

## GEPHE SUMMARY

	Gephebase Gene	GepheID
SCN4A (Nav1.4) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^SCN4A (Nav1.4)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^SCN4A (Nav1.4)^#gephebase-summary-title</a> )	GP00000721	
Published	Entry Status	Main curator

## PHENOTYPIC CHANGE

	Trait Category	Trait	
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title</a> )			
Xenobiotic resistance (TTX) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Xenobiotic resistance (TTX)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Xenobiotic resistance (TTX)^#gephebase-summary-title</a> )	Trait State in Taxon A	Trait State in Taxon B	Ancestral State
Mya arenaria - sensitive			
Mya arenaria - resistant			
Taxon A	Taxonomic Status		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title</a> )			
Taxon A	Latin Name	Taxon B	Latin Name
Mya arenaria ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mya+arenaria^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mya+arenaria^#gephebase-summary-title</a> )		Mya arenaria ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mya+arenaria^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mya+arenaria^#gephebase-summary-title</a> )	
-	Common Name	-	Common Name
softshell	Synonyms	softshell	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheredonta; Myoidea; Myoidea; Myidae; Mya	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheredonta; Myoidea; Myoidea; Myidae; Mya	Lineage
Mya () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603</a> )	Parent	Mya () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603</a> )	Parent
6604 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604</a> )	NCBI Taxonomy ID	6604 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604</a> )	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

## GENOTYPIC CHANGE

	Generic Gene Name		
SCN4A			UniProtKB Homo sapiens
HYPP; SkM1; CMS16; HYKPP; NAC1A; HOKPP2; Nav1.4; Na(V)1.4	Synonyms		GenebankID or UniProtKB
9606.ENSP00000396320 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000396320">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000396320</a> )	String		AAX14719 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AAX14719">https://www.ncbi.nlm.nih.gov/nuccore/AAX14719</a> )
	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 ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005244">https://www.ebi.ac.uk/QuickGO/term/GO:0005244</a> )			
GO:0005248 : voltage-gated sodium channel activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005248">https://www.ebi.ac.uk/QuickGO/term/GO:0005248</a> )			
GO:0005244 : voltage-gated ion channel activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005244">https://www.ebi.ac.uk/QuickGO/term/GO:0005244</a> )	GO - Biological Process		
GO:0006814 : sodium ion transport ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0006814">https://www.ebi.ac.uk/QuickGO/term/GO:0006814</a> )			
GO:0019228 : neuronal action potential ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0019228">https://www.ebi.ac.uk/QuickGO/term/GO:0019228</a> )			

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

Sodium channel mutation leading to saxitoxin resistance in clams increases risk of PSP. (2005) (<https://pubmed.ncbi.nlm.nih.gov/15815630>)

Authors

Bricelj VM; Connell L; Konoki K; Macquarrie SP; Scheuer T; Catterall WA; Trainer VL

Abstract

Bivalve molluscs, the primary vectors of paralytic shellfish poisoning (PSP) in humans, show marked inter-species variation in their capacity to accumulate PSP toxins (PSTs) which has a neural basis. PSTs cause human fatalities by blocking sodium conductance in nerve fibres. Here we identify a molecular basis for inter-population variation in PSP resistance within a species, consistent with genetic adaptation to PSTs. Softshell clams (*Mya arenaria*) from areas exposed to 'red tides' are more resistant to PSTs, as demonstrated by whole-nerve assays, and accumulate toxins at greater rates than sensitive clams from unexposed areas. PSTs lead to selective mortality of sensitive clams. Resistance is caused by natural mutation of a single amino acid residue, which causes a 1,000-fold decrease in affinity at the saxitoxin-binding site in the sodium channel pore of resistant, but not sensitive, clams. Thus PSTs might act as potent natural selection agents, leading to greater toxin resistance in clam populations and increased risk of PSP in humans. Furthermore, global expansion of PSP to previously unaffected coastal areas might result in long-term changes to communities and ecosystems.

Additional References

## RELATED GEPHE

#### Related Genes

No matches found.

Related Haplotypes

No matches found.

## EXTERNAL LINKS

## COMMENTS

