GEPHE SUMMARY

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

GepheID
GP00002286

Entry Status
Published

Main curator
Martin

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
WT

Trait State in Taxon B
Melanin deficient fish (mutation maintained by breeders in Japan)

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Oryzias latipes (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryzias latipes^#gephebase-summary-title)

Common Name
Japanese medaka

Synonyms
Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes

Rank
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; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias

Parent
Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8089)

NCBI Taxonomy ID
8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8090)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Oryzias latipes (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Oryzias latipes^#gephebase-summary-title)

Common Name
Japanese medaka

Synonyms
Poecilia latipes; Japanese medaka; Japanese rice fish; medaka; Oryzias latipes (Temminck & Schlegel, 1846); Poecilia latipes Temminck & Schlegel, 1846; Orizias latipes

Rank
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; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias

Parent
Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8089)

NCBI Taxonomy ID
8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8090)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)

GO - Biological Process

UniProtKB Homo sapiens
Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)

GenebankID or UniProtKB
()

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=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Insertion Size

100-999 bp

Molecular Details of the Mutation

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

Experimental Evidence

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

Main Reference

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

Authors

Fukamachi S; Shimada A; Shima A

Abstract

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.

Additional References

RELATED GEPHE

Related Genes

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

Related Haplotypes

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)

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