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Mc1r gene in *Astroblepus pholeter* and *Astyanax mexicanus*: Convergent regressive evolution of pigmentation across cavefish species

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Abstract

Cave-adapted organisms are often characterized by a reduction in pigmentation, eyesight, and enhanced mechanosensory functions. Previous studies have described the genetic basis for a depigmented phenotype in multiple independent populations of the Blind Mexican Tetra, *Astyanax mexicanus*; the reduction in melanin content (brown; Mc1r). At least seven wild populations express the brown phenotype. In three populations, there are two different coding sequence alterations affecting Mc1r and the remaining four populations show the accumulation of sequence mutations affecting the 5′ regulatory region. Thus, the Mc1r gene has been the repeated and independent location of mutations in *Astyanax*. As such, it would appear that this gene is a target during regressive evolution of cave adapted organisms. If this is the case, it would be expected that other cave adapted fish would have mutations in the same gene. We study here the stygobitic catfish *Astroblepus pholeter*, a depigmented fish found within some river caves in Ecuador. *A. pholeter* displays mutations in ultra-conserved areas of the pigment-controlling gene, Mc1r, that have been linked to pigment regulation in other organisms. It is thus concluded that Mc1r, a gene known to control pigment variation in many organisms, may be the target of cavernicole regressive evolution across species in different families of fish.

Keywords

Astroblepus pholeter, Chaetostoma microps, Astyanax, cave, catfish, Jumandi cave, troglobite, reduction in pigmentation, regressive evolution, OCA2, Mc1r.