**Simons Searchlight Registry Update for STXB1P1**

**December 2021**

**Participant Age at Most Recent Interview**

**Developmental & Behavioral Diagnoses**

- Intellectual Disability/Development Delay: 67
- Autism: 24
- Language Impairment: 48
- ADHD: 6
- Anxiety: 2
- OCD: 1

**Notes:**
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.
- Graphs show data as of the last medical history interview completed on participants who have a pathogenic or likely pathogenic variant.

**How to Participate**

The information in this report is made possible by the active participation of the STXB1P1 community! Progress for individuals in your community with a STXB1P1 variant is shown below — log in to your simonssearchlight.org dashboard today to check for new surveys and tasks. Your data could hold the clues geneticists need to find answers.

**STEP 1**  
Sign up online.

**STEP 2**  
Provide your genetic lab report.

**STEP 3**  
Share your important medical history.

**STEP 4**  
Fill out surveys.

**STEP 5**  
Provide a blood sample if you are interested.

**STEP 6**  
Update us every year.

**Participant Age at Most Recent Interview**

**Developmental & Behavioral Diagnoses**

**Neurological Problems**

- Tic Disorder: 5
- Movement Disorder: 40
- Small Head Size: 7
- Large Head Size: 11
- Low Muscle Tone: 16
- High Muscle Tone: 14
- Clumsy: 7
- Cerebral Palsy: 36
- Cortical blindness: 5
- Seizures: 21

**Gastrointestinal Problems**

- GERD: 36
- Diarrhea: 5
- Constipation: 26
- Celiac Disease: 3

**Other**

- GERD: 5
- Diarrhea: 5
- Constipation: 5
- Celiac Disease: 5

**Participant Age at Most Recent Interview**

- 0-4: 114
- 5-9: 81
- 10-14: 71
- 15-19: 47
- 20-24: 21
- 25+: 19

**Data in these four graphs are from the medical history phone interviews collected in Simons Searchlight on 69 participants with STXB1P1.**
Information Spotlight: **Deeper Dive into Sleep**

The Simons Searchlight Sleep Supplement is a custom survey. We worked with members of the Simons Searchlight community and sleep researchers to design these questions. It asks caregivers about children’s sleep habits and the impact of health issues on sleep behaviors. The survey looks at children between ages 2 and 18 years of age. 28 children with STXBP1 are represented.

**Getting Enough Sleep**

- Always (10.71%)
- Usually (50%)
- Sometimes (17.86%)
- Rarely (17.86%)
- Never (3.57%)

**Children with STXBP1 fall asleep in 31 minutes on average.**

**Medical issues related to sleep problems in children with STXBP1**

- Breathing problems: 3
- Restless legs: 3
- Pain: 5
- GI symptoms: 6
- Sleep apnea/snoring: 1
- Movement disorder: 1
- Medication side effects: 1
- Seizures: 1

**Best Ways to Help Sleep: Community Recommendations**

- Bedtime routine or ritual: 10
- Parent lying in their bed until they fall asleep: 6
- Medication: 5
- Sound/white noise machine: 1

* Caregivers were able to select more than one answer.

Thank you for contributing knowledge to Simons Searchlight!