

## GEPHE SUMMARY

tyrosinase (TYR) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> )	Gephebase Gene	GP00002321	GepheID
Gephebase="tyrosinase (TYR)"#gephebase-summary-title)			Main curator
Published	Entry Status	Martin	

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> )	Trait Category		
Category="Morphology"#gephebase-summary-title)			
Coloration (skin) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> )	Trait		
(skin)"#gephebase-summary-title)			
WT	Trait State in Taxon A		
Complete albinism	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> )	Taxonomic Status		
Status="Intraspecific"#gephebase-summary-title)			

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

## GENOTYPIC CHANGE

Tyr	Generic Gene Name	P11344 ( <a href="http://www.uniprot.org/uniprot/P11344">http://www.uniprot.org/uniprot/P11344</a> )	UniProtKB Mus musculus
c; Oca1; skc35; albino	Synonyms		GenebankID or UniProtKB
	String		
10090.ENSMUSP00000004770			
( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770</a> )			
Belongs to the tyrosinase family.	Sequence Similarities		
	GO - Molecular Function		
GO:0042803 : protein homodimerization activity			
( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042803">https://www.ebi.ac.uk/QuickGO/term/GO:0042803</a> )			
GO:0046982 : protein heterodimerization activity			
( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046982">https://www.ebi.ac.uk/QuickGO/term/GO:0046982</a> )			
GO:0005507 : copper ion binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005507">https://www.ebi.ac.uk/QuickGO/term/GO:0005507</a> )			
GO:0004503 : monophenol monooxygenase activity			
( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004503">https://www.ebi.ac.uk/QuickGO/term/GO:0004503</a> )			

- 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>)

- No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>) Presumptive Null
- Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title>) Molecular Type
- SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>) Aberration Type
- Nonsynonymous SNP Coding Change
- c.169G>A p.Gly57Arg Molecular Details of the Mutation
- Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title>) Experimental Evidence

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

- Spontaneous tyrosinase mutations identified in albinos of three wild frog species. (2018) (<https://pubmed.ncbi.nlm.nih.gov/28674275>) Main Reference
- Miura I; Tagami M; Fujitani T; Ogata M Authors
- 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. Abstract
- Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

