SCN2A
July 2019
Registry Snapshot
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.
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!

<table>
<thead>
<tr>
<th>Step</th>
<th>Number of Families</th>
</tr>
</thead>
<tbody>
<tr>
<td>Families Registered</td>
<td>186</td>
</tr>
<tr>
<td>Parent &amp; Individual Consented</td>
<td>142</td>
</tr>
<tr>
<td>Lab Reports Submitted</td>
<td>120</td>
</tr>
<tr>
<td>Medical History Phone Call</td>
<td>92</td>
</tr>
<tr>
<td>Data Released to Researchers</td>
<td>83</td>
</tr>
</tbody>
</table>

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.

- reports of anxiety: 2
- reports of learning disability: 3
- reports of developmental delay or intellectual disability: 59
- reports of ataxia: 3
- reports of sleep apnea: 2
- reports of attention deficit/hyperactivity disorder: 30
- have been diagnosed with autism spectrum disorder: 5

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

### Most Commonly Reported Conditions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Reported Cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cerebral Palsy</td>
<td>22</td>
</tr>
<tr>
<td>Movement Disorder</td>
<td>21</td>
</tr>
<tr>
<td>Small Head Size</td>
<td>20</td>
</tr>
<tr>
<td>High Muscle Tone</td>
<td>14</td>
</tr>
<tr>
<td>Low Muscle Tone</td>
<td>21</td>
</tr>
<tr>
<td>Tic Disorder</td>
<td>7</td>
</tr>
<tr>
<td>Cyst in Spinal Cord</td>
<td>7</td>
</tr>
<tr>
<td>Large Head Size</td>
<td>8</td>
</tr>
<tr>
<td>Cortical Blindness</td>
<td>15</td>
</tr>
<tr>
<td>Clumsy</td>
<td>22</td>
</tr>
<tr>
<td>Constipation</td>
<td>40</td>
</tr>
<tr>
<td>Diarrhea</td>
<td>35</td>
</tr>
<tr>
<td>Inflammatory Bowel Disease</td>
<td>9</td>
</tr>
<tr>
<td>Reflux</td>
<td>2</td>
</tr>
<tr>
<td>Gastric Ulcer</td>
<td>1</td>
</tr>
<tr>
<td>Other (not specified)</td>
<td>6</td>
</tr>
<tr>
<td>Neurological Conditions</td>
<td></td>
</tr>
<tr>
<td>Gastrointestinal Conditions</td>
<td></td>
</tr>
<tr>
<td>Vision Conditions</td>
<td></td>
</tr>
</tbody>
</table>

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.

- **Breathing**: 13 reports of respiratory conditions.
- **Bone**: 25 reports of bone conditions.
- **Kidney**: 7 reports of kidney conditions.
- **Endocrine**: 19 reports of endocrine conditions.
- **Heart**: 4 reports of heart conditions.
- **Genital**: 3 reports of genital conditions.
Seizures and Medications

Seizures
668 participants reported details of seizure history and treatment.

Types of Seizures
Seizures were reported in 61 individuals. Individual participants may have reported multiple seizure types.

- Myoclonic 3
- Tonic 4
- Drop Attack 12
- Seizures with Fever 15
- Simple Partial 19
- Complex Partial 23
- Petit Mal 24
- Infantile Spasms 25
- Grand Mal 38

Age of first seizure
1 hr - 9 yrs

Receiving polytherapy
78%

Currently having seizures
60%
Medication Reported to Work Best for Seizures

- Trileptal: 3
- Tgretol, Carbatrol: 3
- Phenobarbital: 3
- Dilantin: 4
- Keppra: 5
- Depakote: 7

* 8 participants 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), Topiramate (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.

- Seizures: 52
- Allergies/asthma: 33
- Movement disorders*: 17
- Mood & behavior**: 14
- Gastrointestinal: 8
- Sleep: 6
- Dysautonomia: 5
- Thyroid: 4

* Some with reported dysautonomia
** Includes cannabidiol and medical cannabis
THANK YOU

Source: SimonsSearchlight.org base.sfari.org

Simons Searchlight is the official registry of the FamilieSCN2A Foundation.