

**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 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/>**



### **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](mailto:WhatIsEpilepsy@LurieChildrens.org)  
or contact the FamilieSCN2A  
foundation at [Impact@SCN2A.org](mailto:Impact@SCN2A.org)

# DEMOGRAPHICS

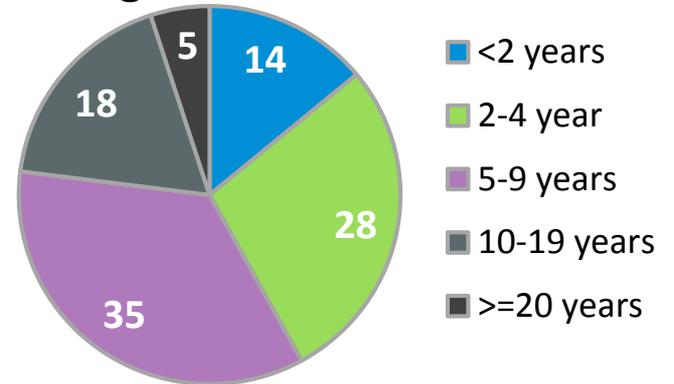


57

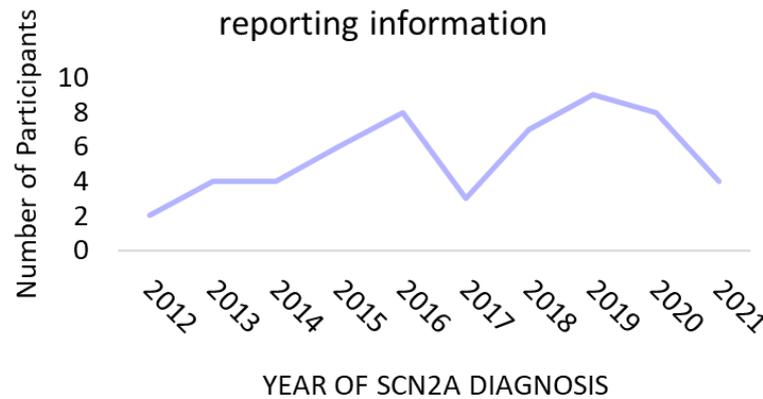
Parents from the USA, Canada, UK, Ireland, Brazil, Columbia, Germany, Spain, Georgia, and India

44% ♀ 56% ♂

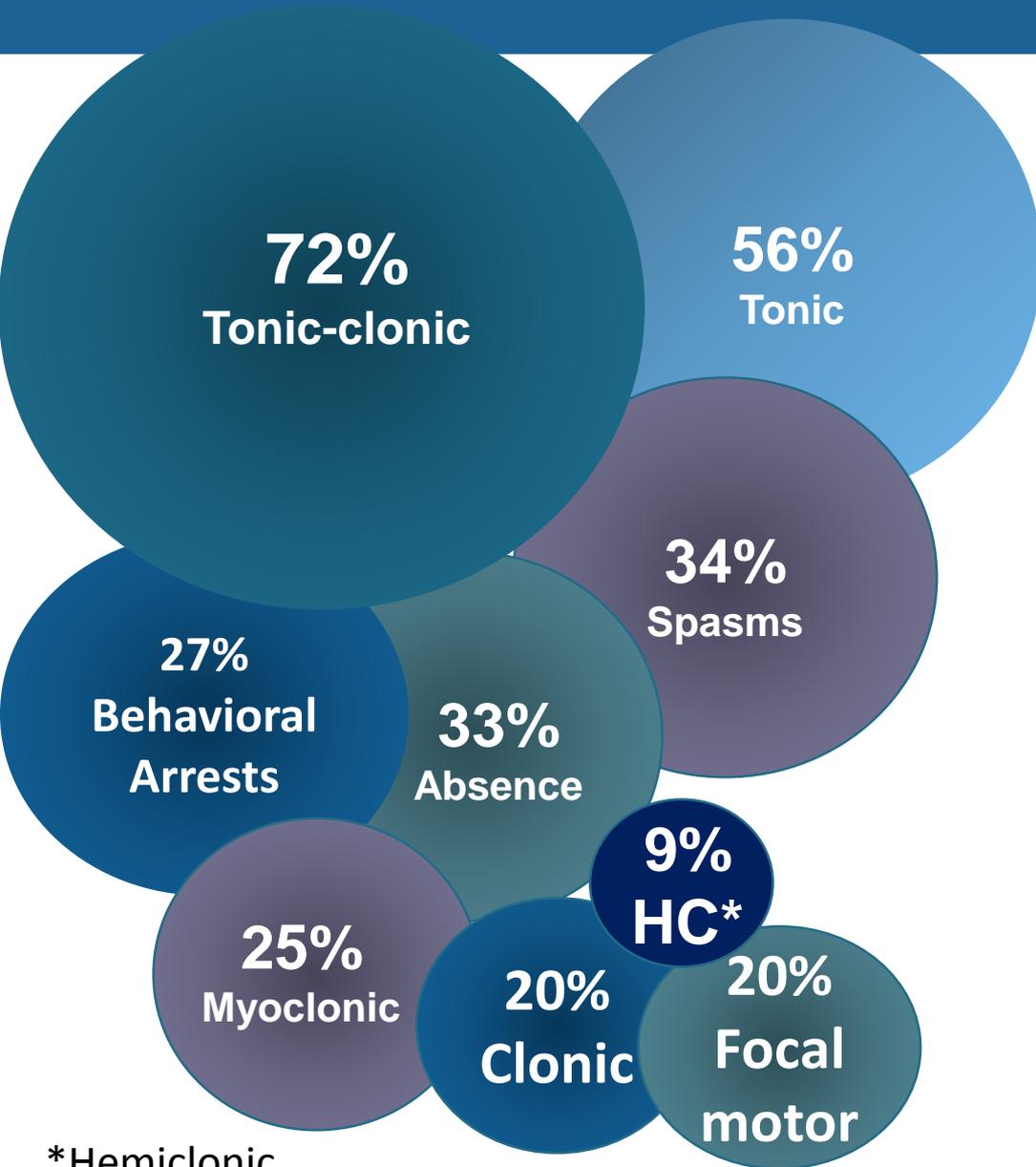
## Age at enrollment



Year of SCN2A diagnosis for 55 families reporting information



# EPILEPSY AND SEIZURES



\*Hemiclonic

48 Children diagnosed with epilepsy

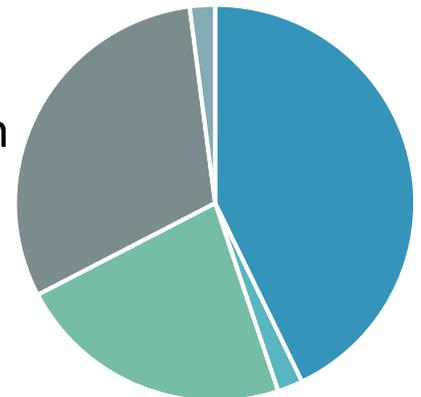
Specific epilepsy diagnoses reported

- 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



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



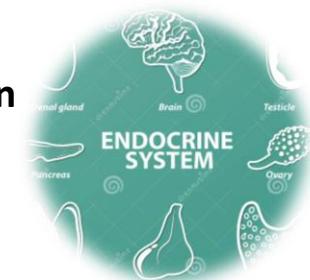
**20% reported conditions affecting the lungs**  
**These were related to low muscle tone, aspiration and tracheostomies**



### 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

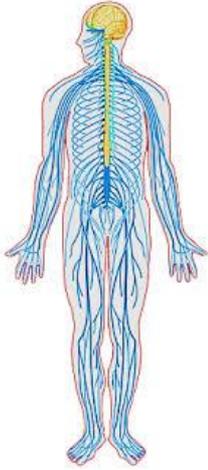


**71% had vision/eye concerns**

- **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

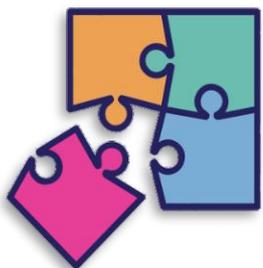
13% Have features



**Anxiety**

10% Have a diagnosis

17% Have features





# BASIC FUNCTION AND ABILITIES



## MOTOR



**75% Moderate to severe gross motor delay**



**21% walk outside independently for distances**



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

Cannot control head

Cannot sit independently

Have to be lifted or hoisted



**18% cannot grasp objects with their hands**



**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**

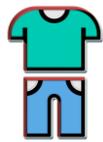
**23% understand >100 words/signs/symbols**



# 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