SIMONS SEARCHLIGHT

Driven by science. United by hope.

SCN2A

July 2019

Registry Snapshot

About this Report

Most of the information in this report comes from *SCN2A* families completing the Simons Searchlight medical history phone call. Families also complete follow-up phone calls every year so we can track progress over time. We are reporting on a total of 76 participants.

The information presented in this report is a summary of data contributed by SCN2A families with genetic changes classified as "pathogenic" or "likely pathogenic". To present an accurate picture of conditions related to *SCN2A*, genetic changes classified as "variants of uncertain/unknown significance" were not included here.

The data included in this report represents **current information** in the registry (as of July 1, 2019), prior to official data cleaning and release. It is not intended for publication.

Participation

After registering for a research account, what does it take for a participant's data to be released to researchers?

1. Sign up online. **SimonsSearchlight.org***

2. Provide your genetic lab report

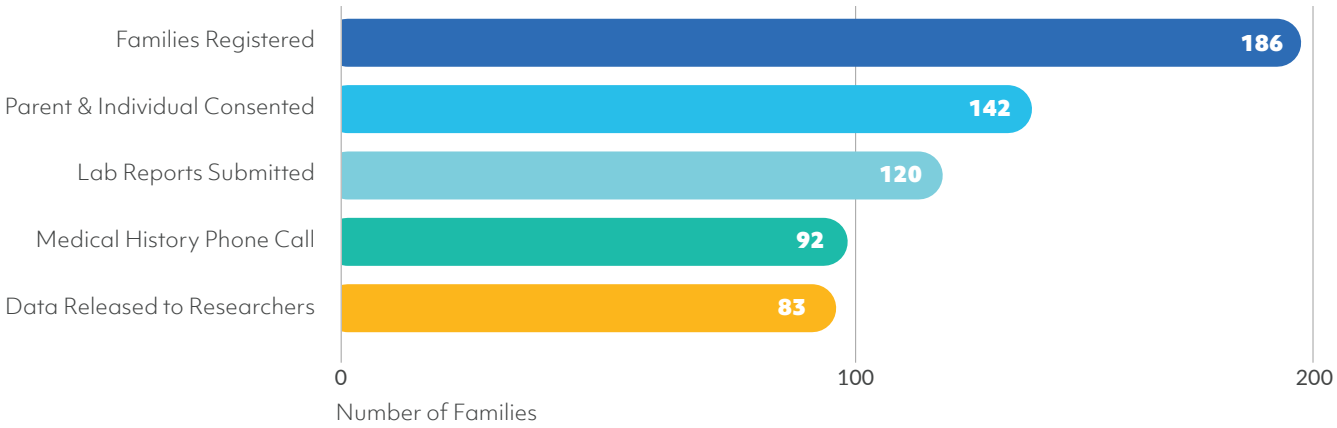
3. Discuss your medical history with a genetic counselor

4. Unite with researchers and other families

* If you had previously signed up for Simons VIP, click the link in your email to link your old account with your new Simons Searchlight account. No need to re-register!

Where are *SCN2A* families in this process?

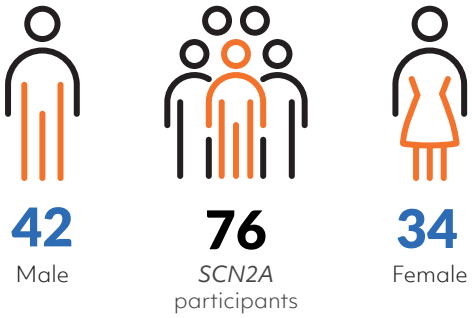
Finish any missing steps to contribute your family's data!



Note: Numbers as of July 26 2019, a new data release is anticipated August 2019.

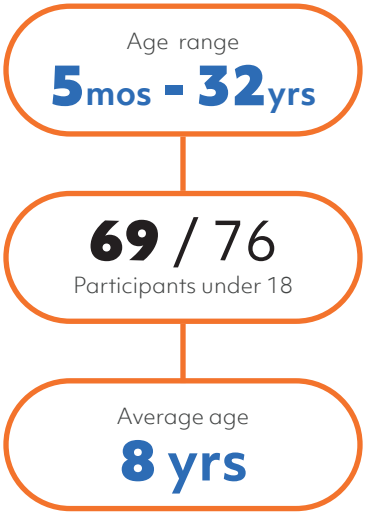
Total Males and Females

Among the 76 participants included, there are 42 males and 34 females who have the *SCN2A* genetic change.



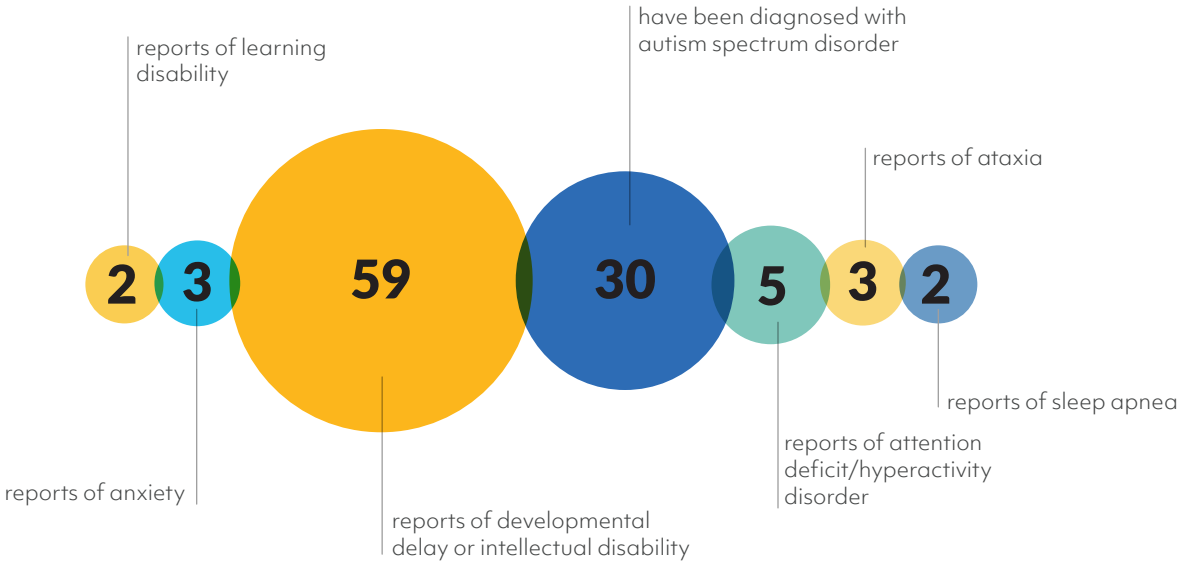
Age Range

The average age is 8 years old. Ages range from 5 months to 32 years, and 69 of the 76 participants are under 18.



Developmental & Behavioral Diagnoses

Simons VIP completed diagnostic history interviews with 77 participants.



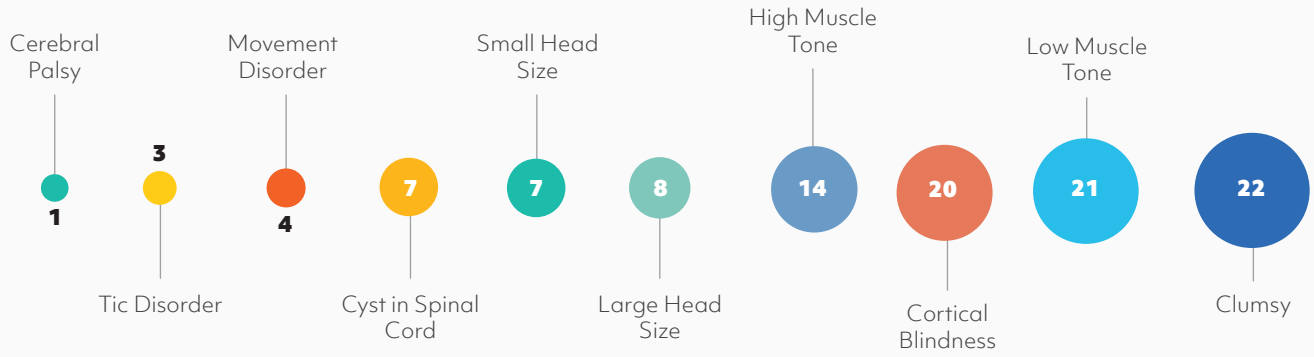
Other: bipolar, depression, PTSD, pica, non-ambulatory, regression due to seizures, incontinence, minimally verbal

Reported Conditions

Most Commonly Reported Conditions

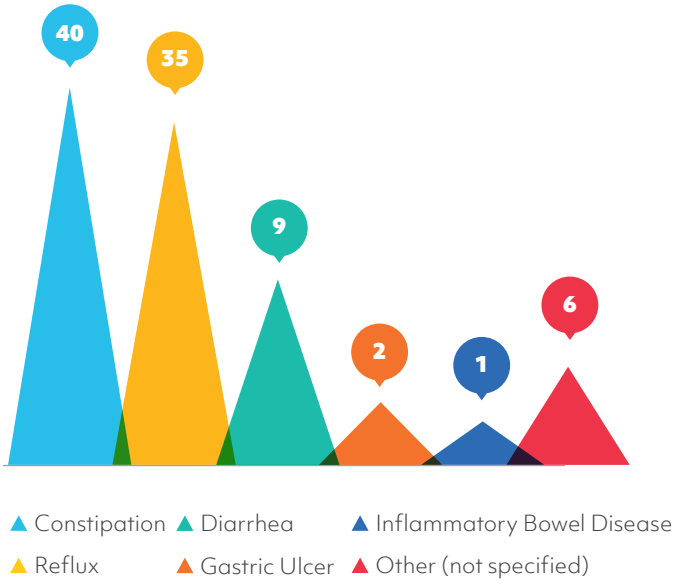
SCN2A families reported a variety of associated medical conditions. The most common medical conditions were reported in the brain, eyes, and digestive system. The specific medical conditions that were reported are displayed in the graphs on this page.

Neurological Conditions



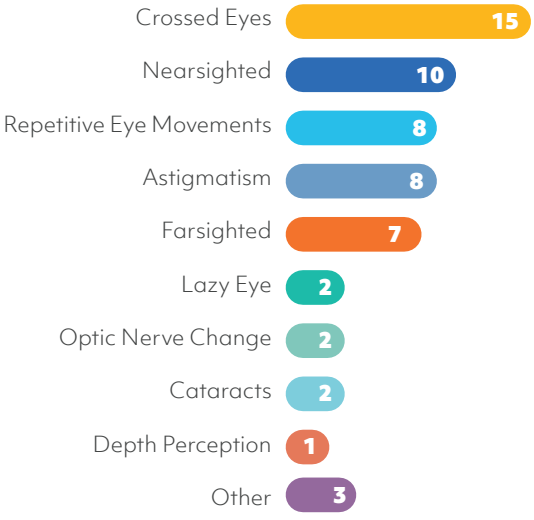
Gastrointestinal Conditions

Note: Individual participants may have reported multiple conditions



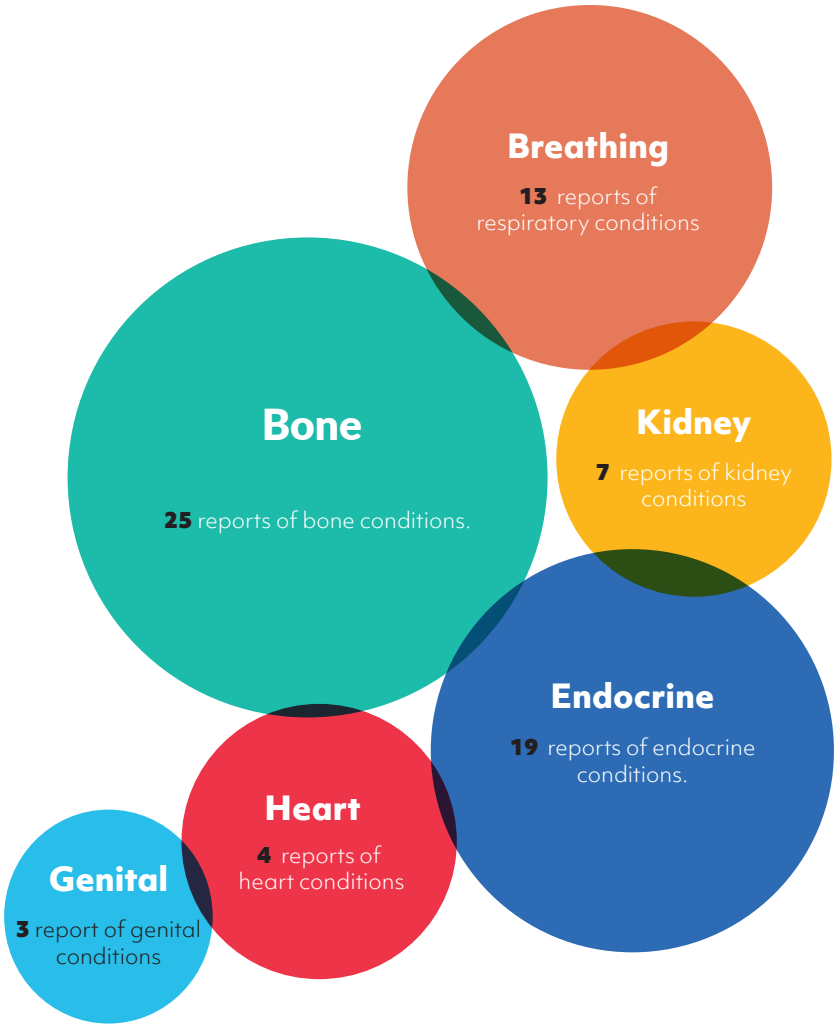
Vision Conditions

Note: Individual participants may have reported multiple conditions



Other Conditions Reported

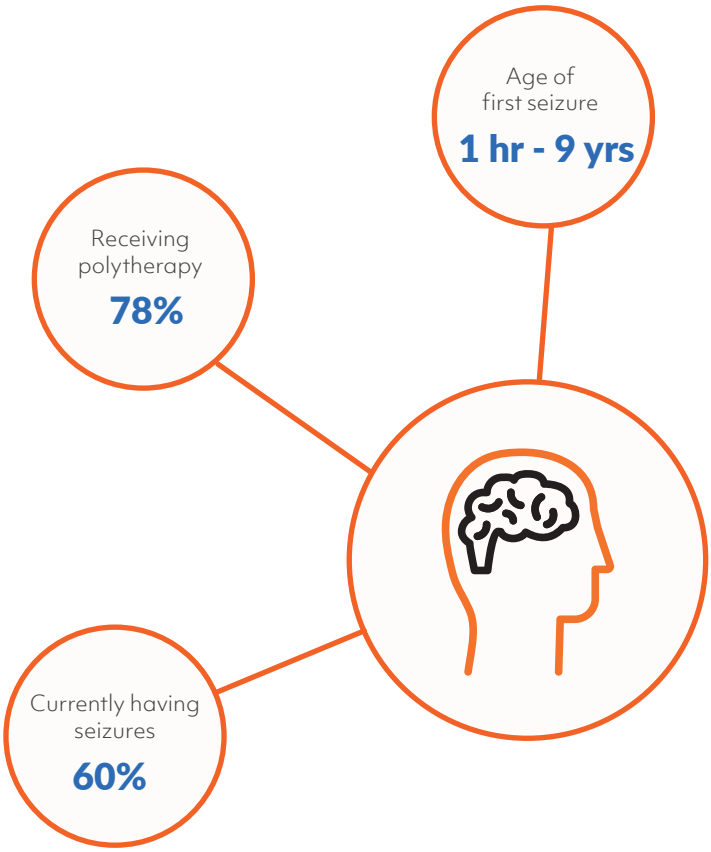
Reports of other conditions varied widely for this group of 76 participants. The graphic below shows the total number of participants reporting a condition. Individual participants may have reported multiple conditions across multiple systems.



Seizures and Medications

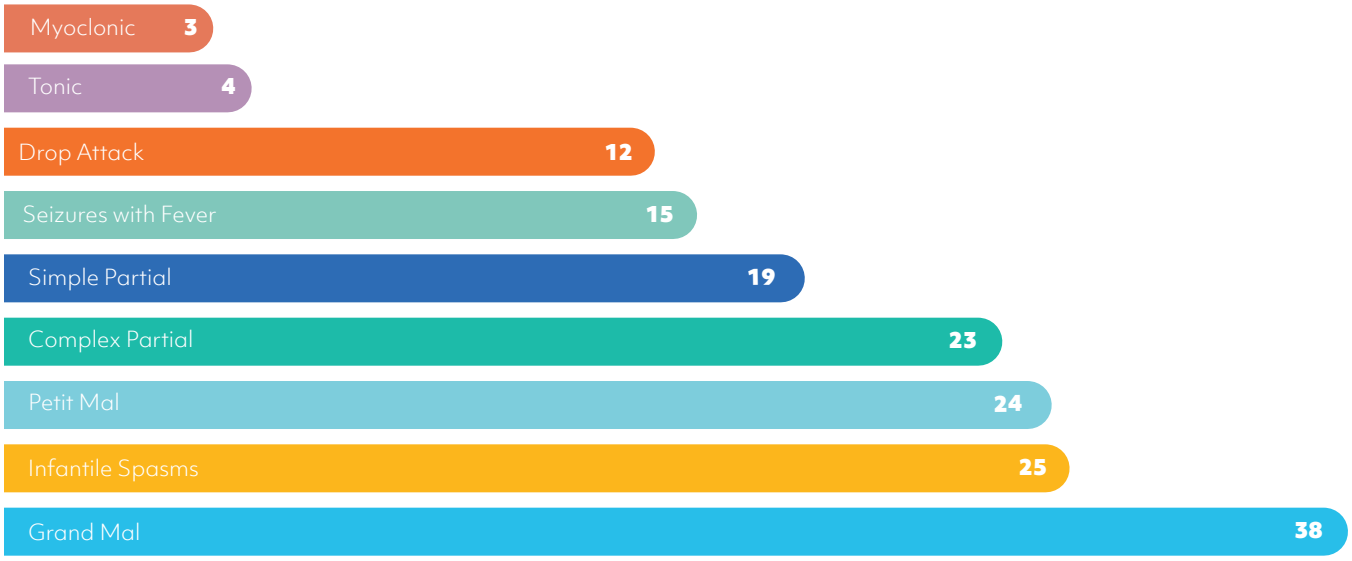
Seizures

668 participants reported details of seizure history and treatment.

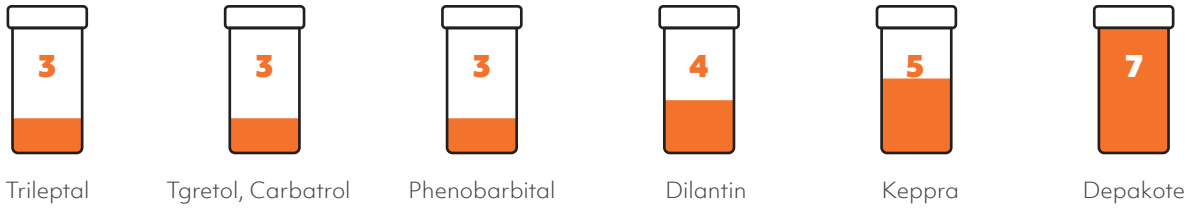


Types of Seizures

Seizures were reported in 61 indiviudals. Individual participants may have reported multiple seizure types.



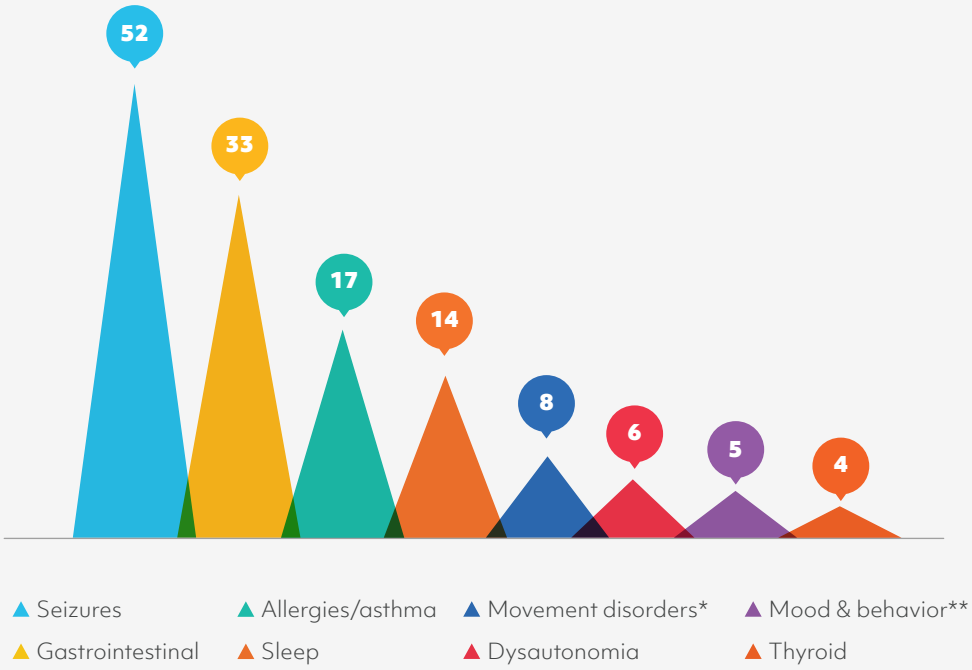
Medication Reported to Work Best for Seizures



* 8 particiapnts reported polytherapy (more than one) - most common are lacosamide, lamotrigine, levetiracetam or diastat in combination with others.
** 13 reported another medication working best - Clobazam (2), Diastat (1), Lamotrigine (2), Topirimate (1), Vigabatrin (1), Zonisamide (1), ACTH (1), not specified (4)

Current Medication Types Taken by Participants

Current medications taken from the most recent medical history interview.



* Some with reported dysautonomia
** includes cannabidiol and medical cannabis

THANK YOU

SIMONS SEARCHLIGHT
Driven by science. United by hope.

SCN2A RELATED AUTISM & EPILEPSY
FAMILIESCN2A FOUNDATION
WWW.SCN2A.ORG

Source:
SimonsSearchlight.org
base.sfari.org

Simons Searchlight is the official registry of the Familie*SCN2A* Foundation.