

GEPHE SUMMARY

	Gephebase Gene		GepheID
tyrosinase (TYR) (<a +tyrosinase+(tyr)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+tyrosinase+(TYR)^#gephebase-summary-title)		GP00002372	
	Entry Status	Santos	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology (<a +morphology^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology^#gephebase-summary-title)			
	Trait		
Coloration (amelanism) (<a +coloration+(amelanism)^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(amelanism)^#gephebase-summary-title)			
	Trait State in Taxon A		
WT			
	Trait State in Taxon B		
reduction of melanic pigmentation having pale orange-red skin but with red-wine-colored eyes			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Domesticated (<a +domesticated^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated^#gephebase-summary-title)			
	Taxon A	Taxon B	
	Latin Name		Latin Name
Oryzias latipes (<a +oryzias+latipes^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryzias+latipes^#gephebase-summary-title)		Oryzias latipes (<a +oryzias+latipes^#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Oryzias+latipes^#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; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; 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; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; Ovalentaria; Atherinomorphae; Beloniformes; Adrianichthyoidei; Adrianichthyidae; Oryziinae; Oryzias	
	Parent		Parent
Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089)		Oryzias () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8089)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090)		8090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=8090)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
Yes		Yes	
	Taxon A Description		Taxon B Description
WT strain		genotype i4/i4	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Mus musculus
Tyr		P11344 (http://www.uniprot.org/uniprot/P11344)	
	Synonyms		GenebankID or UniProtKB
c; Oca1; skc35; albino		P55025 (https://www.ncbi.nlm.nih.gov/nuccore/P55025)	
	String		
10090.ENSMUSP00000004770 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770)			
	Sequence Similarities		
Belongs to the tyrosinase family.			
	GO - Molecular Function		
GO:0042803 : protein homodimerization activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
GO:0046982 : protein heterodimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046982>)
GO:0005507 : copper ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005507>)
GO:0004503 : monophenol monooxygenase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004503>)

GO - Biological Process

GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0033280 : response to vitamin D (<https://www.ebi.ac.uk/QuickGO/term/GO:0033280>)
GO:0051591 : response to cAMP (<https://www.ebi.ac.uk/QuickGO/term/GO:0051591>)
GO:0009411 : response to UV (<https://www.ebi.ac.uk/QuickGO/term/GO:0009411>)
GO:0048538 : thymus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048538>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0043231 : intracellular membrane-bounded organelle
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)
GO:0048471 : perinuclear region of cytoplasm
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)
GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)
GO:0033162 : melanosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

Presumptive Null

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes~#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

1-10 kb

Molecular Details of the Mutation

"i4 allele was found to contain a 4.7- kb transposable element in the 5th exon"

"Tyr-i4 contains a 4.7-kb insertion in exon 5 (Fig. 3). If the inserted fragment was transcribed, the amino acid sequence would be altered after the 469th residue, with a termination codon (UAG) at amino acid position 471. The i4/i4 genotype exhibits a quasi-albino phenotype. This may be explained by generation of a tyrosinase protein that only lacks its C-terminus region. The wild-type tyrosinase has a highly hydrophobic amino acid block at its C terminus (aa 480 - aa 500) which is thought to be part of a membrane spanning domain (Inagaki et al., 1994). A possible explanation is that the mutant tyrosinase encoded by Tyr- i4 lacks this region, so that it cannot be positioned properly on the membrane"

Experimental Evidence

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

Main Reference

Albinism due to transposable element insertion in fish. (1997) (<https://pubmed.ncbi.nlm.nih.gov/9428004>)

Authors

Koga A; Hori H

Abstract

The i locus of the medaka fish, *Oryzias latipes*, is responsible for tyrosinase expression, and several mutant alleles have been identified. The genotype i1/i1 exhibits a complete albino phenotype, having pale orange-red skin and red eyes. This mutant lacks in vivo tyrosinase activity. The genotype i4/i4, on the other hand, shows a quasi-albino phenotype with skin as bright as that of i1/i1 but with red-wine-colored eyes. At the light microscope level, reduced pigmentation is observed both in the skin and eyes of this mutant. The tyrosinase genes for the i1 and the i4 alleles were cloned and sequenced, and compared with that of the wild-type tyrosinase gene. The i1 allele was found to contain a 1.9-kb transposable element in the 1st exon, and the i4 allele was found to contain a 4.7-kb transposable element in the 5th exon. Both i1 and i4 are alleles that were found in a commercial breeding population. The insertion of a transposable element thus appears to constitute a natural cause of mutations that cause albinism in this organism.

Additional References

RELATED GEPHE

Related Genes

1 (SLC45A2=MATP) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=~8090~/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

