

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000567	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)	Trait Category		
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+albinism^#gephebase-summary-title)	Trait		
Surface fish (pigmented)	Trait State in Taxon A		
Cavefish with depigmented phenotype (brown mutation ; distinct from albinism mutation)	Trait State in Taxon B		
	Ancestral State		
	Taxon A		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title)	Taxonomic Status		

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

GENOTYPIC CHANGE

MC1R	Generic Gene Name	Q01726 (http://www.uniprot.org/uniprot/Q01726)	UniProtKB Homo sapiens
CMM5; MSH-R; SHEP2; MSHR	Synonyms	ACN39571 (https://www.ncbi.nlm.nih.gov/nuccore/ACN39571)	GenebankID or UniProtKB
9606.ENSPP00000451605 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)	GO - Molecular Function		
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)			

GO:0004980 : melanocyte-stimulating hormone receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004980>)
GO:0031625 : ubiquitin protein ligase binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031625>)

GO - Biological Process

GO:0007275 : multicellular organism development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007275>)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
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:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)
GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
GO:0019233 : sensory perception of pain
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019233>)
GO:0007189 : adenylate cyclase-activating G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)
GO:0035556 : intracellular signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035556>)
GO:0007187 : G protein-coupled receptor signaling pathway, coupled to cyclic nucleotide second messenger (<https://www.ebi.ac.uk/QuickGO/term/GO:0007187>)
GO:0032720 : negative regulation of tumor necrosis factor production
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032720>)
GO:0010739 : positive regulation of protein kinase A signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010739>)
GO:0090037 : positive regulation of protein kinase C signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0090037>)
GO:0009650 : UV protection (<https://www.ebi.ac.uk/QuickGO/term/GO:0009650>)
GO:0070914 : UV-damage excision repair
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070914>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)

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

1-9 bp

Molecular Details of the Mutation

2bp deletion resulting in frameshift

Experimental Evidence

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

Main Reference

A novel role for Mc1r in the parallel evolution of depigmentation in independent populations of the cavefish *Astyanax mexicanus*. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19119422>)

Authors

Gross JB; Borowsky R; Tabin CJ

Abstract

The evolution of degenerate characteristics remains a poorly understood phenomenon. Only recently has the identification of mutations underlying regressive phenotypes become accessible through the use of genetic analyses. Focusing on the Mexican cave tetra *Astyanax mexicanus*, we describe, here, an analysis of the brown mutation, which was first described in the literature nearly 40 years ago. This phenotype causes reduced melanin content, decreased melanophore number, and brownish eyes in convergent cave forms of *A. mexicanus*. Crosses demonstrate non-complementation of the brown phenotype in F(2) individuals derived from two independent cave populations: Pach³n and the linked Yerbaniz and Japon^s caves, indicating the same locus is responsible for reduced pigmentation in these fish. While the brown mutant phenotype arose prior to the fixation of albinism in Pach³n cave individuals, it is unclear whether the brown mutation arose before or after the fixation of albinism in the linked Yerbaniz/Japon^s caves. Using a QTL approach combined with sequence and functional analyses, we have discovered that two distinct genetic alterations in the coding sequence of the gene *Mc1r* cause reduced pigmentation associated with the brown mutant phenotype in these caves. Our analysis identifies a novel role for *Mc1r* in the evolution of degenerative phenotypes in blind Mexican cavefish. Further, the brown phenotype has arisen independently in geographically separate caves, mediated through different mutations of the same gene. This example of parallelism indicates that certain genes are frequent targets of mutation in the repeated evolution of regressive phenotypes in cave-adapted species.

Additional References

RELATED GEPHE

Related Genes

1 (Oca2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~7994*/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~MC1R*/and+Taxon ID=~7994*/or+Gene Gephebase=~MC1R*/and+Taxon ID=~7994*#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

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