

# GEPHE SUMMARY

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

## PHENOTYPIC CHANGE

	Trait Category	
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category=^Morphology^#gephebase-summary-title)	Trait	
Coloration (albinism) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration</a> (albinism)^#gephebase-summary-title)	Trait State in Taxon A	
WT	Trait State in Taxon B	
Melanin deficient fish (mutation maintained by breeders in Japan)	Ancestral State	
Taxon A	Taxonomic Status	
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status=^Domesticated^#gephebase-summary-title)		
Taxon A	Latin Name	Latin Name
Oryzias latipes ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryzias">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Oryzias</a> latipes^#gephebase-summary-title)		
Japanese medaka	Common Name	Common Name
Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes	Synonyms	
species	Rank	Rank
	Lineage	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; Acanthomorpha; Euacanthomorphacea; Percomorphaceae; 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> )		
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> )	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
SLC45A2		
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	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: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 - Molecular Function	
	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>)

Presumptive Null

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=%20No%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%20No%23gephebase-summary-title))

Molecular Type

Cis-regulatory ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%20Cis-regulatory%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%20Cis-regulatory%23gephebase-summary-title))

Aberration Type

Complex Change ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%20Complex Change%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%20Complex%20Change%23gephebase-summary-title))

Molecular Details of the Mutation

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

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%20Candidate Gene%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%20Candidate%20Gene%23gephebase-summary-title))

Main Reference

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

Authors

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Abstract

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.

Additional References

## RELATED GEPHE

1 (tyrosinase (TYR)) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=%208090%20/and+Trait=Coloration/and+groupHaplotypes=true%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon%20ID=%208090%20/and+Trait=Coloration/and+groupHaplotypes=true%23gephebase-summary-title))

Related Genes

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=%20SLC45A2=MATP%20/and+Taxon ID=%208090%20/or+Gene Gephebase=%20SLC45A2=MATP%20/and+Taxon ID=%208090%23gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene%20Gephebase=%20SLC45A2=MATP%20/and+Taxon%20ID=%208090%20/or+Gene%20Gephebase=%20SLC45A2=MATP%20/and+Taxon%20ID=%208090%23gephebase-summary-title))

## EXTERNAL LINKS

## COMMENTS

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