

SCN4A (Nav1.4a gene copy) (https://www.gephebase.org/search-criteria?/and+Gene)	Gephebase Gene	GP00000722	GepheID
Gephebase=~SCN4A (Nav1.4a gene copy)~#gephebase-summary-title)			Main curator
Published	Entry Status	Martin	

	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
Takifugu rubripes	
	Ancestral State
Data not curated	
	Taxonomic Status
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Interspecific^#gephebase-summary-title)	

GENOTYPIC CHANGE

SCN4A	Generic Gene Name	P35499 (http://www.uniprot.org/uniprot/P35499)	UniProtKB Homo sapiens
HYPP; SkM1; CMS16; HYKPP; NAC1A; HOKPP2; Nav1.4; Na(V)1.4	Synonyms	ABB29442 (https://www.ncbi.nlm.nih.gov/nuccore/ABB29442)	GenebankID or UniProtKB
9606.ENSP00000396320 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000396320)	String		
	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

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>)

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
Y401N	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	-	-	-

Genetic basis of tetrodotoxin resistance in pufferfishes. (2005) (https://pubmed.ncbi.nlm.nih.gov/16303569)	Main Reference
Venkatesh B; Lu SQ; Dandona N; See SL; Brenner S; Soong TW	Authors
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.	Abstract
	Additional References

RELATED GEPHE

5 (AHR2, AIP, ARNT-1c, ARNT-L2a, SCN4A (Nav1.4b gene copy)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~32443~/and+Trait=Xenobiotic resistance/or+Taxon ID=~31033~/and+Trait=Xenobiotic resistance/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~SCN4A (Nav1.4a gene copy)~/and+Taxon ID=~32443~/or+Gene Gephebase=~SCN4A (Nav1.4a gene copy)~/and+Taxon ID=~31033~#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

