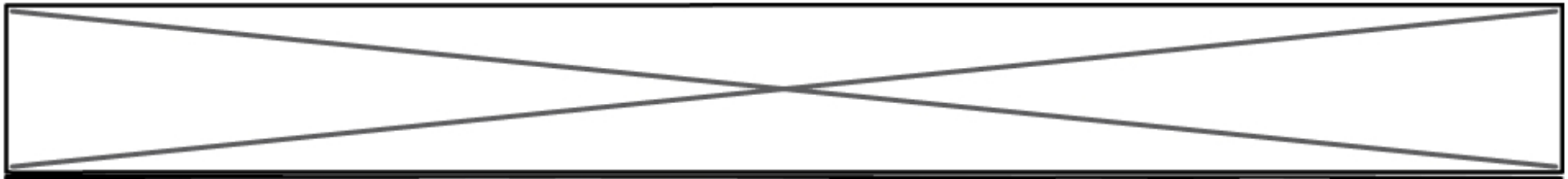
A Web Page

http://

Epilepsiome

search

A database on epilepsy and genes developed by the Genetics Commission of the ILAE



Home | About Epilepsiome | About the GC | About the ILAE | Our blog | EpiGAD | SiteMap

By disease

The genetic epilepsies explained by disease

- Dravet Syndrome
- West Syndrome
- Genetic Generalized Epilepsies
- Benign Neonatal and Infantile Seizures

...additional diseases

most relevant terms in 2014: Dravet Syndrome, Absence Epilepsy, Juvenile Myoclonic Epilepsy, Lennox-Gastaut-Syndrome, West Syndrome, Infantile Spasms

By genes

The genes implicated in human seizure disorders

- *SCN1A*
- *CDKL5*
- *PCDH19*
- *STXBP1*

...additional genes

most relevant terms in 2014: SCN2A, LGI1, STXBP1, SCN9A, CHD2, ALG13, HDAC1, PCDH19, HCN1, PGAP2, ME2, EFHC1, BRD2, ARX, LIS1

By mechanism

Theories on how genes link to epilepsy

- Channelopathies
- Neuronal Migration
- De novo mutations
- Pharmacoresistance

...additional mechanisms

most relevant terms in 2014: de novo mutations, copy number variations, CNV, recessive disorders, GEFS+, genotype-phenotype correlation, brain malformations

By concept

Terms and concepts that you might want to know

- Genome-wide association study
- Exome sequencing
- Epigenetics explained
- Variable expressivity and penetrance

...additional list of relevant terms

most relevant terms: GWAS, exome sequencing, de novo mutations, copy number variations, microdeletions, genome sequencing

Recent blog posts

- *HCN1* in Dravet Syndrome
- *FIG4* in polymicrogyria
- *QARS* in EE
- *PNPO* and neonatal epilepsy

Receive most recent updates on epilepsies together with xxx other people

Most recent updates

- Gene A and disease B
- Gene B and disease C
- Gene C and disease D
- Gene D and disease E
- Gene E and disease F

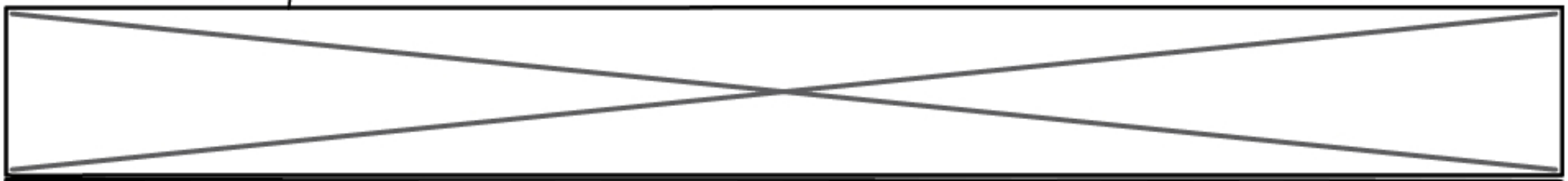
Beyond the Ion channel

http://channelopathist.net

Epilepsiome

A database on epilepsy and genes by the Genetics Commission of the ILAE

search



Home | About Epilepsiome | About the GC | About the ILAE | Our blog

By disease | By genes | By mechanism | By concept

Featured syndrome of the month

XY Syndrome is a severe epilepsy of Infancy that usually present in othewise healthy children. Various genes have been implicated in the etiology of XY, but solid evidence for a contribution of the SCNXA gene could only be obtained in 2013...
...read more

Epilepsy syndromes A-Z

Diseases and Syndromes - A -

Disease A	Disease A	Disease A
Disease B	Disease B	Disease B
Disease C	Disease C	Disease C
Disease D	Disease D	Disease D
Disease E	Disease E	Disease E

Diseases and Syndromes - B -

Disease A	Disease A	Disease A
Disease B	Disease B	Disease B
Disease C	Disease C	Disease C
Disease D	Disease D	Disease D
Disease E	Disease E	Disease E

Diseases and Syndromes - C -

Disease A	Disease A	Disease A
Disease B	Disease B	Disease B
Disease C	Disease C	Disease C
Disease D	Disease D	Disease D
Disease E	Disease E	Disease E

Diseases and Syndromes - E -

Disease A	Disease A	Disease A
Disease B	Disease B	Disease B
Disease C	Disease C	Disease C
Disease D	Disease D	Disease D
Disease E	Disease E	Disease E

Beyond the Ion channel

http://channelopathist.net

Epilepsiome

A database on epilepsy and genes by the Genetics Commission of the ILAE

search

Home | About Epilepsiome | About the GC | About the ILAE | Our blog | EpiGAD

By disease | By genes | By mechanism | By concept

Featured gene of the month

***SCN1A* - alpha 1 subunit of the voltage-gated sodium channel**

SCN1A is the most prominent epilepsy gene to date, causing a range of epilepsy syndromes. Most prominently, mutations in *SCN1A* are the genetic cause of Dravet Syndrome, formerly Severe Myoclonic Epilepsy of Infancy. In addition, *SCN1A* is implicated in the familial syndrome of GEFS+, Genetic/Generalized Epilepsy with Febrile Seizures Plus. But there are some more aspects about *SCN1A* that are less well known

...read more

Epilepsy genes A-Z

A

Gene 1	Gene 5	Gene 9	Gene 13
Gene 2	Gene 6	Gene 10	Gene 14
Gene 3	Gene 7	Gene 11	Gene 15
Gene 4	Gene 8	Gene 12	Gene 16

B

Gene 1	Gene 5	Gene 9	Gene 13
Gene 2	Gene 6	Gene 10	Gene 14
Gene 3	Gene 7	Gene 11	Gene 15
Gene 4	Gene 8	Gene 12	Gene 16

Recent blog posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Receive most recent updates on epilepsies together with xxx other people

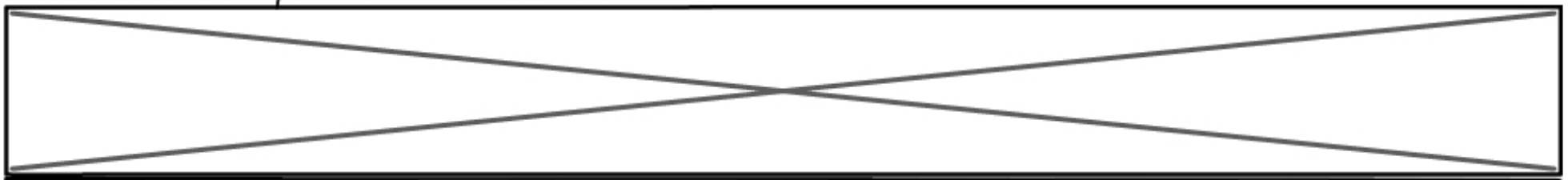
- Gene A and disease B
- Gene B and disease C
- Gene C and disease D
- Gene D and disease E
- Gene E and disease F

Beyond the Ion channel

http://channelopathist.net

Epilepsiome | A database on epilepsy and genes by the Genetics Commission of the ILAE

search



Home Diesease Overview Gene Overview About ILAE GC

SCN1A - quick summary

SCN1A is the most prominent gene for human epilepsies
Mutations in SCN1A cause Dravet Syndrome and GEFS+
SCN1A encodes a voltage-gated sodium channel

.

most recent updates - April 2014

GWAS study finds *SCN1A* variants as risk factor for common epilepsies
A recent large scale genome-wide association study finds that common variants in the SCN1A predispose to a much wider spectrum of epilepsies than previously thought, discovering a trace of the effect of SCN1A in places where researchers did not expect it.
...read blog post of the ILAE-GC here

SCN1A - the gene Expand

SCN1A - related diseases Expand

SCN1A - gene scoring Expand

SCN1A - selected references, recent updates, EpiGAD Expand

Recent posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Follow this blog together with 5 other people

Archives

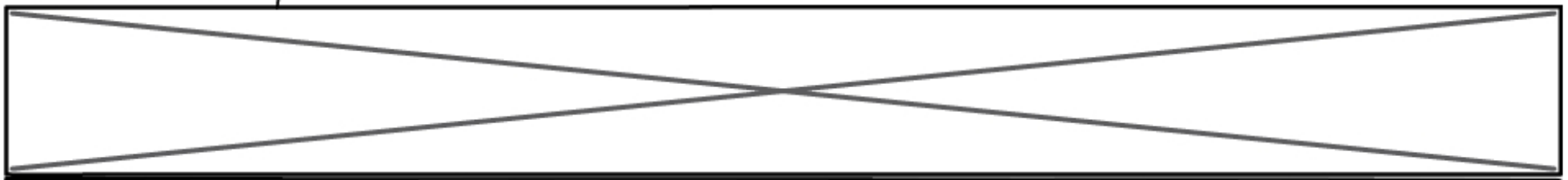
- April 2014
- March 2014
- February 2014
- January 2014
- December 2013

Beyond the Ion channel

http://channelopathist.net

Epilepsiome | A database on epilepsy and genes by the Genetics Commission of the ILAE

search



Home | Diesease Overview | Gene Overview | About ILAE GC

Dravet Syndrome - three things that you might want to know

(1) Dravet Syndrome is a severe genetic childhood epilepsy
(2) Seizures associated with fever are the hallmark of Dravet Syndrome
(3) 80% of patients with Dravet Syndrome carry mutations in SCN1A

.

most recent updates - April 2014

***GABRA1* & *STXBP1* - an unexpected cause of Dravet Syndrome**
A recent study in Neurology finds that some patients with Dravet Syndrome negative for mutations in *SCN1A* carry mutations in *GABRA1* and *STXBP1*. These genes had previously been implicated in other epilepsies and puzzling finding expands our understanding on the genetic basis of Dravet Syndrome
...read blog post of the ILAE-GC here

Dravet Syndrome - clinical features — Expand

Dravet Syndrome - genetics — Expand

Dravet Syndrome - useful references and links — Expand

Recent posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Follow this blog together with 5 other people

Archives

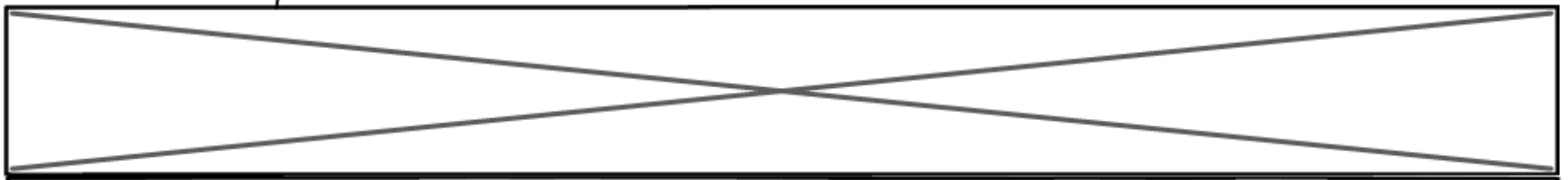
- April 2014
- March 2014
- February 2014
- January 2014
- December 2013

Beyond the Ion channel

http://channelopathist.net

Epilepsiome | A database on epilepsy and genes by the Genetics Commission of the ILAE

search



Home Diesease Overview Gene Overview About ILAE GC

Channelopathies - Are we really beyond the ion channels?

Posted on 01.05.2014 by John Doe

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat. Duis aute irure dolor in reprehenderit in voluptate velit esse cillum dolore eu fugiat nulla pariatur. Excepteur sint occaecat cupidatat non proident, sunt in culpa qui officia deserunt

Section 1

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat. Duis aute irure dolor in reprehenderit in voluptate velit esse cillum dolore eu fugiat nulla pariatur. Excepteur sint occaecat cupidatat non proident, sunt in culpa qui officia deserunt

Section 2

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat.

Recent posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Follow this blog together with 5 other people

Archives

- April 2014
- March 2014
- February 2014
- January 2014
- December 2013

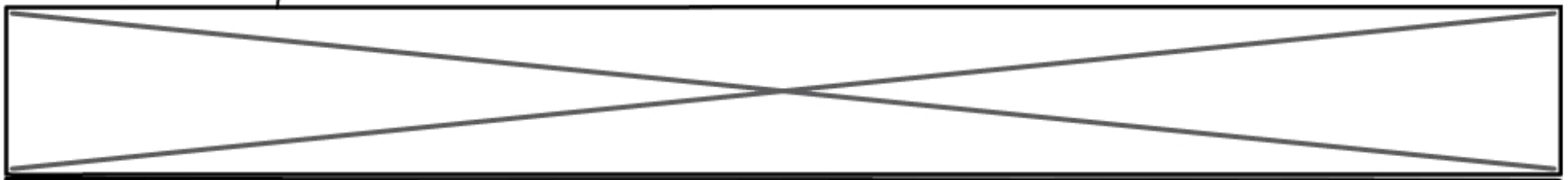
Beyond the Ion channel

http://channelopathist.net

Epilepsiome

A database on epilepsy and genes by the Genetics Commission of the ILAE

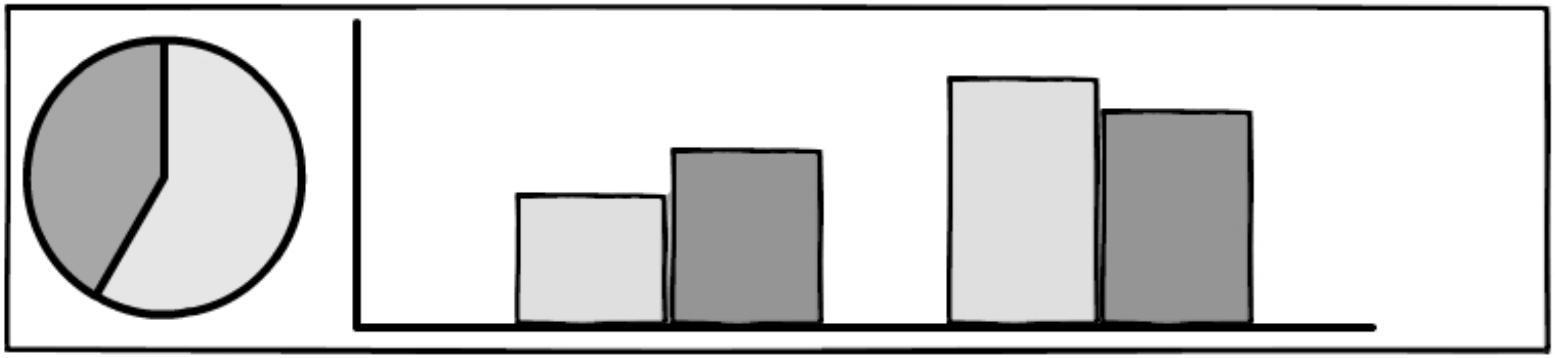
search



Home Diesease Overview Gene Overview About ILAE GC

Finding the tiny, tiny puzzle pieces of genetic risk or what is a genome-wide association study?

Posted on 01.05.2014 by John Doe



Section 1

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat. Duis aute irure dolor in reprehenderit in voluptate velit esse cillum dolore eu fugiat nulla pariatur. Excepteur sint occaecat cupidatat non proident, sunt in culpa qui officia deserunt

Recent posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Follow this blog together with 5 other people

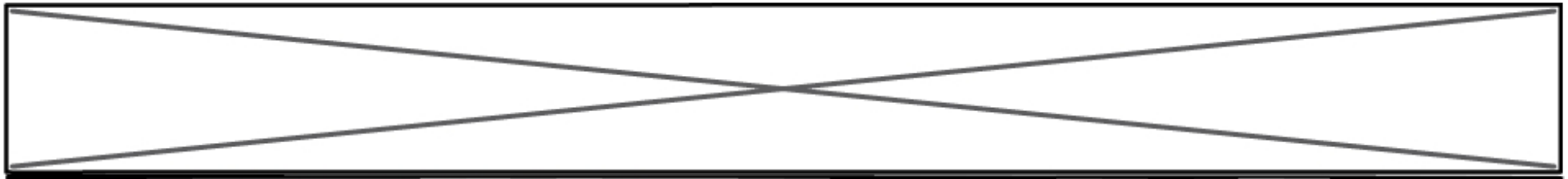
Archives

- April 2014
- March 2014
- February 2014
- January 2014
- December 2013

Beyond the Ion Channel

search

The Channelopathist is a blog
about the genetics of the epilepsies



Blog post on gene 1

Posted on 01.05.2014 by John Doe

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat. Duis aute irure dolor in reprehenderit in voluptate velit esse cillum dolore eu fugiat nulla pariatur. Excepteur sint occaecat cupidatat non proident, sunt in culpa qui officia deserunt

posted in 2014, term 1, term 2, term 3, term 4, term 5

Blog post on gene 2

Posted on 01.05.2014 by John Doe

Lorem ipsum dolor sit amet, consectetur adipisicing elit, sed do eiusmod tempor incididunt ut labore et dolore magna aliqua. Ut enim ad minim veniam, quis nostrud exercitation ullamco laboris nisi ut aliquip ex ea commodo consequat. Duis aute irure dolor in reprehenderit in voluptate velit esse cillum dolore eu

Recent posts

- Blog post on gene 1
- Blog post on gene 2
- Blog post on gene 3
- Blog post on gene 4
- Blog post on gene 5

Follow this blog together with 5 other people

Archives

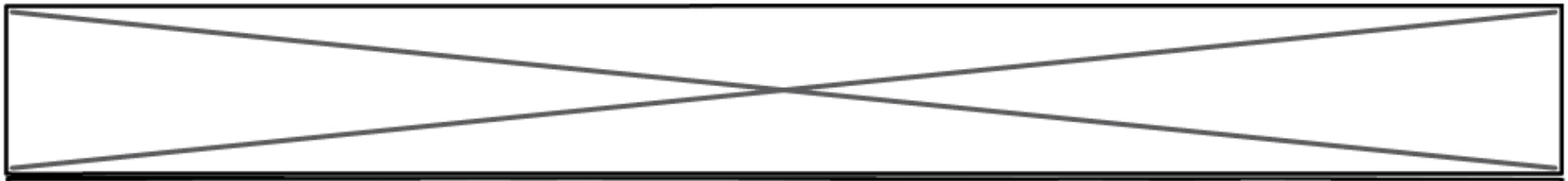
- April 2014
- March 2014
- February 2014
- January 2014
- December 2013

Beyond the Ion channel

http://channelopathist.net

EpiGAD The Genetic Association Database of the ILAE - integrated in Epilepsiome

search



Home About ILAE-GC Beyond the Ion Channel EpiGAD

Gene	Study	phenotype	rs	p-value	OR	comment
SCN1A	Epicure	IGE	rs234235	10 e-5	2.1	GWAS
STXBP1	Epicure	IGE	rs675435	5 e-6	1.1	GWAS
SCN9A	Epicure	IGE	rs34665	7 e-4	0.9	GWAS
GABRG2	Asian collaborative study	BRE	rs43535	10 e-5	1.1	
PNPO	XYZ group	TLE	rs859535	5.6 e-6	1.5	
ME2	TLE study group	TLE	rs234235	10 e-5	2.1	GWAS
BRD2	Doe et al.	IGE	rs234235	10 e-5	2.1	
SYNGAP1	XY Consortium	TLE	rs234235	5 e-5	2.5	GWAS
SLC2A1	Mueller et al.	focal	rs234235	6 e-5	2.1	
SCN3A	Meyer et al.	all epilepsy	rs234235	7 e-5	2.5	

What is EpiGAD

Posted on 01.05.2014 by John Doe

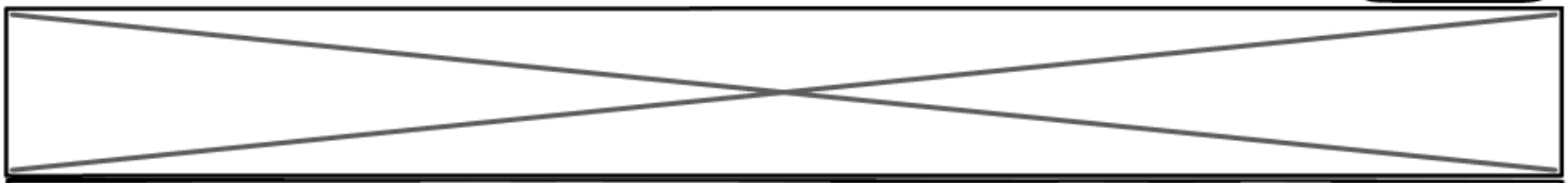
EpiGAD is the genetic association database of the ILAE Genetic Commission. We survey the literature of genetic association studies in the field and collate the data on the reported results within this webpage.

Beyond the Ion channel

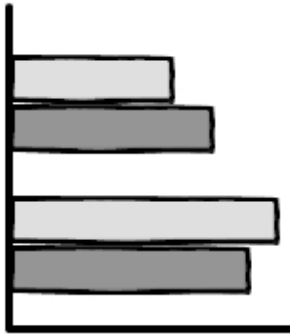
http://channelopathist.net

Epilepsiome | Gene scoring of epilepsy-related genes

search



Home About ILAE-GC Beyond the Ion Channel EpiGAD



The concept of gene scoring

1 - secure & established
2 - very probable
3 - probable
4 - suggestive
5 - some suggestive data, but insufficient
6 - insufficient data
7 - not implicated

Gene	Score	comment
SCN1A	1 - established	
SCN9A	6 - data insufficient	variants in LD with SCN1A
ME2	6 - data insufficient	
EFHC1	5 - some data but inconclusive	
CHD2	1 - secure	

What is Gene Scoring

Posted on 01.05.2014 by John Doe

All genes related to human epilepsies on the Channelopathist have been reviewed by researchers the field and have been assigned a score relating to their probability to be involved in human epilepsies. This mechanism that had been used for autism-related genes in the past has been adapted to the epilepsies.

Beyond the Ion channel

http://channelopathist.net

Epilepsiome | SiteMap

search

Home About ILAE-GC Beyond the Ion Channel EpiGAD