GEPHE SUMMARY

Gephebase Gene

PMEL17 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^PMEL17^#gephebase-summary-title)

GepheID

GP00002028

Entry Status

Published

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Domesticated cattle

Trait State in Taxon B

Domesticated cattle with coat colour dilution

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?

Yes

Taxon A Description

Holstein

Taxon B #1

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?

Yes

Taxon B Description

Hereford

Taxon B #2

Latin Name

Bos grunniens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos grunniens^#gephebase-summary-title)

Common Name

domestic yak

Synonyms

Bos mutus grunniens; Poephagus grunniens; domestic yak; yak

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)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903)

NCBI Taxonomy ID

30521
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30521)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

PMEL

Synonyms

SILV; PMEL17; RPE1

String

9913.ENSBTAP00000005250
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=
9913.ENSBTAP00000005250)

Sequence Similarities

Belongs to the PMEL/NMB family.

GO - Molecular Function

-

GO - Biological Process

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0032438 : melanosome organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0032438)

GO - Cellular Component

GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0005771 : multivesicular body (https://www.ebi.ac.uk/QuickGO/term/GO:0005771)

UniProtKB Bos taurus

Q06154 (http://www.uniprot.org/uniprot/Q06154)

GenebankID or UniProtKB

()

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

3bp deletion - g.57669913_57669915delTTC - c.50_52delTTC - p.Leu19del

Experimental Evidence

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

Main Reference

Coat-colour dilution and hypotrichosis in Hereford crossbred calves. (2008) (https://pubmed.ncbi.nlm.nih.gov/18408794)

Authors

Jolly RD; Wills JL; Kenny JE; Cahill JI; Howe L

Abstract

To investigate cases of coat-colour dilution and hypotrichosis in a group of Hereford x Friesian crossbred calves, and to define the underlying molecular genetics of the disorder.

The investigation was predicated on the hypothesis that this disorder was similar to a known dominantly inherited disorder of calves of black breeds crossed with Simmental cattle, for which there were candidate gene mutations. Sequence analyses of PCR amplicons from exon 1 and exon 11 of the premelanosome protein 17 gene (PMel17) were carried out. Restriction enzyme digestions of amplicons were followed using electrophoresis of digested fragments.

It was shown that an affected calf and its Hereford sire were heterozygous for a three-base deletion in exon 1 of the PMel17 gene. These two animals were also heterozygous for a second mutation in exon 11 of the PMel17 gene. Four other related animals were likewise heterozygous for both mutations in the sire's herd of origin.

Coat-colour dilution and hypotrichosis in Hereford crossbred calves in New Zealand is the same genetic disorder as that previously described in Simmental crossbred calves, and is linked to mutations in the PMel17 gene.

Additional References

RELATED GEPHE

Related Genes

11 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9913^/and+Trait=Coloration/or+Taxon ID=^30521^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9913^/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^9913^/or+Gene Gephebase=^PMEL17^/and+Taxon ID=^30521^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries @Parallelism https://omia.org/OMIA001545/9913/