Simons Searchlight Registry Update STXBP1

June 2021

Ages in Years

Developmental & Behavioral Diagnoses

Notable conditions:
- Low Muscle Tone (57)
- Clumsy (57)
- High Muscle Tone (57)
- Large Head Size (57)
- Small Head Size (57)
- Movement Disorder (57)
- Tic Disorder (57)
- Cerebral Palsy (57)
- Cortical Blindness (57)
- Seizures (57)

Participation & Registration

This ever-growing database is made possible by the active participation of the STXBP1 community! Progress for individuals in your community with a STXBP1 variant is shown below -- log in to your simonssearchlight.org dashboard today to check for new surveys and other priority tasks. Your unique data could hold the clues scientists need to make progress and find answers.

**STEP 1**
Sign up online.

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

**STEP 3**
Schedule medical history phone call with a genetic counselor.

**STEP 4**
Fill out surveys.

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

**STEP 6**
We follow up with you over time.

**Notes:**
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.
- Information on reported seizure types is included on the 2021 quarter 1 registry report available on simonssearchlight.org/STXBP1.
The Child Behavior Checklist (CBCL) is a survey used to understand behavioral and emotional concerns in children.

The bars in the graphs show the overall level of concern reported by Simons Searchlight caregivers who completed the CBCL survey about their children with STXBP1. Individual caregivers may have indicated a lower or higher level of concern than indicated by the overall group.

**Behavioral Concerns for ages 1.5-5yrs**

- Oppositional behavior
- Attention/hyperactivity
- Aggression
- Anxiety
- Sadness

**Behavioral Concerns for ages 6-18 yrs**

- Oppositional behavior
- Attention/hyperactivity
- Aggression
- Anxiety
- Sadness

**Top Behavioral Concerns**

**Ages 1.5-5 yrs**

- Chews on non-food items
- Doesn't display fear of getting hurt
- Doesn't respond when spoken to
- Quickly moves from one activity to the next

**Top Behavioral Concerns**

**Ages 6-18 yrs**

- Hyperactive
- Impulsive
- Frequently distracted
- Obsessions