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)

GepheID

GP00002152

Entry Status

Published

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

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

Latin Name

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

Common Name

rabbit

Synonyms

Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

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; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus

Parent

Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID

9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

rabbit

Synonyms

Lepus cuniculus; rabbit; European rabbit; Japanese white rabbit; domestic rabbit; rabbits

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; Euarchontoglires; Glires; Lagomorpha; Leporidae; Oryctolagus

Parent

Oryctolagus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9984)

NCBI Taxonomy ID

9986 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9986)

is Taxon B an Infraspecies?

No

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0010436)
GO:0004744 : retinal isomerase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004744)

UniProtKB Homo sapiens

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

GenebankID or UniProtKB

()

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

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

Experimental Evidence

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

Main Reference

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)

Authors

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

Abstract

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 Î²-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.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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