

The SCN2A Study consist of two inter-connected studies:

SCN2A
Clinical Trial Readiness Study (CTRS)

The SCN2A Clinical Trial Readiness Study (CTRS) is a longitudinal study designed with parents to assess outcomes in their children that are life changing, life limiting, and ultimately important to parents. A primary goal is to prepare the SCN2A community with necessary outcomes measures for precision medicine clinical trials when those new therapies are available.

SCN2A Global Survey

The SCN2A Global survey is a one-time survey that contains many of the same questions as the CTRS.

If you are interested in registering for one of these studies, please visit our website to learn more at <https://scn2a.study/>

Who is funding the study?

This study is entirely funded by the FamilieSCN2A Foundation.

Need to get back to the survey?

Did you start the CTRS or Global survey and lost the address? No worries, just visit the site from this link and sign in with your email and password. You can reset your password if you forgot

<https://scn2actrs.study/> . You can also see the results from our other Natural History projects with different groups.

Dissemination of results and data sharing:

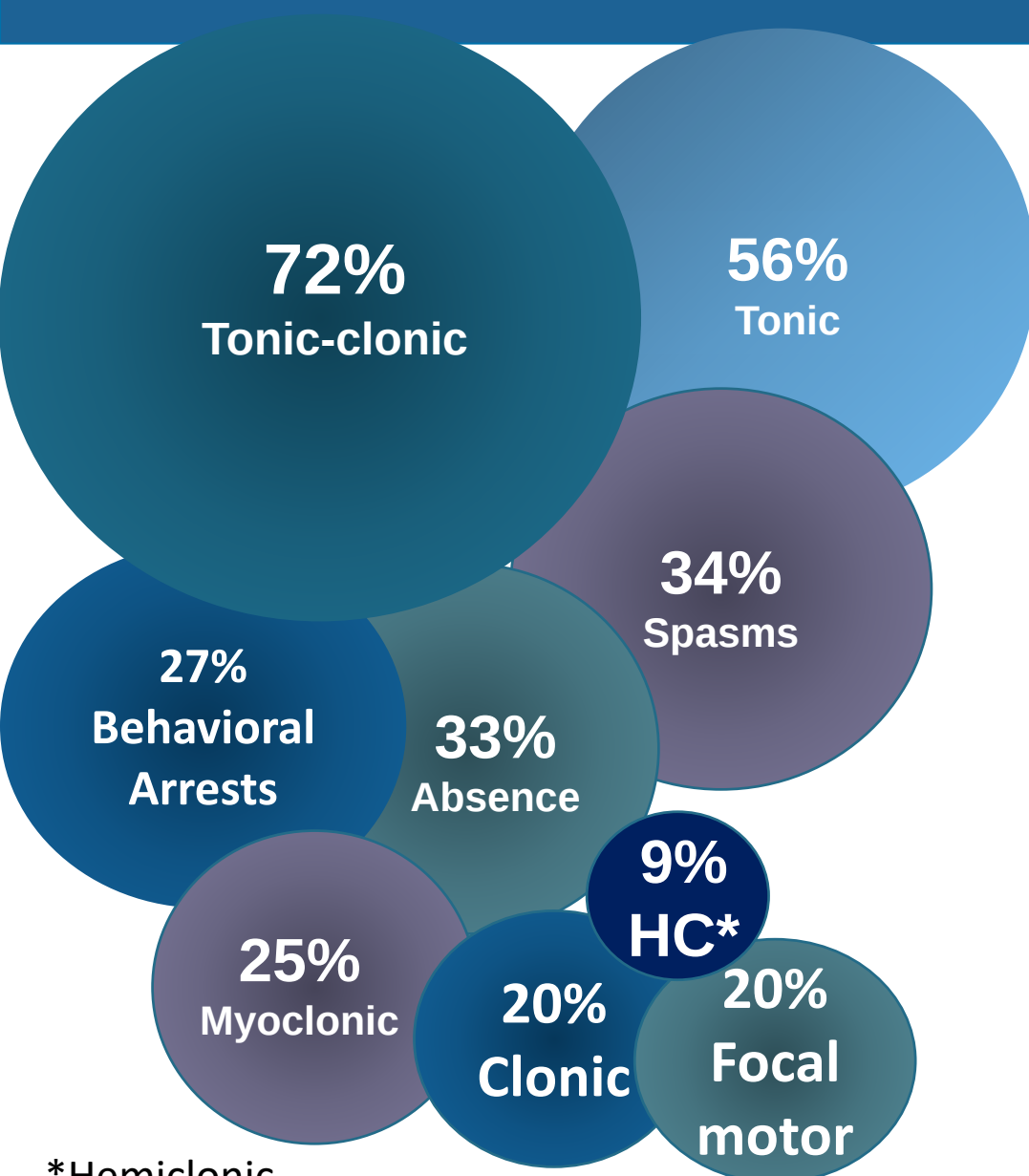
The data from this study are shared with the family community through graphics such as this first one.

They will be presented and published for the scientific community. Completed and locked data will also be archived with the Simons Foundation and available to authorized investigators. At the end of the study, parents may also download a copy of all of their own data.

Have questions?

Contact us at
WhatIsEpilepsy@LurieChildrens.org
or contact the FamilieSCN2A
foundation at **Impact@SCN2A.org**

EPILEPSY AND SEIZURES



48 Children diagnosed with epilepsy

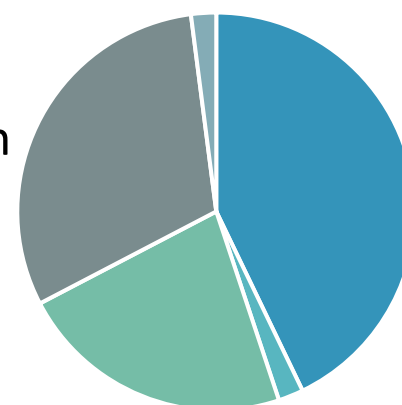
Specific epilepsy diagnoses reported

- 28% Infantile spasms
- 13% Ohtahara
- 26% Lennox-Gastaut
- 4% ESES
- 2% Migrating focal seizures

Age at first seizure

Median age = 3.5 months

- First week
- 1-4 weeks
- 1-11 month
- 1-4 years
- 5+ years



*Hemiclonic



93% currently on anti-seizure medications

During past 3 months



31 (69%) had seizures

Of those...

21 (68%) –convulsive seizures

16 (52%) – behavioral and myoclonic sz.

7 (23%) – other seizure types



22% children had prolonged seizures



20% children needed rescue medications

MEDICAL HISTORY



LABOR – DELIVERY – FIRST MONTH



23% - pre-term
35% - complications during labor&delivery
22% - emergency c-section

53% - evaluated & treated for serious condition in neonatal period

32% problems feeding
9% failure to thrive
20% jaundice
12% breathing problems/apnea
16% encephalopathy
34% neonatal epilepsy

40% admitted to NICU
25% NICU admission ≥ 1 month



COMMON MEDICAL CONDITIONS



20% reported conditions affecting the lungs
These were related to low muscle tone, aspiration and tracheostomies



Dental

13% Teeth grinding
11% Late baby teeth

18% reported endocrine dysfunction
9% precocious puberty



Musculoskeletal

20% Dystonia
38% Hypotonia
17% Spasticity
23% Scoliosis



Vision



71% had vision/eye concerns

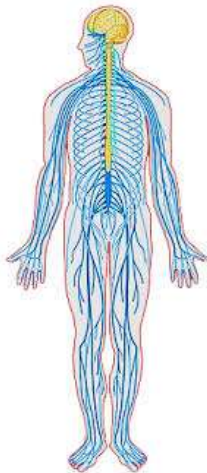
- 41% Cortical-visual impairment
- 13% Difficulty judging distance, depth, visual-motor impairment

AUTONOMIC SYMPTOMS, SLEEP & BEHAVIOR

DYSAUTONOMIAS

Autonomic symptoms in past 3 months

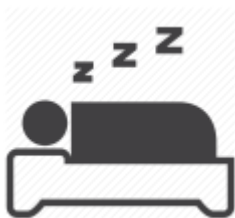
- 57% Temperature regulation
- 32% Sweating
- 29% Reflux/vomiting
- 53% High pain tolerance
- 27% Abnormal Pupil Dilation/contraction
- 54% Gastro-intestinal
- 26% Urinary retention
- 43% Excessive irritability



82% Of children had 1 or more autonomic symptoms
5 Median number of autonomic symptoms reported

Autonomic Storms: 13% Yes, in past 3 months
8 % Yes but not recently

SLEEP DISTURBANCE



37% of those with epilepsy reported seizures in sleep
87% of all children had non-seizure awakenings
46% had awakenings 3-7 nights/week



BEHAVIOR

Autism
43% Have a diagnosis
23% Have features



Aggressive behavior
2% Have a diagnosis
21% Have features

Obsessive-Compulsive Disorder
2% Have a diagnosis
13% Have features



Anxiety
10% Have a diagnosis
17% Have features





BASIC FUNCTION AND ABILITIES



MOTOR



75% Moderate to severe gross motor delay



39% 2y and older depend on a mobility device for any distance



18% cannot grasp objects with their hands



21% walk outside independently for distances

Cannot control head

Cannot sit independently

Have to be lifted or hoisted



12% pick up raisin with thumb and forefinger

COMMUNICATION



89% Moderate to severe language delay



72% inconsistently or rarely communicate even with people they know (e.g. family)



29% speak a few or more words

16% speak as their primary form of communication

15% communicate by sign or device only

56% have no symbolic language

15% use >100 words/signs/symbols

8% combine 3+ words into sentences

23% understand >100 words/signs/symbols



ACTIVITIES OF DAILY LIVING



SELF-CARE



5% dress themselves



21% use spoon and fork



5% brush own teeth



9% wash and dry their hands



12% drink from a cup



22% use a touchscreen device



14% scribble or write with crayon



5% are completely independent for toilet use

EATING



44% are completely dependent on a caregiver for feeding



Feeding tube

20% Exclusively G-tube fed
9% Partially G-tube-fed

Can drink or eat these foods with no difficulty*

60% Water

66% Soft solids (apple sauce)

57% Bite-sized soft food like piece of bread or banana



46% Bite-sized crunchy food like crackers

23% Bite-sized piece of chewy food like meat



20% Bite into a crispy food like an apple



*Non G-tube dependent only