

GEPHE SUMMARY

Oca2 (https://www.gephebase.org/search-criteria?/and+Gene)	Gephebase Gene	GP00000746	GepheID
Gephebase="Oca2"#gephebase-summary-title)			Main curator
Published	Entry Status	Courtier	

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait)	Trait
Category="Morphology"#gephebase-summary-title)	Trait State in Taxon A
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait)	Trait State in Taxon B
Criteria="Coloration (albinism)"#gephebase-summary-title)	unpigmented fish - Pachon cave
pigmented surface fish	
unpigmented fish - Pachon cave	

Trait #2	Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait)	Trait
Category="Physiology"#gephebase-summary-title)	Trait State in Taxon A
Catecholamine metabolism (https://www.gephebase.org/search-criteria?/and+Trait)	Trait State in Taxon B
Criteria="Catecholamine metabolism"#gephebase-summary-title)	unpigmented fish - Pachon cave
pigmented surface fish	
unpigmented fish - Pachon cave	

Taxon A	Ancestral State
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic)	Taxonomic Status
Status="Intraspecific"#gephebase-summary-title)	

Taxon A	Latin Name	Taxon B	Latin Name
Astyanax mexicanus (https://www.gephebase.org/search-criteria?/and+Taxon)			
Mexican tetra	Mexican tetra	Mexican tetra	Mexican tetra
Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)	Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)	Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)	Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)
species	species	species	species
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	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	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	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
Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993)	Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993)	Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993)	Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993)
7994 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994)			
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
			Taxon B Description
			Astyanax mexicanus - Pachon cave

GENOTYPIC CHANGE

Oca2	Generic Gene Name	Q62052 (http://www.uniprot.org/uniprot/Q62052)	UniProtKB Mus musculus
p; D7Nic1; p<cas>; D7H15S12; D7lcr28RN; P	Synonyms	ABB29299 (https://www.ncbi.nlm.nih.gov/nucore/ABB29299)	GenebankID or UniProtKB
10090.ENSMUSP00000032633 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000032633)	String		
Belongs to the CitM (TC 2.A.11) transporter family.	Sequence Similarities		
-	GO - Molecular Function		
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 - Biological Process		
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)	GO - Cellular Component		
Yes (https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title)	Presumptive Null		
Coding (https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title)	Molecular Type		
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion^#gephebase-summary-title)	Aberration Type		
-	Deletion Size		
Almost complete deletion of exon 24; + 2 a.a substitutions 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 Pach ^Å population	Molecular Details of the Mutation		
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Linkage+Mapping^#gephebase-summary-title)	Experimental Evidence		
Genetic analysis of cavefish reveals molecular convergence in the evolution of albinism. (2006) (https://pubmed.ncbi.nlm.nih.gov/16341223)	Main Reference		
Protas ME; Hersey C; Kochanek D; Zhou Y; Wilkens H; Jeffery WR; Zon LI; Borowsky R; Tabin CJ	Authors		
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.	Abstract		
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)	Additional References		

RELATED GEPHE

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 Genes
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)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

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