## GEPHE SUMMARY

Gephebase Gene

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

Entry Status

Published

GepheID

GP00002321

Main curator

Martin

## PHENOTYPIC CHANGE

Trait Category

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait

Coloration (skin) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (skin)^#gephebase-summary-title)

Trait State in Taxon A

WT

Trait State in Taxon B

Complete albinism

Ancestral State

Taxon A

Taxonomic Status

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

| | Taxon A | Taxon B |
|---|---|---|
| Latin Name | Fejervarya kawamurai (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Fejervarya kawamurai^#gephebase-summary-title) | Fejervarya kawamurai (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Fejervarya kawamurai^#gephebase-summary-title) |
| Common Name | - | - |
| Synonyms | Fejervarya kawamurai Djong, Matsui, Kuramoto, Nishioka & Sumida, 2011; IABHU F2184 | Fejervarya kawamurai Djong, Matsui, Kuramoto, Nishioka & Sumida, 2011; IABHU F2184 |
| Rank | species | species |
| Lineage | cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Anura; Neobatrachia; Ranoidea; Dicroglossidae; Dicroglossinae; Fejervarya | cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amphibia; Batrachia; Anura; Neobatrachia; Ranoidea; Dicroglossidae; Dicroglossinae; Fejervarya |
| Parent | Fejervarya () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 127010 ) | Fejervarya () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 127010 ) |
| NCBI Taxonomy ID | 1772025 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 1772025 ) | 1772025 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 1772025 ) |
| is Taxon A an Infraspecies? / is Taxon B an Infraspecies? | No | No |

## GENOTYPIC CHANGE

Generic Gene Name

Tyr

Synonyms

c; Oca1; skc35; albino

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)

UniProtKB Mus musculus

P11344 (http://www.uniprot.org/uniprot/P11344)

GenebankID or UniProtKB

()

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

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

c.169G>A p.Gly57Arg

Experimental Evidence

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

| | Taxon A | Taxon B | Position |
|---|---|---|---|
| Codon | - | - | - |
| Amino-acid | Gly | Arg | 57 |

Main Reference

Spontaneous tyrosinase mutations identified in albinos of three wild frog species. (2018) (https://pubmed.ncbi.nlm.nih.gov/28674275)

Authors

Miura I; Tagami M; Fujitani T; Ogata M

Abstract

The present study reports spontaneous tyrosinase gene mutations identified in oculocutaneous albinos of three Japanese wild frog species, Pelophylax nigromaculatus, Glandirana rugosa and Fejervarya kawamurai. This represents the first molecular analyses of albinic phenotypes in frogs. Albinos of P. nigromaculatus collected from two different populations were found to suffer from frameshift mutations. These mutations were caused by the insertion of a thymine residue within each of exons 1 and 4, while albinos in a third population lacked three nucleotides encoding lysine in exon 1. Albinos from the former two P. nigromaculatus populations were also associated with splicing variants of mRNA that lacked either exons 2-4 or exon 4. In the other two frog species examined, missense mutations that resulted in amino acid substitutions from glycine to arginine and glycine to aspartic acid were identified in exons 1 and 3, respectively. The two glycines in F. kawamurai and G. rugosa, and the lysine deleted in one P. nigromaculatus albino, were highly conserved in vertebrates, which suggested that they were situated in regions of critical importance to tyrosinase function. In fact, the glycine of G. rugosa is located within a predicted copper-binding domain. The five mutations identified in the present study are candidates for causing the albinic phenotypes, and, if directly confirmed, they are all unique among vertebrates, which suggests that molecular analysis of albino frogs could contribute to research on albinos in humans and vertebrates by providing new information about tyrosinase structure and transcript processing.

Additional References

## RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS