GEPHE SUMMARY

Gephebase Gene
BCO2 = beta-carotene oxygenase 2

GephelD
GP00002039

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Carotenoid content (bill, legs)

Trait State in Taxon A
wild and domesticated canari

Trait State in Taxon B
canari - urucum breed - bright red bills and legs

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Serinus canaria

Common Name
common canary

Synonyms
Serinus canarius; common canary; canary; Serinus canaria (Linnaeus, 1758)

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Passeriformes; Passeroidea; Fringillidae; Carduelinae; Serinus

Parent
Serinus () - (Rank: genus)

NCBI Taxonomy ID
9135

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Serinus canaria

Common Name
common canary

Synonyms
Serinus canarius; common canary; canary; Serinus canaria (Linnaeus, 1758)

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Passeriformes; Passeroidea; Fringillidae; Carduelinae; Serinus

Parent
Serinus () - (Rank: genus)

NCBI Taxonomy ID
9135

is Taxon B an Infraspecies?
Yes

Taxon B Description
urucum canari breed

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

UniProtKB Homo sapiens
Q9BYV7

GenebankID or UniProtKB
AAI51704

GO - Biological Process
GO:0055114 : oxidation-reduction process
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
Nonsynonymous

Molecular Details of the Mutation
point mutation at nucleotide position 837,806 that is predicted to be a nonsynonymous mutation in the exon 9 of the BCO2 gene. This variant results in the substitution of a histidine for an arginine at residue 413 of the protein (R413H) . In silico analysis suggests that the R413H could cause structural distortion of the protein and lead to a possible loss of activity. In vitro assays with the urucum BCO2 variant show that the enzyme activity is lost.

Experimental Evidence
Association Mapping

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	His	Arg	413

Main Reference
Genetic Basis Of De Novo Appearance Of Carotenoid Ornamentation In Bare-Parts Of Canaries . (2019/01/01)

Authors
Gazda Malgorzata A.; Toomey Matthew B.; Araujo Pedro M.; Lopes Ricardo J.; Afonso Sandra; Myers Connie A.; Serres Kyla; Kiser Phylip D.; Hill Geoffrey; Corbo Joseph C.; Carneiro Miguel

Abstract
-

Additional References

RELATED GEPHE

Related Genes
1 (SCARB1)
Related Haplotypes
No matches found.

COMMENTS

Paper is on BioRxiv - https://www.biorxiv.org/content/10.1101/762112v1 . Allele-specific expression in the shared cellular environment of individuals heterozygous for the urucum and wild-type alleles show same expression levels of both alleles, suggesting that there is no cis-regulatory mutations in the urucum breed.