GEPHE SUMMARY

Gephebase Gene
Oca2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Oca2^#gephebase-summary-title)

GepheID
GP00001619

Entry Status
Published

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (albinism)^#gephebase-summary-title)

Trait State in Taxon A
Corn snake with black borders

Trait State in Taxon B
Amelanistic corn snake with white borders

Ancestral State
Taxon A

Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Pantherophis guttatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Pantherophis guttatus^#gephebase-summary-title)

Common Name
-

Synonyms
Elaphe guttata; ZMUU 147; ZMUU:147

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Serpentes; Colubroidea; Colubridae; Colubrinae; Pantherophis

Parent
Pantherophis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 201800)

NCBI Taxonomy ID
94885 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 94885)

is Taxon A an Infraspecies?
Yes

Taxon A Description
Corn snake with black borders

Taxon B

Latin Name
Pantherophis guttatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Pantherophis guttatus^#gephebase-summary-title)

Common Name
-

Synonyms
Elaphe guttata; ZMUU 147; ZMUU:147

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Serpentes; Colubroidea; Colubridae; Colubrinae; Pantherophis

Parent
Pantherophis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 201800)

NCBI Taxonomy ID
94885 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 94885)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Corn snake with white borders

GENOTYPIC CHANGE

Generic Gene Name
OCA2

Synonyms
P; SHEP1; BEY; PED; BEY1; BEY2; BOCA; EYCL; HCL3; EYCL2; EYCL3; D15S12

String
9606.ENSP00000346659 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000346659)

Sequence Similarities
Belongs to the CitM (TC 2.A.11) transporter family.

GO - Molecular Function
GO:0005215 : transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005215)
GO:0005302 : L-tyrosine transmembrane transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005302)

GO - Biological Process

UniProtKB Homo sapiens
Q04671 (http://www.uniprot.org/uniprot/Q04671)

GenebankID or UniProtKB
()

GO:0042438 : melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
GO:0030318 : melanocyte differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0030318)
GO:0007286 : spermatid development
(https://www.ebi.ac.uk/QuickGO/term/GO:0007286)
GO:0006726 : eye pigment biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006726)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0010008 : endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0010008)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)
GO:0005765 : lysosomal membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005765)
GO:0033162 : melanosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

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

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

Insertion Size

100-999 bp

Molecular Details of the Mutation

insertion of an LTR-retrotransposon (5832-bp) in the 11th intron resulting after splicing in an additional 397-bp fragment constituted of 3 new exons inserted between exons 11 and 12. Generates truncated protein with two stop codons

Experimental Evidence

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

Main Reference

Amelanism in the corn snake is associated with the insertion of an LTR-retrotransposon in the OCA2 gene. (2015) (https://pubmed.ncbi.nlm.nih.gov/26597053)

Authors

Saenko SV; Lamichhaney S; Martinez Barrio A; Rafati N; Andersson L; Milinkovitch MC

Abstract

The corn snake (Pantherophis guttatus) is a new model species particularly appropriate for investigating the processes generating colours in reptiles because numerous colour and pattern mutants have been isolated in the last five decades. Using our captive-bred colony of corn snakes, transcriptomic and genomic next-generation sequencing, exome assembly, and genotyping of SNPs in multiple families, we delimit the genomic interval bearing the causal mutation of amelanism, the oldest colour variant observed in that species. Proceeding with sequencing the candidate gene OCA2 in the uncovered genomic interval, we identify that the insertion of an LTR-retrotransposon in its 11(th) intron results in a considerable truncation of the p protein and likely constitutes the causal mutation of amelanism in corn snakes. As amelanistic snakes exhibit white, instead of black, borders around an otherwise normal pattern of dorsal orange saddles and lateral blotches, our results indicate that melanocytes lacking melanin are able to participate to the normal patterning of other colours in the skin. In combination with research in the zebrafish, this work opens the perspective of using corn snake colour and pattern variants to investigate the generative processes of skin colour patterning shared among major vertebrate lineages.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@TE; possible null mutation