | |
| GO:0008134 : transcription factor binding | | | |
| (https://www.ebi.ac.uk/QuickGO/term/GO:0008134) | | | |

GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
GO:0003682 : chromatin binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003682>)
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:0006357 : regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006357>)
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:0008285 : negative regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008285>)
GO:0001649 : osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001649>)
GO:0048701 : embryonic cranial skeleton morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048701>)
GO:0030335 : positive regulation of cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030335>)
GO:0043392 : negative regulation of DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043392>)
GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
GO:0045668 : negative regulation of osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045668>)
GO:0044092 : negative regulation of molecular function
(<https://www.ebi.ac.uk/QuickGO/term/GO:0044092>)
GO:0061303 : cornea development in camera-type eye
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061303>)
GO:0060325 : face morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060325>)
GO:0045638 : negative regulation of myeloid cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045638>)
GO:0010838 : positive regulation of keratinocyte proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010838>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0005667 : transcription factor complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005667>)
GO:0005730 : nucleolus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005730>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

10-100 kb

Molecular Details of the Mutation

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

Experimental Evidence

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

Main Reference

A structural variant in the 5'-flanking region of the TWIST2 gene affects melanocyte development in belted cattle. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28658273>)

Authors

Awasthi Mishra N; DrÄ¼gemÄ¼ller C; Jagannathan V; Keller I; WÄ¼thrich D; Bruggmann R; Beck J; SchÄ¼tz E; Brenig B; Demmel S; Moser S; Signer-Hasler H; PieÄ¼kowska-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

Remapping of the belted phenotype in cattle on BTA3 identifies a multiplication event as the candidate causal mutation. (2018) (<https://pubmed.ncbi.nlm.nih.gov/29980171>)

A shared 336 kb haplotype associated with the belt pattern in three divergent cattle breeds. (2010) (<https://pubmed.ncbi.nlm.nih.gov/19917047>)

RELATED GEPHE

Related Genes

11 (Agouti, coatmer 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 (TYR), tyrosinase-related protein 1 (TYRP1)) (<https://www.gephebase.org/search-criteria?/or+TaxonID=^9913^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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