

GEPHE SUMMARY

	Gephebase Gene		GepheID
tyrosinase-related protein 1 (TYRP1) (https://www.gephebase.org/search-criteria?/and+Gene=Gephebase=^tyrosinase-related protein 1 (TYRP1)^#gephebase-summary-title)		GP00001145	
	Entry Status	Martin	Main curator
Published			

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		
Bos bovis		Trait State in Taxon B	
Bos bovis - Dexter cattle			
	Ancestral State		
Data not curated		Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)		Bos taurus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Bos taurus^#gephebase-summary-title)	
	Common Name		Common Name
cattle		cattle	
	Synonyms		Synonyms
Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus		Bos bovis; Bos primigenius taurus; cattle; bovine; cow; dairy cow; domestic cattle; domestic cow; Bos taurus Linnaeus, 1758; Bos Taurus	
	Rank		Rank
species		species	
	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		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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)		Bos (oxen, cattle) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9903)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)		9913 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9913)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Tyrp1		P07147 (http://www.uniprot.org/uniprot/P07147)	
	Synonyms		GenebankID or UniProtKB
b; isa; Oca3; TRP1; Tyrp; TRP-1; brown; Tyrp-2		AAL38167 (https://www.ncbi.nlm.nih.gov/nuccore/AAL38167)	
	String		
10090.ENSMUSP00000006151 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000006151)			
	Sequence Similarities		
Belongs to the tyrosinase family.			
	GO - Molecular Function		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)			
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)			
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)			

GO:0004497 : monooxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004497>)

GO - Biological Process

GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0006583 : melanin biosynthetic process from tyrosine
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006583>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0043438 : acetoacetic acid metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043438>)
GO:0006582 : melanin metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006582>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)
GO:0033162 : melanosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.1300C>T p.H434Y

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	His	Tyr	434

Main Reference

TYRP1 is associated with dun coat colour in Dexter cattle or how now brown cow?. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12755816>)

Authors

Berryere TG; Schmutz SM; Schimpf RJ; Cowan CM; Potter J

Abstract

Tyrosinase related protein 1 (TYRP1), which is involved in the coat colour pathway, was mapped to BTA8 between microsatellites BL1080 and BM4006, using a microsatellite in intron 5 of TYRP1. The complete coding sequence of bovine TYRP1 was determined from cDNA derived from skin biopsies of cattle with various colours. Sequence data from exons 2-8 from cattle with diluted phenotypes was compared with that from non-diluted phenotypes. In addition, full-sib families of beef cattle generated by embryo transfer and half-sib families from traditional matings in which coat colour was segregating were used to correlate TYRP1 sequence variants with dilute coat colours. Two non-conservative amino acid changes were detected in Simmental, Charolais and Galloway cattle but these polymorphisms were not associated with diluted shades of black or red, nor with the dun coat colour of Galloway cattle or the taupe brown colour of Braunvieh and Brown Swiss cattle. However, in Dexter cattle all 25 cattle with a dun brown coat colour were homozygous for a H424Y change. One Dexter that was also homozygous Y434 was red because of an "E+/E+" genotype at MC1R which lead to the production of only phaeomelanin. None of the 70 remaining black or red Dexter cattle were homozygous for Y434. This tyrosine mutation was not found in any of the 121 cattle of other breeds that were examined.

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, PMEL17, SLC45A2=MATP, Twist2, tyrosinase (TYR)) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~9913~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001249/9913/>