Simons Searchlight Registry Update STXBP1
April, 2022

Ages in Years

Developmental and Behavioral Conditions

Neurological Problems

Gastrointestinal Problems

Notes:
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.

How to Participate

The information in this report is made possible by the active participation of the STXBP1 community! Progress for individuals in your community with STXBP1 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.

<table>
<thead>
<tr>
<th>STEP 1</th>
<th>STEP 2</th>
<th>STEP 3</th>
<th>STEP 4</th>
<th>STEP 5</th>
<th>STEP 6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sign up online</td>
<td>Provide your genetic lab report</td>
<td>Share your important medical history</td>
<td>Fill out surveys</td>
<td>Provide a blood sample if you are interested</td>
<td>Update us every year</td>
</tr>
</tbody>
</table>

227 151 87 124 34  
Log in to see next steps
Information Spotlight: **Quality of Life**

The **Quality of Life Inventory** is a scientific survey where we asked caregivers to describe how their family member with STXB1 responds to life experiences.

It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

These graphs reflect the percentage of people who reported "Often" or "Very Often" for each question.

**Health & Well-being**

- **Has energy for daily activities**: 57%
- **Is alert and aware**: 71%
- **Is in good health**: 93%
- **Sleeps well**: 64%

**Feelings & Emotions**

- **In a good mood**: 75%
- **Smiles**: 50%
- **Expresses happiness using body language**: 61%
- **Laughs or giggles**: 46%
- **Unsettled for no reason**: 18%
- **Displays aggression**: 11%
- **Feels upset or angry**: 11%
- **Withdrawn, low mood**: 0%
- **Intentionally self harms**: 11%
- **Dislikes changes in routine**: 7%
- **Displays anxiety or agitation**: 14%
"Quality of life is a common survey of patient-centered outcomes in clinical trials and natural history studies."

- Alison Holbrook, PhD

### Daily Life
- Expresses their needs: 50%
- Makes own choices for activities: 14%
- Likes using technology: 29%
- Helps with routines: 11%
- Enjoys making things: 7%

### Activities & the Outdoors
- Enjoys moving their body: 68%
- Enjoys feeling steady during activities: 57%
- Enjoys physical activities: 71%
- Enjoys outings: 57%
- Enjoys time outdoors: 68%

### Family & Friends
- Happy when they are understood: 46%
- Feels relaxed while making eye contact: 71%
- Initiates greetings with others: 68%
- Happy to be included: 75%
- Enjoys socializing at mealtimes: 57%
- Enjoys interacting with others: 68%
- Excited by upcoming activities: 46%

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Thank you for contributing knowledge to Simons Searchlight!