Data in this report are from 9 participants with SETBP1. 2 females and 7 males are represented. Participants were between 3 and 20 years old. The average age was 9 years.

Symptoms Reported for People with SETBP1

Caregivers were asked to report all the current symptoms experienced by their family members with SETBP1. A list of 32 symptoms was provided. Bars in the graph show the number of people who currently experience the symptom.

Intellectual disability/delay 8
Difficulty with fine motor control 8
Limited or absent speech 7
Attention issues 6
Repetitive or rigid behavior 5
Aggression toward others 4
Irritability 4
Sensory issues 4
Not using toilet for bowel movements 3
Difficulty walking 3
Limited means of communication 3
Low muscle tone 3
Anxiety 3
Gastrointestinal problems 3
Mood swings 2
Hyperactivity 2
Difficulty with social interaction 2
Tantrums 2
Not using toilet for urinating 2
Pain or discomfort 2
Visual impairment 1
Hearing impairment 1
Epilepsy 1
High muscle tone 1
Uncontrolled movements 1
Self-injurious behavior 1
Picky eating, food aversions or refusal 1
Apnea 1

Note: People may be shown in more than one bar if they reported multiple symptoms.
If people reported having pain or discomfort as a symptom, they were asked to describe where they experienced it.

This graph shows the number of people who experienced each type of pain.

**Type of Pain or Discomfort**

<table>
<thead>
<tr>
<th>Type</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Head</td>
<td>1</td>
</tr>
<tr>
<td>Muscular</td>
<td>1</td>
</tr>
<tr>
<td>Limbs/joints</td>
<td>1</td>
</tr>
</tbody>
</table>

**Symptoms with the Greatest Impact**

Caregivers were asked to rank 3 symptoms that have the most impact on them or their family members with SETBP1. Below, we show the 3 symptoms that were reported to have the most impact.

1. Intellectual disability / delay
2. Limited or absent speech
3. Aggression toward others

**Symptoms with a High or Very High Impact**

Caregivers were asked to rate the impact for every symptom experienced by their family members with SETBP1. They rated the impact for each symptom from ‘no impact’ to ‘very high impact.’

This table shows the number of people with SETBP1 who are either highly impacted or very highly impacted by a given symptom.

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Number</th>
</tr>
</thead>
<tbody>
<tr>
<td>Limited or absent speech</td>
<td>6</td>
</tr>
<tr>
<td>Intellectual disability/delays</td>
<td>5</td>
</tr>
<tr>
<td>Attention issues</td>
<td>5</td>
</tr>
</tbody>
</table>

Notes:
- People may be shown in more than one row or category if they reported multiple symptoms.
- Symptoms that affected at least 5 or more people were included.
Most Challenging Daily Activities

Caregivers selected all the daily activities that are most challenging for their family members with SETBP1. Bars in the graph show the number of people with SETBP1 that have trouble with each daily activity.

- Independence in everyday care: 7
- Using speech/language to communicate: 6
- Engaging in learning: 5
- Using the toilet: 4
- Using their hands: 2
- Nonverbal communication: 2
- Attending school or having a job: 2
- Participating in community outings: 2
- Sitting up: 1

Barriers to Participating in Community Outings

If people reported difficulty participating in community outings, they were asked to describe the biggest barrier to participation.

This graph shows the most common barriers to participating in community outings.

Note: People may appear in more than one bar if they reported multiple challenges.
Important Targets for Drug Treatments

Caregivers were asked to rank 3 treatment targets that are most important to their family members with SETBP1. This graph shows the 3 treatment targets ranked as most important.

1. Improve learning and cognitive ability
2. Improve speech / language
3. Reduce repetitive or rigid behaviors

Simons Searchlight is grateful to all the caregivers representing their family members. This survey is an initial step to better understand the symptoms and challenges faced by your community. Your participation can help guide future research efforts to fully describe the needs and priorities of your community and plan for effective treatments.

Thank you for contributing knowledge to Simons Searchlight!