

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

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	
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+Synonyms=~Astyanax+mexicanus~#gephebase-summary-title)		Astyanax mexicanus (https://www.gephebase.org/search-criteria?/and+Taxon+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 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994)		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 - Pachon cave	

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR	String	
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)	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
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
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 <i>Astyanax mexicanus</i> . (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: Pach^{Å³n} and the linked Yerbaniz and Japon^{Å©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 Pach^{Å³n} cave individuals, it is unclear whether the brown mutation arose before or after the fixation of albinism in the linked Yerbaniz/Japon^{Å©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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