Resource Summary Report

Generated by SPARC SAWG on Aug 9, 2026

scn1a

RRID:SCR_006987
Type: Tool

Proper Citation

scn1a (RRID:SCR_006987)

Resource Information

URL: http://scicrunch.org

Proper Citation: scn1a (RRID:SCR_006987)

Description: THIS RESOURCE IS NO LONGER IN SERVICE, documented on August 29,2019.

Locus-specific database listing variants of the SCN1A gene. To provide a continued platform that is in keeping with the latest data, this web site has been set up where all information can be accessed and downloaded. A Mutation submission form encourages data submissions. Over the past ten years, mutations in voltage-gated sodium channels (Navs) have become closely associated with inheritable forms of epilepsy. One isoform in particular, Nav1.1 (gene symbol SCN1A), appears to be a superculprit, with virtually hundreds of mutations. The associated phenotypes range from benign febrile seizures to extremely serious conditions like Dravet syndrome (a.k.a. severe myoclonic epilepsy in infancy or SMEI). Despite the wealth of information, mutational analyses are cumbersome, owing to inconsistencies among the Nav1.1 sequences to which different research groups refer. Splicing variability is the core problem: Nav1.1 exists in the brain in 3 different isoforms: full-length (2009 AA) as well as two shorter versions that lack 11 or 28 amino acids compared to the former. This online database ?????? SCN1A infobase ?????? establishes a standardized nomenclature for Nav1.1 variants so as to provide a platform from which future mutation analyses can be started without up-front data normalization.

Abbreviations: SCN1A Infobase

Synonyms: The SCN1A Infobase

Resource Type: data or information resource, database, service resource, storage service resource, data repository

Defining Citation: PMID:18804930

Keywords: scn1a, nav1.1, variation, epilepsy, dravet, smei, gefs, locus-specific, mutation, voltage-gated sodium channel, seizure, sodium channel, scn1a mutation

Funding: private endeavor

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: scn1a

Resource ID: SCR_006987

Alternate IDs: nlx_143761

Old URLs: http://web.scn1a.info, http://www.scn1a.info

Record Creation Time: 20220129T080239+0000

Record Last Update: 20260808T185852+0000

Ratings and Alerts

No rating or validation information has been found for scn1a.

No alerts have been found for scn1a.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 25 mentions in open access literature.

Listed below are recent publications. The full list is available at SPARC SAWG .

Bao Z, et al. (2024) Effect of SCN1Aand SCN2A gene polymorphisms on the efficacy of valproic acid treatment in Chinese children with epilepsy. PloS one, 19(6), e0304869.

Fidry M, et al. (2024) Pharmacogenetic Testing in Treatment-resistant Panic Disorder: a Preliminary Analysis. Clinical practice and epidemiology in mental health : CP & EMH, 20, e17450179337258.

Balestrini S, et al. (2023) A registry for Dravet syndrome: The Italian experience. Epilepsia open, 8(2), 517.

Alam A, et al. (2022) Targeted Whole Exome Sequencing in Children With Early-Onset Epilepsy: Parent Experiences. Journal of child neurology, 37(10-11), 840.

Petrasek T, et al. (2021) mTOR inhibitor improves autistic-like behaviors related to Tsc2 haploinsufficiency but not following developmental status epilepticus. Journal of neurodevelopmental disorders, 13(1), 14.

Ozyurt IB, et al. (2020) Bio-AnswerFinder: a system to find answers to questions from biomedical texts. Database : the journal of biological databases and curation, 2020.

Piccolo SR, et al. (2020) ShinyLearner: A containerized benchmarking tool for machine-learning classification of tabular data. GigaScience, 9(4).

Katoozi S, et al. (2020) Functional specialization of retinal M??ller cell endfeet depends on an interplay between two syntrophin isoforms. Molecular brain, 13(1), 40.

Anwar MZ, et al. (2019) To assemble or not to resemble-A validated Comparative Metatranscriptomics Workflow (CoMW). GigaScience, 8(8).

Shi HS, et al. (2019) Ca2+-dependent recruitment of voltage-gated sodium channels underlies bilirubin-induced overexcitation and neurotoxicity. Cell death & disease, 10(10), 774.

Nazish HR, et al. (2018) The possible effect of SCN1A and SCN2A genetic variants on carbamazepine response among Khyber Pakhtunkhwa epileptic patients, Pakistan. Therapeutics and clinical risk management, 14, 2305.

Sourbron J, et al. (2017) Pharmacological Analysis of the Anti-epileptic Mechanisms of Fenfluramine in scn1a Mutant Zebrafish. Frontiers in pharmacology, 8, 191.

Do TT, et al. (2017) SCN1A Gene Mutation and Adaptive Functioning in 18 Vietnamese Children with Dravet Syndrome. Journal of clinical neurology (Seoul, Korea), 13(1), 62.

Ogden KK, et al. (2016) Prioritizing the development of mouse models for childhood brain disorders. Neuropharmacology, 100, 2.

Wong JC, et al. (2016) Huperzine A Provides Robust and Sustained Protection against Induced Seizures in Scn1a Mutant Mice. Frontiers in pharmacology, 7, 357.

Sun Y, et al. (2016) A deleterious Nav1.1 mutation selectively impairs telencephalic inhibitory neurons derived from Dravet Syndrome patients. eLife, 5.

Bechi G, et al. (2015) Rescuable folding defective NaV1.1 (SCN1A) mutants in epilepsy: properties, occurrence, and novel rescuing strategy with peptides targeted to the endoplasmic reticulum. Neurobiology of disease, 75, 100.

Haerian BS, et al. (2015) RORA gene rs12912233 and rs880626 polymorphisms and their interaction with SCN1A rs3812718 in the risk of epilepsy: a case-control study in Malaysia. Genomics, 105(4), 229.

Xu X, et al. (2015) Amplicon Resequencing Identified Parental Mosaicism for Approximately 10% of "de novo" SCN1A Mutations in Children with Dravet Syndrome. Human mutation, 36(9), 861.

Lossin C, et al. (2012) Compromised function in the Na(v)1.2 Dravet syndrome mutation R1312T. Neurobiology of disease, 47(3), 378.