

GEPHE SUMMARY

	Gephebase Gene		GepheID
Rx1 ( <a +rx1+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Rx1+"#gephebase-summary-title</a> )		GP00001441	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology ( <a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title</a> )			
	Trait		
Color vision (expression of SWS2b opsin) ( <a +color+vision+(expression+of+sws2b+opsin)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Color+vision+(expression+of+SWS2b+opsin)+"#gephebase-summary-title</a> )			
	Trait State in Taxon A		
Expresses predominantly SWS2B			
	Trait State in Taxon B		
Expresses predominantly SWS2A			
	Ancestral State		
Unknown			
	Taxonomic Status		
Interspecific ( <a +interspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Interspecific+"#gephebase-summary-title</a> )			
	Taxon A	Taxon B	
	Latin Name		Latin Name
Aulonocara baenschii ( <a +aulonocara+baenschii+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Aulonocara+baenschii+"#gephebase-summary-title</a> )		Tramitichromis intermedius ( <a +tramitichromis+intermedius+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Tramitichromis+intermedius+"#gephebase-summary-title</a> )	
	Common Name		Common Name
Nkhomo-benga peacock cichlid		-	
	Synonyms		Synonyms
Nkhomo-benga peacock cichlid; Nkhomo-benga peacock; Aulonocara baenschii Meyer & Riehl, 1985		-	
	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; Percomorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Aulonocara		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; Percomorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Tramitichromis	
	Parent		Parent
Aulonocara () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=27780">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=27780</a> )		Tramitichromis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=136859">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=136859</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
143496 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=143496">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=143496</a> )		323801 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=323801">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=323801</a> )	
	is Taxon A an Infrasppecies?		is Taxon B an Infrasppecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Danio rerio
rx1		O42356 ( <a href="http://www.uniprot.org/uniprot/O42356">http://www.uniprot.org/uniprot/O42356</a> )	
	Synonyms		GenebankID or UniProtKB
-		0	
	String		
7955.ENSADARP00000096944 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=7955.ENSADARP00000096944">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=7955.ENSADARP00000096944</a> )			
	Sequence Similarities		
Belongs to the paired homeobox family, Bicoid subfamily.			
	GO - Molecular Function		
GO:0003700 : DNA-binding transcription factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003700">https://www.ebi.ac.uk/QuickGO/term/GO:0003700</a> )			
GO:0043565 : sequence-specific DNA binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0043565">https://www.ebi.ac.uk/QuickGO/term/GO:0043565</a> )			

GO:0003407 : neural retina development

<https://www.ebi.ac.uk/QuickGO/term/GO:0003407>

GO:0006366 : transcription by RNA polymerase II

<https://www.ebi.ac.uk/QuickGO/term/GO:0006366>

GO:0060219 : camera-type eye photoreceptor cell differentiation

<https://www.ebi.ac.uk/QuickGO/term/GO:0060219>GO:0010842 : retina layer formation (<https://www.ebi.ac.uk/QuickGO/term/GO:0010842>)

GO - Cellular Component

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~No^#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Cis-regulatory^#gephebase-summary-title>)

Aberration Type

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

Indel Size

100-999 bp

Molecular Details of the Mutation

413bp deletion located 2.5-kb upstream of the Rx1 translation start site correlating with decreased Rx1 expression

Experimental Evidence

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

Main Reference

Interspecific variation in Rx1 expression controls opsin expression and causes visual system diversity in African cichlid fishes. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24859246>)

Authors

Schulte JE; O'Brien CS; Conte MA; O'Quin KE; Carleton KL

Abstract

The mechanisms underlying natural phenotypic diversity are key to understanding evolution and speciation. Cichlid fishes are among the most speciose vertebrates and an ideal model for identifying genes controlling species differences. Cichlids have diverse visual sensitivities that result from species expressing subsets of seven cichlid cone opsin genes. We previously identified a quantitative trait locus (QTL) that tunes visual sensitivity by varying *SWS2A* (short wavelength sensitive 2A) opsin expression in a genetic cross between two Lake Malawi cichlid species. Here, we identify Rx1 (retinal and anterior neural fold homeobox) as the causative gene for the QTL using fine mapping and RNAseq in retinal transcriptomes. Rx1 is differentially expressed between the parental species and correlated with *SWS2A* expression in the F2 progeny. Expression of Rx1 and *SWS2A* is also correlated in a panel of 16 Lake Malawi cichlid species. Association mapping in this panel identified a 413-bp deletion located 2.5-kb upstream of the Rx1 translation start site that is correlated with decreased Rx1 expression. This deletion explains 62% of the variance in *SWS2A* expression across 53 cichlid species in 29 genera. The deletion occurs in both the sand and rock-dwelling cichlid clades, suggesting that it is an ancestral polymorphism. Our finding supports the hypothesis that mixing and matching of ancestral polymorphisms can explain the diversity of present day cichlid phenotypes.

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Additional References

## RELATED GEPHE

Related Genes

1 (opsin - (*SWS2B*)) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=~143496^/and+Trait=Color+vision/or+Taxon+ID=~323801^/and+Trait=Color+vision/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

Deletion occurs in both the sand and rock-dwelling cichlid clades suggesting ILS of an ancestral polymorphism or introgressions - @Introgression @ILS