GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000746

Entry Status

Published

Main curator

Courtier

PHENOTYPIC CHANGE

Trait #1

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

pigmented surface fish

Trait State in Taxon B

unpigmented fish - Pachon cave

Trait #2

Trait Category

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

Trait

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

Trait State in Taxon A

pigmented surface fish

Trait State in Taxon B

unpigmented fish - Pachon cave

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

Mexican tetra

Synonyms

Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Characiphysae; Characiformes; Characoidei; Characidae; Characidae incertae sedis; Astyanax clade; Astyanax

Parent

Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7993)

NCBI Taxonomy ID

7994 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

Mexican tetra

Synonyms

Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Otomorpha; Ostariophysi; Otophysi; Characiphysae; Characiformes; Characoidei; Characidae; Characidae incertae sedis; Astyanax clade; Astyanax

Parent

Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7993)

NCBI Taxonomy ID

7994 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Astyanax mexicanus - Pachon cave

GENOTYPIC CHANGE

Generic Gene Name

Oca2

Synonyms

p; D7Nic1; p<cas>; D7H15S12; D7Icr28RN; P

String

10090.ENSMUSP00000032633 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000032633)

Sequence Similarities

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

GO - Molecular Function

-

GO - Biological Process

GO:0055085 : transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0055085)
GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0043473 : pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0043473)
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)
GO:0048066 : developmental pigmentation (https://www.ebi.ac.uk/QuickGO/term/GO:0048066)
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 - Cellular Component

GO:0016021 : integral component of membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
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)

UniProtKB Mus musculus

Q62052 (http://www.uniprot.org/uniprot/Q62052)

GenebankID or UniProtKB

ABB29299 (https://www.ncbi.nlm.nih.gov/nuccore/ABB29299)

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

-

Molecular Details of the Mutation

Almost complete deletion of exon 24; + 2 a.a subsitutions at conserved positions - the two point mutations do not drastically affect the function of OCA2 in cell lines suggesting that the exon 24 deletion is the mutation that causes albinism in the PachÃ³n population

Experimental Evidence

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

Main Reference

Genetic analysis of cavefish reveals molecular convergence in the evolution of albinism. (2006) (https://pubmed.ncbi.nlm.nih.gov/16341223)

Authors

Protas ME; Hersey C; Kochanek D; Zhou Y; Wilkens H; Jeffery WR; Zon LI; Borowsky R; Tabin CJ

Abstract

The genetic basis of vertebrate morphological evolution has traditionally been very difficult to examine in naturally occurring populations. Here we describe the generation of a genome-wide linkage map to allow quantitative trait analysis of evolutionarily derived morphologies in the Mexican cave tetra, a species that has, in a series of independent caves, repeatedly evolved specialized characteristics adapted to a unique and well-studied ecological environment. We focused on the trait of albinism and discovered that it is linked to Oca2, a known pigmentation gene, in two cave populations. We found different deletions in Oca2 in each population and, using a cell-based assay, showed that both cause loss of function of the corresponding protein, OCA2. Thus, the two cave populations evolved albinism independently, through similar mutational events.

Additional References

Behavioural changes controlled by catecholaminergic systems explain recurrent loss of pigmentation in cavefish. (2018) (https://pubmed.ncbi.nlm.nih.gov/29720416)
CRISPR mutagenesis confirms the role of oca2 in melanin pigmentation in Astyanax mexicanus. (2018) (https://pubmed.ncbi.nlm.nih.gov/29555241)

RELATED GEPHE

Related Genes

1 (MC1R) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^7994^/and+Trait=Coloration/or+Taxon ID=^7994^/and+Trait=Catecholamine metabolism/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Oca2^/and+Taxon ID=^7994^/or+Gene Gephebase=^Oca2^/and+Taxon ID=^7994^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@Parallelism @Pleiotropy https://omia.org/OMIA002130/7994/