

# GEPHE SUMMARY

	Gephebase Gene	GephelD
PMEL17 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase=^PMEL17">#gephebase-summary-title)	GP00002028	
PUBLICATION	Entry Status	Main curator
Published	Courier	

## PHENOTYPIC CHANGE

	Trait Category	
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category=^Morphology">#gephebase-summary-title)	Trait	
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration</a> (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 ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status=^Domesticated">#gephebase-summary-title)		
Taxon A	Latin Name	Latin Name
Bos taurus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos</a> taurus)#gephebase-summary-title)		
cattle	Common Name	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus	Synonyms	Synonyms
species	Rank	Rank
	Lineage	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	Parent
Bos (oxen, cattle) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a> )	NCBI Taxonomy ID	NCBI Taxonomy ID
9913 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913</a> )		
Yes	Taxon A Description	Taxon B Description
Holstein		
Taxon B #1	Latin Name	Latin Name
Bos taurus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos</a> taurus)#gephebase-summary-title)		
cattle	Common Name	Common Name
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Tauurus	Synonyms	Synonyms
species	Rank	Rank
	Lineage	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	Parent
Bos (oxen, cattle) - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903</a> )	NCBI Taxonomy ID	NCBI Taxonomy ID
9913 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913</a> )		
Yes	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
Hereford		
Taxon B #2	Latin Name	Latin Name
Bos grunniens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos</a> grunniens)#gephebase-summary-title)		
domestic yak	Common Name	Common Name
Bos mutus grunniens; Poephagus grunniens; domestic yak; yak	Synonyms	Synonyms
species	Rank	Rank
	Lineage	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	Parent

	Taxon B #2	
Bos grunniens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Bos</a> grunniens)#gephebase-summary-title)	Latin Name	Latin Name
domestic yak	Common Name	Common Name
Bos mutus grunniens; Poephagus grunniens; domestic yak; yak	Synonyms	Synonyms
species	Rank	Rank
	Lineage	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	Parent

Bos (oxen, cattle) - (Rank: genus)	Related Genes
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9903</a> )	
30521	NCBI Taxonomy ID
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30521">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 30521</a> )	is Taxon B an Infraspecies?

No

## GENOTYPIC CHANGE

PMEL	Generic Gene Name	UniProtKB Bos taurus
SILV; PMEL17; RPE1	Synonyms	GenebankID or UniProtKB
9913.ENSBTAP00000005250 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9913.ENSBTAP00000005250">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9913.ENSBTAP00000005250</a> )	String	0
Belongs to the PMEL/NMB family.	Sequence Similarities	
-	GO - Molecular Function	
GO:0042438 : melanin biosynthetic process ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042438">https://www.ebi.ac.uk/QuickGO/term/GO:0042438</a> )	GO - Biological Process	
GO:0032438 : melanosome organization ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0032438">https://www.ebi.ac.uk/QuickGO/term/GO:0032438</a> )	GO - Cellular Component	
GO:0005887 : integral component of plasma membrane ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005887">https://www.ebi.ac.uk/QuickGO/term/GO:0005887</a> )		Presumptive Null
GO:0005576 : extracellular region ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005576">https://www.ebi.ac.uk/QuickGO/term/GO:0005576</a> )		Molecular Type
GO:0005794 : Golgi apparatus ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005794">https://www.ebi.ac.uk/QuickGO/term/GO:0005794</a> )		Aberration Type
GO:0005789 : endoplasmic reticulum membrane ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005789">https://www.ebi.ac.uk/QuickGO/term/GO:0005789</a> )		Deletion Size
GO:0042470 : melanosome ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042470">https://www.ebi.ac.uk/QuickGO/term/GO:0042470</a> )		
GO:0005771 : multivesicular body ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005771">https://www.ebi.ac.uk/QuickGO/term/GO:0005771</a> )		
No ( <a href="https://www.gephebase.org/search-criteria/?and+Presumptive Null^No">#gephebase-summary-title</a> )		
Coding ( <a href="https://www.gephebase.org/search-criteria/?and+Molecular Type^Coding">#gephebase-summary-title</a> )		
Deletion ( <a href="https://www.gephebase.org/search-criteria/?and+Aberration Type^Deletion">#gephebase-summary-title</a> )		
1-9 bp	Molecular Details of the Mutation	
3bp deletion - g.57669913_57669915delTTC - c.50_52delTTC - p.Leu19del	Experimental Evidence	
Candidate Gene ( <a href="https://www.gephebase.org/search-criteria/?and+Experimental Evidence^Candidate Gene">#gephebase-summary-title</a> )	Main Reference	
Coat-colour dilution and hypotrichosis in Hereford crossbred calves. (2008) ( <a href="https://pubmed.ncbi.nlm.nih.gov/18408794">https://pubmed.ncbi.nlm.nih.gov/18408794</a> )	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), Microphthalmia-associated transcription factor, SLC45A2=MATP, Twist2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) ( <a href="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">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</a> )

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/>