

	Gephebase Gene	GepheID
Oca2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~Oca2^#gephebase-summary-title)	GP00001955	
	Entry Status	Courtier
Published		Main curator

Trait Category		Trait	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=~Morphology~#gephebase-summary-title)			
			Trait
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait=~Coloration+albinism~#gephebase-summary-title)			
		Trait State in Taxon A	
wild-type coloration			
		Trait State in Taxon B	
absence of all black and brown pigmentation in the skin; iris and retina			
		Ancestral State	
Taxon A			
		Taxonomic Status	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific~#gephebase-summary-title)			
Taxon A		Taxon B	
	Latin Name		Latin Name
Melanochromis auratus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Melanochromis+auratus~#gephebase-summary-title)		Melanochromis auratus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=~Melanochromis+auratus~#gephebase-summary-title)	
	Common Name		Common Name
-		-	
	Synonyms		Synonyms
Chromis auratus; Tilapia aurea; golden mbuna; Melanochromis auratus (Boulenger, 1897)		Chromis auratus; Tilapia aurea; golden mbuna; Melanochromis auratus (Boulenger, 1897)	
	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; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percormorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Melanochromis		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percormorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Melanochromis	
	Parent		Parent
Melanochromis (black cichlids) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27750)		Melanochromis (black cichlids) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27750)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
27751 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27751)		27751 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27751)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

Oca2	Generic Gene Name	UniProtKB Mus musculus
	Synonyms	Q62052 (http://www.uniprot.org/uniprot/Q62052)
p; D7Nic1; p<cas>; D7H15S12; D7lcr28RN; P		GenebankID or UniProtKB Mus musculus
	String	Q62052 (https://www.ncbi.nlm.nih.gov/nuccore/Q62052)
10090.ENSMUSP00000032633 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000032633)	Sequence Similarities	
Belongs to the CitM (TC 2.A.11) transporter family.	GO - Molecular Function	
-	GO - Biological Process	
GO:0055085 : transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0055085)		
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:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0048066 : developmental pigmentation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048066>)
GO:0030318 : melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030318>)
GO:0007286 : spermatid development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007286>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
GO:0005789 : endoplasmic reticulum membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>)
GO:0005765 : lysosomal membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005765>)
GO:0033162 : melanosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

	Presumptive Null
Yes (#gephebase-summary-title)	
	Molecular Type
Coding (#gephebase-summary-title)	
	Aberration Type
Deletion (#gephebase-summary-title)	
	Deletion Size
1-10 kb	
	Molecular Details of the Mutation
deletion covering a total of 5.4kb including exon 2 and parts of the flanking introns 1 and 2	
	Experimental Evidence
Candidate Gene (#gephebase-summary-title)	
	Main Reference
Genome of the Malawi golden cichlid fish (<i>Melanochromis auratus</i>) reveals exon loss of oca2 in an amelanistic morph. (2019) (https://pubmed.ncbi.nlm.nih.gov/31131985)	
	Authors
Kratochwil CF; Urban S; Meyer A	

Abstract

The tropical freshwater fish family Cichlidae is famous for its record-breaking rates of speciation and diversity in colors and color patterns. Here, we sequenced the genome of the Lake Malawi cichlid *Melanochromis auratus* to study the genetic basis of an amelanistic morph of this species that lacks the typical melanistic stripes and markings. Genome sequencing of the amelanistic and wild-type morph revealed the loss of the second exon of the known pigmentation gene *oculocutaneous albinism II* (*oca2*), also known as *p*(ink-eyed dilution) gene or melanocyte-specific transporter gene. Additional genotyping confirms the complete association with this recessive Mendelian phenotype. The deletion results in a shorter transcript, lacking an acidic di-leucine domain that is crucial for trafficking of the *Oca2* protein to melanosomes. The fact that *oca2* is involved in a wide range of amelanistic morphs across vertebrates demonstrates its highly conserved function.

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Additional References

RELATED GEPHE

	Related Genes
No matches found.	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

COMMENTS