

## GEPHE SUMMARY

SLC45A2=MATP ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase= <sup>^</sup> SLC45A2=MATP <sup>^</sup> #gephebase-summary-title)	Gephebase Gene	GP00002287	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Morphology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Coloration (albinism) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> = <sup>^</sup> Coloration (albinism) <sup>^</sup> #gephebase-summary-title)	Trait		
WT	Trait State in Taxon A		
Melanin deficient fish (mutation maintained by breeders in Japan)	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status= <sup>^</sup> Domesticated <sup>^</sup> #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
	Latin Name		Latin Name
Oryzias latipes ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Oryzias latipes <sup>^</sup> #gephebase-summary-title)		Oryzias latipes ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and Synonyms= <sup>^</sup> Oryzias latipes <sup>^</sup> #gephebase-summary-title)	
	Common Name		Common Name
Japanese medaka		Japanese medaka	
	Synonyms		Synonyms
Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes		Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes	
	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; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias		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; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias	
	Parent		Parent
Oryzias () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089</a> )		Oryzias () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
8090 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090</a> )		8090 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090</a> )	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 ( <a href="http://www.uniprot.org/uniprot/Q9UMX9">http://www.uniprot.org/uniprot/Q9UMX9</a> )	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000296589 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000296589">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000296589</a> )	String		
	Sequence Similarities		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.			
	GO - Molecular Function		
GO:0008506 : sucrose:proton symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008506">https://www.ebi.ac.uk/QuickGO/term/GO:0008506</a> )			
	GO - Biological Process		

GO:0042438 : melanin biosynthetic process  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)  
 GO:0048066 : developmental pigmentation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0048066)  
 GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)  
 GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)  
 GO:0015770 : sucrose transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015770)  
 GO - Cellular Component

GO:0016021 : integral component of membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)  
 GO:0033162 : melanosome membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

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

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

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

Promoter complex change resulting in loss of expression : an inversion of 167 bp ; an insertion of 48 bp ; and a deletion of 172 bp

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

Rescue from oculocutaneous albinism type 4 using medaka slc45a2 cDNA driven by its own promoter. (2008) (https://pubmed.ncbi.nlm.nih.gov/18245373)

Fukamachi S; Kinoshita M; Tsujimura T; Shimada A; Oda S; Shima A; Meyer A; Kawamura S; Mitani H

Patients and vertebrate mutants with oculocutaneous albinism type 4 (OCA4) have mutations in the solute carrier family 45 member 2 (slc45a2) gene. However, there is no empirical evidence for this gene-phenotype relationship. There is a unique OCA4 mutant in medaka (b) that exhibits albinism only in the skin, but the mechanism underlying this phenotype is also unknown. In this study, we rescued medaka OCA4 phenotypes, in both the eyes and the skin, by micro-injection of an slc45a2-containing genomic fragment or slc45a2 cDNA driven by its own 0.9-kb promoter. We also identified a spontaneous nucleotide change of 339 bp in the promoter as the b mutation. There are multiple transcription start sites in medaka slc45a2, as in its human ortholog, and only the shortest and eye-specific mRNA is transcribed with the b mutation. Interestingly, we further revealed a conserved pyrimidine (Py)-rich sequence of approximately 10 bp in the promoter by medaka-pufferfish comparative genomics and verified that it plays an indispensable role for expression of slc45a2 in the skin. Further studies of the 0.9-kb promoter identified in this study should provide insights into the cis/trans-regulatory mechanisms underlying the ocular and cutaneous expression of slc45a2.

Presumptive Null

Molecular Type

Aberration Type

Molecular Details of the Mutation

Experimental Evidence

Main Reference

Authors

Abstract

Additional References

## RELATED GEPHE

1 (tyrosinase (TYR)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^8090^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^8090^/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^8090^#gephebase-summary-title)

Related Genes

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

@Parallelism https://omia.org/OMIA001821/8090/ @AllelicSeries