GEPHE SUMMARY

Gephebase Gene

SCN4A (Nav1.4b gene copy) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SCN4A (Nav1.4b gene copy)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000733

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Other fishes

Trait State in Taxon B

Tetraodon nigroviridis

Ancestral State

Data not curated

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

teleost fishes

Synonyms

teleost fishes

Rank

infraclass

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii

Parent

Neopterygii () - (Rank: subclass) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 41665)

NCBI Taxonomy ID

32443 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32443)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Tetraodon nigroviridis (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Tetraodon nigroviridis^#gephebase-summary-title)

Common Name

spotted green pufferfish

Synonyms

spotted green pufferfish; Tetraodon nigroviridis Marion de Proce, 1822

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; Eupercaria; Tetraodontiformes; Tetraodontoidei; Tetradontoidea; Tetraodontidae; Tetraodon

Parent

Tetraodon () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 47144)

NCBI Taxonomy ID

99883 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 99883)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

SCN4A

Synonyms

HYPP; SkM1; CMS16; HYKPP; NAC1A; HOKPP2; Nav1.4; Na(V)1.4

String

9606.ENSP00000396320 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000396320)

Sequence Similarities

Belongs to the sodium channel (TC 1.A.1.10) family. Nav1.4/SCN4A subfamily.

GO - Molecular Function

GO:0005244 : voltage-gated ion channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005244)
GO:0005248 : voltage-gated sodium channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005248)

GO - Biological Process

UniProtKB Homo sapiens

P35499 (http://www.uniprot.org/uniprot/P35499)

GenebankID or UniProtKB

ABB29442 (https://www.ncbi.nlm.nih.gov/nuccore/ABB29442)

GO:0006814 : sodium ion transport (https://www.ebi.ac.uk/QuickGO/term/GO:0006814)
GO:0019228 : neuronal action potential
(https://www.ebi.ac.uk/QuickGO/term/GO:0019228)
GO:0034765 : regulation of ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0034765)
GO:0086010 : membrane depolarization during action potential
(https://www.ebi.ac.uk/QuickGO/term/GO:0086010)
GO:0006936 : muscle contraction (https://www.ebi.ac.uk/QuickGO/term/GO:0006936)
GO:0035725 : sodium ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0035725)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0030424 : axon (https://www.ebi.ac.uk/QuickGO/term/GO:0030424)
GO:0001518 : voltage-gated sodium channel complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0001518)

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

E945D

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	-	-	-

Main Reference

Genetic basis of tetrodotoxin resistance in pufferfishes. (2005) (https://pubmed.ncbi.nlm.nih.gov/16303569)

Authors

Venkatesh B; Lu SQ; Dandona N; See SL; Brenner S; Soong TW

Abstract

Tetrodotoxin (TTX) is a highly potent neurotoxin that selectively binds to the outer vestibule of voltage-gated sodium channels. Pufferfishes accumulate extremely high concentrations of TTX without any adverse effect. A nonaromatic amino acid (Asn) residue present in domain I of the pufferfish, Takifugu pardalis, Na v1.4 channel has been implicated in the TTX resistance of pufferfishes . However, the effect of this residue on TTX sensitivity has not been investigated, and it is not known if this residue is conserved in all pufferfishes. We have investigated the genetic basis of TTX resistance in pufferfishes by comparing the sodium channels from two pufferfishes (Takifugu rubripes [fugu] and Tetraodon nigroviridis) and the TTX-sensitive zebrafish. Although all three fishes contain duplicate copies of Na v1.4 channels (Na v1.4a and Na v1.4b), several substitutions were found in the TTX binding outer vestibule of the two pufferfish channels. Electrophysiological studies showed that the nonaromatic residue (Asn in fugu and Cys in Tetraodon) in domain I of Na v1.4a channels confers TTX resistance. The Glu-to-Asp mutation in domain II of Tetraodon channel Na v1.4b is similar to that in the saxitoxin- and TTX-resistant Na+ channels of softshell clams . Besides helping to deter predators, TTX resistance enables pufferfishes to selectively feed on TTX-bearing organisms.

Additional References

Toxin-resistant sodium channels: parallel adaptive evolution across a complete gene family. (2008) (https://pubmed.ncbi.nlm.nih.gov/18258611)

RELATED GEPHE

Related Genes

5 (AHR2, AIP, ARNT-1c, ARNT-L2a, SCN4A (Nav1.4a gene copy)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^32443^/and+Trait=Xenobiotic resistance/or+Taxon ID=^99883^/and+Trait=Xenobiotic resistance/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SCN4A (Nav1.4b gene copy)^/and+Taxon ID=^32443^/or+Gene Gephebase=^SCN4A (Nav1.4b gene copy)^/and+Taxon ID=^99883^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS