The SCN2A Study consists 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 outcome 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/](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/](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
Parents from the USA, Canada, UK, Ireland, Brazil, Columbia, Germany, Spain, Georgia, and India

44% ♀ 56% ♂

Age at enrollment

- <2 years: 5
- 2-4 year: 14
- 5-9 years: 28
- 10-19 years: 35
- >=20 years: 18

Year of SCN2A diagnosis for 55 families reporting information

Number of Participants

YEAR OF SCN2A DIAGNOSIS
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

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

93% currently on anti-seizure medications

During past 3 months

20% children needed rescue medications
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

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
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

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

Aggressive behavior
- 2% Have a diagnosis
- 21% 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
- **12%** pick up raisin with thumb and forefinger
- **21%** walk outside independently for distances
- **Cannot control head**
- **Cannot sit independently**
- **Have to be lifted or hoisted**

### COMMUNICATION

- **89%** Moderate to severe language delay
- **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
- **72%** inconsistently or rarely communicate even with people they know (e.g. family)
ACTIVITIES OF DAILY LIVING

SELF-CARE

- 5% dress themselves
- 21% use spoon and fork
- 22% use a touchscreen device
- 14% scribble or write with crayon
- 5% brush own teeth
- 9% wash and dry their hands
- 5% are completely independent for toilet use
- 12% drink from a cup

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