GEPHE SUMMARY

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

GepheID
GP00000721

Entry Status
Published

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
Mya arenaria - sensitive

Trait State in Taxon B
Mya arenaria - resistant

Ancestral State
Taxon A

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

Taxon A

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

Common Name
-

Synonyms
softshell

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheterodonta; Myoida; Myoidea; Myidae; Mya

Parent
Mya () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603)

NCBI Taxonomy ID
6604 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
-

Synonyms
softshell

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Lophotrochozoa; Mollusca; Bivalvia; Heterodonta; Euheterodonta; Myoida; Myoidea; Myidae; Mya

Parent
Mya () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6603)

NCBI Taxonomy ID
6604 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6604)

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
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)

UniProtKB Homo sapiens
P35499 (http://www.uniprot.org/uniprot/P35499)

GenebankID or UniProtKB
AAX14719 (https://www.ncbi.nlm.nih.gov/nuccore/AAX14719)

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