

GEPHE SUMMARY

Gephebase Gene

BCO2 = beta-carotene oxygenase 2

Entry Status

Published

GepheID

GP00000138

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Physiology

Trait

Carotenoid content

Trait State in Taxon A

Bos bovis

Trait State in Taxon B

Bos bovis; yellow serum breeds

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent

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

NCBI Taxonomy ID

9913

is Taxon A an Intraspecies?

No

Taxon B

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; Cetartiodactyla; Ruminantia; Pecora; Bovidae; Bovinae; Bos

Parent

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

NCBI Taxonomy ID

9913

is Taxon B an Intraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

BCO2

Synonyms

BCDO2; B-DIOX-II

String

9606.ENSP00000350314

Sequence Similarities

Belongs to the carotenoid oxygenase family.

GO - Molecular Function

GO:0046872 : metal ion binding

GO:0003834 : beta-carotene 15,15'-monooxygenase activity

GO:0010436 : carotenoid dioxygenase activity

GO:0004744 : retinal isomerase activity

GO:0102076 : beta,beta-carotene-9',10'-cleaving oxygenase activity

GO:0016702 : oxidoreductase activity, acting on single donors with incorporation of molecular oxygen, incorporation of two atoms of oxygen

GO - Biological Process

GO:0055114 : oxidation-reduction process

UniProtKB Homo sapiens

Q9BYV7

GenebankID or UniProtKB

AAI51704

GO:0001523 : retinoid metabolic process
GO:0016121 : carotene catabolic process
GO:0042574 : retinal metabolic process
GO:0016119 : carotene metabolic process
GO:0016116 : carotenoid metabolic process
GO:0051881 : regulation of mitochondrial membrane potential
GO:2000377 : regulation of reactive oxygen species metabolic process
GO:0042573 : retinoic acid metabolic process
GO:0016122 : xanthophyll metabolic process

GO - Cellular Component

GO:0005739 : mitochondrion
GO:0005622 : intracellular
GO:0005759 : mitochondrial matrix

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsense

Molecular Details of the Mutation

Substitution creating premature Stop (codon 79)

Experimental Evidence

Linkage Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

[Mutation in bovine beta-carotene oxygenase 2 affects milk color. \(2009\)](#)

Authors

Berry SD; Davis SR; Beattie EM; Thomas NL; Burrett AK; Ward HE; Stanfield AM; Biswas M; Ankersmit-Udy AE; Oxley PE; Barnett JL; Pearson JF; van der Does Y; Macgibbon AH; Spelman RJ; Lehnert K; Snell RG

Abstract

beta-Carotene biochemistry is a fundamental process in mammalian biology. Aberrations either through malnutrition or potentially through genetic variation may lead to vitamin A deficiency, which is a substantial public health burden. In addition, understanding the genetic regulation of this process may enable bovine improvement. While many bovine QTL have been reported, few of the causative genes and mutations have been identified. We discovered a QTL for milk beta-carotene and subsequently identified a premature stop codon in bovine beta-carotene oxygenase 2 (BCO2), which also affects serum beta-carotene content. The BCO2 enzyme is thereby identified as a key regulator of beta-carotene metabolism.

Additional References

[Genetic variation in the beta, beta-carotene-9', 10'-dioxygenase gene and association with fat colour in bovine adipose tissue and milk. \(2010\)](#)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

COMMENTS

<https://omia.org/OMIA001079/9913/>