

GEPHE SUMMARY

tyrosinase (TYR) (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^tyrosinase+(TYR)^#gephebase-summary-title)	Gephebase Gene	GP00002315	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

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

GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 (http://www.uniprot.org/uniprot/P11344)	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP0000004770 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP0000004770)	String		
Belongs to the tyrosinase family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)			
GO:0005507 : copper ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005507)			
GO:0004503 : monophenol monooxygenase activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0004503>)

GO - Biological Process

GO:0042438 : melanin biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)

GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)

GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)

GO:0033280 : response to vitamin D (<https://www.ebi.ac.uk/QuickGO/term/GO:0033280>)

GO:0051591 : response to cAMP (<https://www.ebi.ac.uk/QuickGO/term/GO:0051591>)

GO:0009411 : response to UV (<https://www.ebi.ac.uk/QuickGO/term/GO:0009411>)

GO:0048538 : thymus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048538>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

GO:0043231 : intracellular membrane-bounded organelle

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)

GO:0048471 : perinuclear region of cytoplasm

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)

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

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title))

Molecular Type

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

Aberration Type

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

c.925_926insC

Experimental Evidence

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

Main Reference

A form of albinism in cattle is caused by a tyrosinase frameshift mutation. (2004) (<https://pubmed.ncbi.nlm.nih.gov/14727143>)

Authors

Schmutz SM; Berryere TG; Ciobanu DC; Mileham AJ; Schmidtz BH; Fredholm M

Abstract

We used PCR amplification of cDNA prepared from skin biopsies to determine the full-length protein-coding sequence of tyrosinase (TYR) in cattle of several coat colors. An insertion of a cytosine was detected in an albino Braunvieh calf, which resulted in a frameshift which caused a premature stop codon at residue 316. This insertion was found in the homozygous state in this calf and the genomic DNA of two related albino calves. All six parents of these calves were heterozygous for this insertion. However, an albino Holstein calf did not have this insertion, nor was any other mutation detected in the partial TYR sequence obtained from the genomic DNA available. Diagnostic genotyping tests were developed to detect this mutation in Braunvieh cattle.

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-related protein 1 (TYRP1)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9913^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title](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

@Parallelism <https://omia.org/OMIA000202/9913/>

