GEPHE SUMMARY

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

GepheID
GP00002287

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

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

Aberration Type

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

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

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

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