

## GEPHE SUMMARY

	Gephebase Gene	GephelD
BCO2 = beta-carotene oxygenase 2 ( <a href="https://www.gephebase.org/search-criteria/?and+Gene">https://www.gephebase.org/search-criteria/?and+Gene</a>	GP00000140	
Gephebase=^BCO2 = beta-carotene oxygenase 2 "#gephebase-summary-title)		Main curator
	Entry Status	Martin
Published		

## PHENOTYPIC CHANGE

	Trait Category	
Physiology ( <a href="https://www.gephebase.org/search-criteria/?and+Trait">https://www.gephebase.org/search-criteria/?and+Trait</a>		
Category="Physiology">#gephebase-summary-title)		
	Trait	
Carotenoid content (yellow fat) ( <a href="https://www.gephebase.org/search-criteria/?and+Trait=^Carotenoid+content+(yellow+fat)+#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Trait=^Carotenoid+content+(yellow+fat)+#gephebase-summary-title</a> )		
	Trait State in Taxon A	
White fat breeds		
	Trait State in Taxon B	
yellow fat breeds		
	Ancestral State	
Taxon A		
	Taxonomic Status	
Domesticated ( <a href="https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=^Domesticated">#gephebase-summary-title"&gt;https://www.gephebase.org/search-criteria/?and+Taxonomic+Status=^Domesticated"&gt;#gephebase-summary-title</a> )		
Taxon A		Taxon B
	Latin Name	Latin Name
Ovis aries		
( <a href="https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Ovis+aries">#gephebase-summary-title"&gt;https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Ovis+aries"&gt;#gephebase-summary-title</a> )		
	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)		
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9935</a> )		
	NCBI Taxonomy ID	
9940		
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9940</a> )		
	is Taxon A an Infraspecies?	
No		
		is Taxon B an Infraspecies?
		Yes
		Taxon B Description
		Norwegian White Sheep

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
BCO2		
	Synonyms	GenebankID or UniProtKB
BCDO2; B-DIOX-II		
	String	
9606.ENSP00000350314		
( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000350314">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000350314</a> )		
	Sequence Similarities	
Belongs to the carotenoid oxygenase family.		
	GO - Molecular Function	
GO:0046872 : metal ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a> )		
GO:0003834 : beta-carotene 15,15'-monoxygenase activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003834">https://www.ebi.ac.uk/QuickGO/term/GO:0003834</a> )		
GO:0010436 : carotenoid dioxygenase activity		

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsense

Molecular Details of the Mutation

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

Experimental Evidence

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

		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

VÅge 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'-monoxygenase 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 AI-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 AI rams for c.196C>T can now be actively used in selection against the yellow fat trait.

Additional References

Biallelic  $\beta^2$ -carotene oxygenase 2 knockout results in yellow fat in sheep via CRISPR/Cas9. (2017) (<https://pubmed.ncbi.nlm.nih.gov/27862083>)

## RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

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