

## GEPHE SUMMARY

MC4R ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC4R^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^MC4R^#gephebase-summary-title</a> )	Gephebase Gene	GP00000637	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Behavior ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Behavior^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Behavior^#gephebase-summary-title</a> )	Trait Category		
Feeding behavior ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Feeding+behavior^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Feeding+behavior^#gephebase-summary-title</a> )	Trait		
Astyanax mexicanus surface	Trait State in Taxon A		
Astyanax mexicanus - Tinaja cave (nutrient-poor; hyperphagia)	Trait State in Taxon B		
Data not curated	Ancestral State		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
	Latin Name		Latin Name
Astyanax mexicanus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Astyanax+mexicanus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Astyanax+mexicanus^#gephebase-summary-title</a> )	Latin Name	Astyanax mexicanus ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Astyanax+mexicanus^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Astyanax+mexicanus^#gephebase-summary-title</a> )	Latin Name
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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993</a> )	Parent	Astyanax () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7993</a> )	Parent
7994 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994</a> )	NCBI Taxonomy ID	7994 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7994</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
			Taxon B Description
			Astyanax mexicanus - Tinaja cave (nutrient-poor; hyperphagia)

## GENOTYPIC CHANGE

mc4r	Generic Gene Name	BoV1P1 ( <a href="http://www.uniprot.org/uniprot/BoV1P1">http://www.uniprot.org/uniprot/BoV1P1</a> )	UniProtKB Danio rerio
zMC4; MC4-R; si:dkey-151m6.62	Synonyms	0	GenebankID or UniProtKB
7955.ENS DARP00000027547 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=7955.ENS DARP00000027547">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=7955.ENS DARP00000027547</a> )	String		
Belongs to the G-protein coupled receptor 1 family.	Sequence Similarities		
GO:0004977 : melanocortin receptor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004977">https://www.ebi.ac.uk/QuickGO/term/GO:0004977</a> )	GO - Molecular Function		
GO:0040008 : regulation of growth ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0040008">https://www.ebi.ac.uk/QuickGO/term/GO:0040008</a> )	GO - Biological Process		

GO:0007189 : adenylylase cyclase-activating G protein-coupled receptor signaling pathway

(<https://www.ebi.ac.uk/QuickGO/term/GO:0007189>)

GO:2000252 : negative regulation of feeding behavior

(<https://www.ebi.ac.uk/QuickGO/term/GO:2000252>)

GO:0097009 : energy homeostasis (<https://www.ebi.ac.uk/QuickGO/term/GO:0097009>)

GO:0006112 : energy reserve metabolic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0006112>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

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

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Gly145Ser (candidate mutation)

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

Melanocortin 4 receptor mutations contribute to the adaptation of cavefish to nutrient-poor conditions. (2015) (<https://pubmed.ncbi.nlm.nih.gov/26170297>)

Authors

Aspiras AC; Rohner N; Martineau B; Borowsky RL; Tabin CJ

Abstract

Despite recent advances in the understanding of morphological evolution, the genetic underpinnings of behavioral and physiological evolution remain largely unknown. Here, we study the metabolic changes that evolved in independently derived populations of the Mexican cavefish, *Astyanax mexicanus*. A hallmark of cave environments is scarcity of food. Cavefish populations rely almost entirely on sporadic food input from outside of the caves. To survive under these conditions, cavefish have evolved a range of adaptations, including starvation resistance and binge eating when food becomes available. The use of these adaptive strategies differs among independently derived cave populations. Although all cavefish populations tested lose weight more slowly than their surface conspecifics during restricted rations, only a subset of cavefish populations consume more food than their surface counterparts. A candidate gene-based screen led to the identification of coding mutations in conserved residues of the melanocortin 4 receptor (MC4R) gene, contributing to the insatiable appetite found in some populations of cavefish. Intriguingly, one of the mutated residues has been shown to be linked to obesity in humans. We demonstrate that the allele results in both reduced maximal response and reduced basal activity of the receptor in vitro. We further validate in vivo that the mutated allele contributes to elevated appetite, growth, and starvation resistance. The allele appears to be fixed in cave populations in which the overeating phenotype is present. The presence of the same allele in multiple caves appears to be due to selection from standing genetic variation present in surface populations.

Additional References

## RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

@Pleiotropy - Parallelism: the Gly-to-Ser shift in the encoded MC4R protein is particularly interesting because mutations in this highly conserved residue are associated with obesity in human patients. The coding mutation needs functional verification

