

GEPHE SUMMARY

MC1R (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MC1R^#gephebase-summary-title)	Gephebase Gene	GP00000628	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Coloration (scales) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration(scales)^#gephebase-summary-title)		Trait	
Holbrookia maculata		Trait State in Taxon A	
Holbrookia maculata		Trait State in Taxon B	
Data not curated		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Holbrookia maculata (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Holbrookia maculata^#gephebase-summary-title)		Holbrookia maculata (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Holbrookia maculata^#gephebase-summary-title)	
lesser earless lizard		lesser earless lizard	
lesser earless lizard; Holbrookia maculata Girard 1851		lesser earless lizard; Holbrookia maculata Girard 1851	
species		species	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Iguania; Phrynosomatidae; Phrynosomatinae; Holbrookia		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Iguania; Phrynosomatidae; Phrynosomatinae; Holbrookia	
Holbrookia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=43596)		Holbrookia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=43596)	
43597 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=43597)		43597 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=43597)	
No		No	

GENOTYPIC CHANGE

MC1R	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q01726 (http://www.uniprot.org/uniprot/Q01726)
	String	GenebankID or UniProtKB
CMM5; MSH-R; SHEP2; MSHR		AAT90228 (https://www.ncbi.nlm.nih.gov/nuccore/AAT90228)
9606.ENSPP00000451605 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000451605)		
Belongs to the G-protein coupled receptor 1 family.		Sequence Similarities
GO:0008528 : G protein-coupled peptide receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008528)		GO - Molecular Function
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>)

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

V168I; but failed functional validation in Rosenblum et al. 2010

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence+Candidate Gene^#gephebase-summary-title>)

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

Main Reference

Adaptive reptile color variation and the evolution of the Mc1r gene. (2004) (<https://pubmed.ncbi.nlm.nih.gov/15446431>)

Authors

Rosenblum EB; Hoekstra HE; Nachman MW

Abstract

The wealth of information on the genetics of pigmentation and the clear fitness consequences of many pigmentation phenotypes provide an opportunity to study the molecular basis of an ecologically important trait. The melanocortin-1 receptor (Mc1r) is responsible for intraspecific color variation in mammals and birds. Here, we study the molecular evolution of Mc1r and investigate its role in adaptive intraspecific color differences in reptiles. We sequenced the complete Mc1r locus in seven phylogenetically diverse squamate species with melanic or blanded forms associated with different colored substrates or thermal environments. We found that patterns of amino acid substitution across different regions of the receptor are similar to the patterns seen in mammals, suggesting comparable levels of constraint and probably a conserved function for Mc1r in mammals and reptiles. We also found high levels of silent-site heterozygosity in all species, consistent with a high mutation rate or large long-term effective population size. Mc1r polymorphisms were strongly associated with color differences in *Holbrookia maculata* and *Aspidoscelis inornata*. In *A. inornata*, several observations suggest that Mc1r mutations may contribute to differences in color: (1) a strong association is observed between one Mc1r amino acid substitution and dorsal color; (2) no significant population structure was detected among individuals from these populations at the mitochondrial ND4 gene; (3) the distribution of allele frequencies at Mc1r deviates from neutral expectations; and (4) patterns of linkage disequilibrium at Mc1r are consistent with recent selection. This study provides comparative data on a nuclear gene in reptiles and highlights the utility of a candidate-gene approach for understanding the evolution of genes involved in vertebrate adaptation.

Additional References

Molecular and functional basis of phenotypic convergence in white lizards at White Sands. (2010) (<https://pubmed.ncbi.nlm.nih.gov/20080544>)

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS