

GEPHE SUMMARY

	Gephebase Gene		GepheID
BCO2 = beta-carotene oxygenase 2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+BCO2+beta-carotene+oxygenase+2+gephebase-summary-title)		GP00002152	
	Entry Status	Martin	Main curator
Published			

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		
New Zealand Red (NZR) with white fat			
	Trait State in Taxon B		
New Zealand Red (NZR) with yellow fat (autosomal recessive)			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Domesticated+gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Oryctolagus+cuniculus+gephebase-summary-title)		Oryctolagus cuniculus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Oryctolagus+cuniculus+gephebase-summary-title)	
	Common Name		Common Name
rabbit		rabbit	
	Synonyms		Synonyms
Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits		Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits	
	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; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus	
	Parent		Parent
Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)		Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9984)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)		9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9986)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
BCO2		Q9BYV7 (http://www.uniprot.org/uniprot/Q9BYV7)	
	Synonyms		GenebankID or UniProtKB
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 (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>)

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))

Presumptive Null

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))

Molecular Type

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

Aberration Type

1-9 bp

Deletion Size

AAT-deletion mutation at Asp codon 248 of the BCO2 gene located at the beginning of exon 6 which results in the removal of an Asp

Molecular Details of the Mutation

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))

Experimental Evidence

A novel AAT-deletion mutation in the coding sequence of the BCO2 gene in yellow-fat rabbits. (2015) (<https://pubmed.ncbi.nlm.nih.gov/26002694>)

Main Reference

Strychalski J; Brym P; Czarnik U; GugoÅ,ek A

Authors

The carcasses of yellow-fat rabbits may be attractive to modern consumers, because they have a relatively high content of biologically active compounds. One of the main candidate genes associated with the yellow-fat trait is β -carotene 9',10'-oxygenase (BCO2). This study is the first report of the novel AAT-deletion mutation at codon 248 of the BCO2 gene, which has been found in homozygous yellow-fat rabbits. The deletion mutation, located at the beginning of exon 6, results in the absence of asparagine in protein. We also developed a PCR-RFLP test that supports intravital genotyping of indel polymorphism based on genomic DNA.

Abstract

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

<https://omia.org/OMIA001079/9986/>

