

## GEPHE SUMMARY

**Gephebase Gene**  
PMEL17

**Entry Status**  
Published

**GepheID**  
GP00002028

**Main curator**  
Courtier

## PHENOTYPIC CHANGE

**Trait Category**  
Morphology

**Trait**  
Coloration (coat)

**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

### 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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

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

**NCBI Taxonomy ID**  
9913

**is Taxon A an Intraspecies?**  
Yes

**Taxon A Description**  
Holstein

### Taxon B #1

**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; Artiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

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

**NCBI Taxonomy ID**  
9913

**is Taxon B an Intraspecies?**  
Yes

**Taxon B Description**  
Hereford

### Taxon B #2

**Latin Name**  
*Bos grunniens*

**Common Name**  
domestic yak

**Synonyms**  
Bos mutus grunniens; Poepagus 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)

**NCBI Taxonomy ID**  
30521

is Taxon B an Intraspecies?

No

## GENOTYPIC CHANGE

### Generic Gene Name

PMEL

UniProtKB *Bos taurus*

[Q06154](#)

### Synonyms

SILV; PMEL17; RPE1

GenebankID or UniProtKB

### String

[9913.ENSBTAP00000005250](#)

### Sequence Similarities

Belongs to the PMEL/NMB family.

### GO - Molecular Function

-

### GO - Biological Process

GO:0042438 : melanin biosynthetic process

GO:0032438 : melanosome organization

### GO - Cellular Component

GO:0005887 : integral component of plasma membrane

GO:0005576 : extracellular region

GO:0005794 : Golgi apparatus

GO:0005789 : endoplasmic reticulum membrane

GO:0042470 : melanosome

GO:0005771 : multivesicular body

### Presumptive Null

No

### Molecular Type

Coding

### Aberration Type

Deletion

### Deletion Size

1-9 bp

### Molecular Details of the Mutation

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

### Experimental Evidence

Candidate Gene

### Main Reference

[Coat-colour dilution and hypotrichosis in Hereford crossbred calves. \(2008\)](#)

### 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

10 (Agouti, coatomer protein complex subunit alpha (COPA), Kit (type III receptor protein-tyrosine kinase), Kit ligand, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, SLC45A2=MATP, Twist2, tyrosinase-related protein 1 (TYRP1))

### Related Haplotypes

1

## EXTERNAL LINKS

## COMMENTS

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