

GEPHE SUMMARY

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

PHENOTYPIC CHANGE

Trait Category	
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	Latin Name	Taxon B	Latin Name
Melanochromis auratus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Melanochromis+auratus^#gephebase-summary-title)	Common Name	Melanochromis auratus (https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Melanochromis+auratus^#gephebase-summary-title)	Common Name
-	Synonyms	-	Synonyms
Chromis auratus; Tilapia aurea; golden mbuna; Melanochromis auratus (Boulenger, 1897)	Rank	Chromis auratus; Tilapia aurea; golden mbuna; Melanochromis auratus (Boulenger, 1897)	Rank
species	Lineage	species	Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Melanochromis	Parent	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Melanochromis	Parent
Melanochromis (black cichlids) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27750)	NCBI Taxonomy ID	Melanochromis (black cichlids) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 27750)	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 Infraspecies?		is Taxon B an Infraspecies?
No		No	

GENOTYPIC CHANGE

Generic Gene Name	UniProtKB Mus musculus
Oca2	Q62052 (http://www.uniprot.org/uniprot/Q62052)
p; D7Nic1; p<cas>; D7H15S12; D7Icr28RN; P	GenebankID or UniProtKB Mus musculus
10090.ENSMUSP00000032633 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000032633)	Q62052 (https://www.ncbi.nlm.nih.gov/nuccore/Q62052)
Belongs to the CitM (TC 2.A.11) transporter family.	Sequence Similarities
-	GO - Molecular Function
GO:0055085 : transmembrane transport (https://www.ebi.ac.uk/QuickGO/term/GO:0055085)	GO - Biological Process
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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes))

Molecular Type

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

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene))

Main Reference

Genome of the Malawi golden cichlid fish (*Melanochromis auratus*) 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