GEPHE SUMMARY

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

GepheID
GP00000745

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
Astyanax mexicanus surface

Trait State in Taxon B
Astyanax mexicanus Molino

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
Astyanax mexicanus surface

Trait State in Taxon B
Astyanax mexicanus Molino

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 Molino

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)

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

Deletion of exon 21

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)

UniProtKB Mus musculus

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

GenebankID or UniProtKB

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

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/