Most of the information in this report comes from STXBP1 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 39 participants.

The information presented in this report is a summary of data contributed by STXBP1 families with genetic changes classified as “pathogenic” or “likely pathogenic”. To present an accurate picture of conditions related to STXBP1, 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, 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 STXBP1 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>109</td>
</tr>
<tr>
<td>Parent &amp; Individual Consented</td>
<td>64</td>
</tr>
<tr>
<td>Lab Reports Submitted</td>
<td>67</td>
</tr>
<tr>
<td>Medical History Phone Call</td>
<td>39</td>
</tr>
<tr>
<td>Data Released to Researchers*</td>
<td>32</td>
</tr>
</tbody>
</table>

* Additional data to be release July 2019
Total Males and Females
Among the 39 participants included, there are 16 males and 23 females who have the STXBP1 genetic change.

Age Range
The average age is 6.7 years old. Ages range from 4 months to 23 years, and 37 of the 39 participants are under 18.

Developmental & Behavioral Diagnoses
Simons Searchlight completed diagnostic history interviews with 38 individuals.

- 2 reported other learning disorder
- 13 have been diagnosed with autism spectrum disorder
- 34 reported to be minimally verbal
- 19 reports of developmental delay or intellectual disability
- 14 reports of language disorder (without intellectual disability)
- 13 regression due to seizures*

* Regression and sensory processing may be underestimates; more families may report these issues over time
** Other conditions reported once: ADHD, sensory processing disorder without autism, pica
Neurological Conditions
Note: Individual participants may have reported multiple conditions

- Small head size: 1
- Large head size: 3
- Clumsy: 4
- Cerebral palsy: 9
- Low muscle tone: 33

Gastrointestinal Conditions
Note: Individual participants may have reported multiple conditions

- Reflux: 23
- Diarrhea: 16
- Constipation: 5
- Other (not specified): 2

Vision Problems
Note: Individual participants may have reported multiple conditions

- Cortical Blindness: 5
- Astigmatism: 4
- Crossed Eyes: 4
- Nearsighted: 3
- Farsighted: 3
- Repetitive Eye Movements: 2
- Drooping Eyelid: 1
- Other: 2

STXBP1 families reported a variety of associated medical conditions. Some 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.
**Other Conditions Reported**

Some other conditions reported for this group of 39 participants are shown in the graphic below as total number of conditions reported. Individual participants may have reported multiple conditions across multiple systems.

*Heart conditions include: high BP, atrial septal defect, sinus tachycardia, pulmonary AVM (hereditary hemorrhagic telangiectasia), pulmonary fistula

**Types of Seizures**

Note: Individual participants may have reported multiple conditions

- Febrile: 1
- Myoclonic: 1
- Drop Attack: 4
- Petit Mal: 8
- Simple Partial: 9
- Infantile Spasms: 12
- Complex Partial: 13
- Grand Mal: 16
Medication Use
Simons Searchlight collected medication use from 38 participants. Below we list the number of participants taking each medication type. Participants may be taking multiple medication types.

Medication Reported to Work Best for Seizures*

* Other meds less frequently reported by families to have good or complete seizure control: clonazepam (1) and phenobarb (1) (meds were weaned); Depakote/valproic acid (1); Tegretol/carbamazepine (1; med is still currently taken)