

## GEPHE SUMMARY

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

## PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Physiology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Xenobiotic resistance (TTX) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=&lt;sup&gt;^&lt;/sup&gt;Xenobiotic resistance (TTX)&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Xenobiotic resistance (TTX)<sup>^</sup>#gephebase-summary-title</a> )	Trait		
Mya arenaria - sensitive	Trait State in Taxon A		
Mya arenaria - resistant	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> Status= <sup>^</sup> Intraspecific <sup>^</sup> #gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Mya arenaria ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Mya arenaria&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Mya arenaria<sup>^</sup>#gephebase-summary-title</a> )	Latin Name	Mya arenaria ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Mya arenaria&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Mya arenaria<sup>^</sup>#gephebase-summary-title</a> )	Latin Name
-	Common Name	-	Common Name
softshell	Synonyms	softshell	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheterodonta; Myoidea; Myoidea; Myidae; Mya	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheterodonta; 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 Intraspecies?	No	is Taxon B an Intraspecies?

## GENOTYPIC CHANGE

SCN4A	Generic Gene Name	P35499 ( <a href="http://www.uniprot.org/uniprot/P35499">http://www.uniprot.org/uniprot/P35499</a> )	UniProtKB Homo sapiens
HYPP; SkM1; CMS16; HYKPP; NAC1A; HOKPP2; Nav1.4; Na(V)1.4	Synonyms	AAX14719 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AAX14719">https://www.ncbi.nlm.nih.gov/nuccore/AAX14719</a> )	GenebankID or UniProtKB
9606.ENSPP00000396320 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000396320">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000396320</a> )	String		
Belongs to the sodium channel (TC 1.A.1.10) family. Nav1.4/SCN4A subfamily.	Sequence Similarities		
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 - Molecular Function		
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: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 - Biological Process		
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

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

