

GEPHE SUMMARY

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] SLC45A2=MATP [^] #gephebase-summary-title)	Gephebase Gene	GP00002286	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Morphology [^] #gephebase-summary-title)	Trait Category		
Coloration (albinism) (https://www.gephebase.org/search-criteria?/and+Trait = [^] Coloration (albinism) [^] #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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Domesticated [^] #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
	Latin Name		Latin Name
Oryzias latipes (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Oryzias latipes [^] #gephebase-summary-title)	Latin Name	Oryzias latipes (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Oryzias latipes [^] #gephebase-summary-title)	Latin Name
Japanese medaka	Common Name	Japanese medaka	Common Name
Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes	Synonyms	Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percormorphaceae; Ovalentaria; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Percormorphaceae; Ovalentaria; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias	Lineage
Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089)	Parent	Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089)	Parent
8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090)	NCBI Taxonomy ID	8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

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

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

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

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

100-999 bp

tandem insertion of 195 nt identical to the adjacent ORF sequence. This tandem repeat produces an insertion of 65 amino acids

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

Mutations in the gene encoding B, a novel transporter protein, reduce melanin content in medaka. (2001) (https://pubmed.ncbi.nlm.nih.gov/11479596)

Fukamachi S; Shimada A; Shima A

Pigmentation of the skin is of great social, clinical and cosmetic significance. Several genes that, when mutated, give rise to altered coat color in mice have been identified; their analysis has provided some insight into melanogenesis and human pigmentation diseases. Such analyses do not, however, fully inform on the pigmentation of lower vertebrates because mammals have only one kind of chromatophore, the melanocyte. In contrast, the medaka (a small, freshwater teleost) is a suitable model of the lower vertebrates because it has all kinds of chromatophores. The basic molecular genetics of fish are known and approximately 70 spontaneous pigmentation mutants have been isolated. One of these, an orange-red variant, is a homozygote of a well-known and common allele, b, and has been bred for hundreds of years by the Japanese. Here, we report the first successful positional cloning of a medaka gene (AIM1): one that encodes a transporter that mediates melanin synthesis. The protein is predicted to consist of 12 transmembrane domains and is 55% identical to a human EST of unknown function isolated from melanocytes and melanoma cells. We also isolated a highly homologous gene from the mouse, indicating a conserved function of vertebrate melanogenesis. Intriguingly, these proteins have sequence and structural similarities to plant sucrose transporters, suggesting a relevance of sucrose in melanin synthesis. Analysis of AIM1 orthologs should provide new insights into the regulation of melanogenesis in both teleosts and mammals.

Presumptive Null

Molecular Type

Aberration Type

Insertion Size

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 ; another mutant allele called b seems to be due to cis-regulatory loss of SLC45A2 expression