

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R~#gephebase-summary-title)	Gephebase Gene	GP00000568	GepheID
Published	Entry Status	Martin	Main curator

Morphology (https://www.gephebase.org/search-criteria?/and+Trait=^Morphology^#gephebase-summary-title)		Trait Category	
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (albinism)^#gephebase-summary-title)		Trait	
Surface cavefish (pigmented)		Trait State in Taxon A	
Cavefish with depigmented phenotype (brown mutation ; distinct from albinism mutation)		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Latin Name		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)	
Common Name		Common Name	
Mexican tetra		Mexican tetra	
Synonyms		Synonyms	
Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)		Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)	
Rank		Rank	
species		species	
Lineage		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		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		Parent	
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)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
7994		7994	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		Yes	
		Taxon B Description	
		Astyanax mexicanus - Yerbaniz and Japon_s caves (possible gene flow)	

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/nucore/ACN39571)	GenebankID or UniProtKB
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	String		
	Sequence Similarities		
Belongs to the G-protein coupled receptor 1 family.			
	GO - Molecular Function		
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)			
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>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null="+No^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type="+Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular Type=))

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Arg164Cys (position 160 in human; so this mutation resembles the human R160W hypomorphic human mutation)

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

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

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

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephabase=`MC1R`/and+Taxon ID=`7994`/or+Gene Gephabase=`MC1R`/and+Taxon ID=`7994`#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS

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