GEPHE SUMMARY

Gephebase Gene
MC4R (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MC4R^#gephebase-summary-title)

GepheID
GP00000637

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Behavior (https://www.gephebase.org/search-criteria?/and+Trait Category=^Behavior^#gephebase-summary-title)

Trait
Feeding behavior (https://www.gephebase.org/search-criteria?/and+Trait=^Feeding behavior^#gephebase-summary-title)

Trait State in Taxon A
Astyanax mexicanus surface

Trait State in Taxon B
Astyanax mexicanus - Tinaja cave (nutrient-poor; hyperphagia)

Ancestral State
Data not curated

Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Astyanax mexicanus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Astyanax mexicanus^#gephebase-summary-title)

Common Name
Mexican tetra

Synonyms
Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)

Rank
species

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

Parent
Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7993)

NCBI Taxonomy ID
7994 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Astyanax mexicanus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Astyanax mexicanus^#gephebase-summary-title)

Common Name
Mexican tetra

Synonyms
Mexican tetra; blind cave fish; Astyanax mexicanus (De Filippi, 1853)

Rank
species

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

Parent
Astyanax () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7993)

NCBI Taxonomy ID
7994 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7994)

is Taxon B an Infraspecies?
Yes

Taxon B Description
Astyanax mexicanus - Tinaja cave (nutrient-poor; hyperphagia)

GENOTYPIC CHANGE

Generic Gene Name
mc4r

Synonyms
zMC4; MC4-R; si:dkey-151m6.62

String
7955.ENSDARP00000027547 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=7955.ENSDARP00000027547)

Sequence Similarities
Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function
GO:0004977 : melanocortin receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004977)

GO - Biological Process
GO:0040008 : regulation of growth (https://www.ebi.ac.uk/QuickGO/term/GO:0040008)

UniProtKB Danio rerio
B0V1P1 (http://www.uniprot.org/uniprot/B0V1P1)

GenebankID or UniProtKB
()

GO:0007189 : adenylate 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 (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Gly145Ser (candidate mutation)

Experimental Evidence

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

	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

Related Genes

No matches found.

Related Haplotypes

No matches found.

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