GEPHE SUMMARY

Gephebase Gene

BCO2 = beta-carotene oxygenase 2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^BCO2 = beta-carotene oxygenase 2^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000140

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait

Carotenoid content (yellow fat) (https://www.gephebase.org/search-criteria?/and+Trait=^Carotenoid content (yellow fat)^#gephebase-summary-title)

Trait State in Taxon A

White fat breeds

Trait State in Taxon B

yellow fat breeds

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name

Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name

sheep

Synonyms

Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Caprinae; Ovis

Parent

Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID

9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Ovis aries (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Ovis aries^#gephebase-summary-title)

Common Name

sheep

Synonyms

Ovis ammon aries; Ovis orientalis aries; Ovis ovis; sheep; domestic sheep; lambs; wild sheep; Ovis aries Linnaeus, 1758

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; Caprinae; Ovis

Parent

Ovis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935)

NCBI Taxonomy ID

9940 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Norwegian White Sheep

GENOTYPIC CHANGE

Generic Gene Name

BCO2

Synonyms

BCDO2; B-DIOX-II

String

9606.ENSP00000350314 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000350314)

Sequence Similarities

Belongs to the carotenoid oxygenase family.

GO - Molecular Function

GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0003834 : beta-carotene 15,15′-monooxygenase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003834)
GO:0010436 : carotenoid dioxygenase activity

UniProtKB Homo sapiens

Q9BYV7 (http://www.uniprot.org/uniprot/Q9BYV7)

GenebankID or UniProtKB

CAX63048 (https://www.ncbi.nlm.nih.gov/nuccore/CAX63048)

(https://www.ebi.ac.uk/QuickGO/term/GO:0010436)
GO:0004744 : retinal isomerase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004744)
GO:0102076 : beta,beta-carotene-9′,10′-cleaving oxygenase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0102076)
GO:0016702 : oxidoreductase activity, acting on single donors with incorporation of molecular oxygen, incorporation of two atoms of oxygen
(https://www.ebi.ac.uk/QuickGO/term/GO:0016702)

GO - Biological Process

GO:0055114 : oxidation-reduction process
(https://www.ebi.ac.uk/QuickGO/term/GO:0055114)
GO:0001523 : retinoid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0001523)
GO:0016121 : carotene catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0016121)
GO:0042574 : retinal metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042574)
GO:0016119 : carotene metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0016119)
GO:0016116 : carotenoid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0016116)
GO:0051881 : regulation of mitochondrial membrane potential
(https://www.ebi.ac.uk/QuickGO/term/GO:0051881)
GO:2000377 : regulation of reactive oxygen species metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:2000377)
GO:0042573 : retinoic acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042573)
GO:0016122 : xanthophyll metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0016122)

GO - Cellular Component

GO:0005739 : mitochondrion (https://www.ebi.ac.uk/QuickGO/term/GO:0005739)
GO:0005622 : intracellular (https://www.ebi.ac.uk/QuickGO/term/GO:0005622)
GO:0005759 : mitochondrial matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0005759)

Presumptive Null

Yes (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)

Aberration Type

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

g.21947481C>T ; c.196C>T ; p.Q66*

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	CAG	TAG	195
Amino-acid	Gln	STP	65

Main Reference

A nonsense mutation in the beta-carotene oxygenase 2 (BCO2) gene is tightly associated with accumulation of carotenoids in adipose tissue in sheep (Ovis aries). (2010) (https://pubmed.ncbi.nlm.nih.gov/20122251)

Authors

VÃ¥ge DI; Boman IA

Abstract

Sheep carcasses with yellow fat are sporadically observed at Norwegian slaughter houses. This phenomenon is known to be inherited as a recessive trait, and is caused by accumulation of carotenoids in adipose tissue. Two enzymes are known to be important in carotenoid degradation in mammals, and are therefore potential candidate genes for this trait. These are beta-carotene 15,15′-monooxygenase 1 (BCMO1) and the beta-carotene oxygenase 2 (BCO2).

In the present study the coding region of the BCMO1 and the BCO2 gene were sequenced in yellow fat individuals and compared to the corresponding sequences from control animals with white fat. In the yellow fat individuals a nonsense mutation was found in BCO2 nucleotide position 196 (c.196C>T), introducing a stop codon in amino acid position 66. The full length protein consists of 575 amino acids. In spite of a very low frequency of this mutation in the Norwegian Al-ram population, 16 out of 18 yellow fat lambs were found to be homozygous for this mutation.

In the present study a nonsense mutation (c.196C>T) in the beta-carotene oxygenase 2 (BCO2) gene is found to strongly associate with the yellow fat phenotype in sheep. The existence of individuals lacking this mutation, but still demonstrating yellow fat, suggests that additional mutations may cause a similar phenotype in this population. The results demonstrate a quantitatively important role for BCO2 in carotenoid degradation, which might indicate a broad enzyme specificity for carotenoids. Animals homozygous for the mutation are not reported to suffer from any negative health or development traits, pointing towards a minor role of BCO2 in vitamin A formation. Genotyping Al rams for c.196C>T can now be actively used in selection against the yellow fat trait.

Additional References

Biallelic Î²-carotene oxygenase 2 knockout results in yellow fat in sheep via CRISPR/Cas9. (2017) (https://pubmed.ncbi.nlm.nih.gov/27862083)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA001079/9940/