GEPHE SUMMARY

Gephebase Gene
SCARB1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SCARB1^#gephebase-summary-title)
Entry Status
Published

GepheID
GP00001681
Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)
Trait
Carotenoid content (https://www.gephebase.org/search-criteria?/and+Trait=^Carotenoid content^#gephebase-summary-title)
Trait State in Taxon A
yellow canari
Trait State in Taxon B
white canari
Ancestral State
Taxon A
Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Serinus canaria (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Serinus canaria^#gephebase-summary-title)
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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9134)
NCBI Taxonomy ID
9135 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9135)
is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Serinus canaria (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Serinus canaria^#gephebase-summary-title)
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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9134)
NCBI Taxonomy ID
9135 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9135)
is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Scarb1
Synonyms
CD36; Cla1; SRBI; Srb1; Cla-1; Hdlq1; SR-B1; SR-BI; Cd36l1; Chohd1; Hlb398; mSR-BI; AI120173; D5Ertd460e
String
10090.ENSMUSP00000083242 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 10090.ENSMUSP00000083242)
Sequence Similarities
Belongs to the CD36 family.
GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0001540 : amyloid-beta binding (https://www.ebi.ac.uk/QuickGO/term/GO:0001540)
GO:0001530 : lipopolysaccharide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0001530)

UniProtKB Mus musculus
Q61009 (http://www.uniprot.org/uniprot/Q61009)
GenebankID or UniProtKB
()

GO:0001875 : lipopolysaccharide receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0001875)
GO:0001786 : phosphatidylserine binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0001786)
GO:0034186 : apolipoprotein A-I binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0034186)
GO:0034185 : apolipoprotein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0034185)
GO:0008035 : high-density lipoprotein particle binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0008035)
GO:0070506 : high-density lipoprotein particle receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0070506)
GO:0030169 : low-density lipoprotein particle binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0030169)

GO - Biological Process

GO:0033344 : cholesterol efflux (https://www.ebi.ac.uk/QuickGO/term/GO:0033344)
GO:0006707 : cholesterol catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006707)
GO:0042632 : cholesterol homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0042632)
GO:0034384 : high-density lipoprotein particle clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0034384)
GO:0034375 : high-density lipoprotein particle remodeling
(https://www.ebi.ac.uk/QuickGO/term/GO:0034375)
GO:0051000 : positive regulation of nitric-oxide synthase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0051000)
GO:0043691 : reverse cholesterol transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0043691)
GO:0070328 : triglyceride homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0070328)
GO:0010867 : positive regulation of triglyceride biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0010867)
GO:0034383 : low-density lipoprotein particle clearance
(https://www.ebi.ac.uk/QuickGO/term/GO:0034383)
GO:0010886 : positive regulation of cholesterol storage
(https://www.ebi.ac.uk/QuickGO/term/GO:0010886)
GO:0050892 : intestinal absorption (https://www.ebi.ac.uk/QuickGO/term/GO:0050892)
GO:0043534 : blood vessel endothelial cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0043534)
GO:0044406 : adhesion of symbiont to host
(https://www.ebi.ac.uk/QuickGO/term/GO:0044406)
GO:0006702 : androgen biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006702)
GO:0070508 : cholesterol import (https://www.ebi.ac.uk/QuickGO/term/GO:0070508)
GO:0030301 : cholesterol transport (https://www.ebi.ac.uk/QuickGO/term/GO:0030301)
GO:0032497 : detection of lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0032497)
GO:0001935 : endothelial cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001935)
GO:0006869 : lipid transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006869)
GO:0015920 : lipopolysaccharide transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0015920)
GO:0006910 : phagocytosis, recognition
(https://www.ebi.ac.uk/QuickGO/term/GO:0006910)
GO:0015914 : phospholipid transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015914)
GO:0043654 : recognition of apoptotic cell
(https://www.ebi.ac.uk/QuickGO/term/GO:0043654)
GO:0050764 : regulation of phagocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0050764)
GO:0010899 : regulation of phosphatidylcholine catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0010899)
GO:0035461 : vitamin transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0035461)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005901 : caveola (https://www.ebi.ac.uk/QuickGO/term/GO:0005901)
GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)
GO:0031528 : microvillus membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031528)

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

-

Molecular Details of the Mutation

T>G nucleotide substitution in the splice-donor site immediately downstream of exon 4 of the SCARB1 gene. This mutation results in abnormal splicing; with the most abundant transcript lacking exon 4. Isoform 4 of SCARB1; the most abundant transcript isoform in the white recessive canary; is predicted to lack 68 amino acids; a deficiency which disrupts SCARB1 protein function.

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

High-density lipoprotein receptor SCARB1 is required for carotenoid coloration in birds. (2017) (https://pubmed.ncbi.nlm.nih.gov/28465440)

Authors

Toomey MB; Lopes RJ; AraÃºjo PM; Johnson JD; Gazda MA; Afonso S; Mota PG; Koch RE; Hill GE; Corbo JC; Carneiro M

Abstract

Yellow, orange, and red coloration is a fundamental aspect of avian diversity and serves as an important signal in mate choice and aggressive interactions. This coloration is often produced through the deposition of diet-derived carotenoid pigments, yet the mechanisms of carotenoid uptake and transport are not well-understood. The white recessive breed of the common canary (Serinus canaria), which carries an autosomal recessive mutation that renders its plumage pure white, provides a unique opportunity to investigate mechanisms of carotenoid coloration. We carried out detailed genomic and biochemical analyses comparing the white recessive with yellow and red breeds of canaries. Biochemical analysis revealed that carotenoids are absent or at very low concentrations in feathers and several tissues of white recessive canaries, consistent with a genetic defect in carotenoid uptake. Using a combination of genetic mapping approaches, we show that the white recessive allele is due to a splice donor site mutation in the scavenger receptor B1 (SCARB1; also known as SR-B1) gene. This mutation results in abnormal splicing, with the most abundant transcript lacking exon 4. Through functional assays, we further demonstrate that wild-type SCARB1 promotes cellular uptake of carotenoids but that this function is lost in the predominant mutant isoform in white recessive canaries. Our results indicate that SCARB1 is an essential mediator of the expression of carotenoid-based coloration in birds, and suggest a potential link between visual displays and lipid metabolism.

Additional References

RELATED GEPHE

Related Genes

1 (BCO2 = beta-carotene oxygenase 2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9135^/and+Trait=Carotenoid content/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

The mutation is in an intron and affects @Splicing and thus the coding sequence.