Simons Searchlight created this Voice of the Community survey to hear about the challenges that family members experience related to their genetic condition.

Researchers use Voice of the Community surveys to help inform assessment and treatment decisions from a patient-centered approach.

Data in this report are from **20 participants** with a 1q21.1 duplication. **5 females and 15 males** are represented.

Participants were between **6 and 17 years old**. The average age was **11 years**.

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Symptoms Reported for People with a 1q21.1 Duplication

Caregivers were asked to report all the current symptoms experienced by their family members with a 1q21.1 duplication. A list of 32 symptoms was provided. Bars in the graph show the number of people who currently experience the symptom.

- **Intellectual disability/delays**: 16
- **Picky eating, food aversions or refusal**: 14
- **Difficulty with social interaction**: 13
- **Attention issues**: 10
- **Repetitive or rigid behavior**: 10
- **Sensory issues**: 9
- **Difficulty with fine motor control**: 9
- **Anxiety**: 9
- **Hyperactivity**: 9
- **Sleep problems**: 8
- **Low muscle tone**: 8
- **Tantrums**: 8
- **Overeating or sneaking food**: 7
- **Limited or absent speech**: 7
- **Limited means of communication in...**: 7
- **Irritability**: 6
- **Mood swings**: 6
- **Aggression toward others**: 4
- **Not using toilet for bowel movements**: 4
- **Not using toilet for urinating**: 4
- **Visual impairment**: 2
- **Self-injurious behavior**: 2
- **Gastrointestinal problems**: 2
- **Respiratory/breathing problems**: 2
- **Apnea**: 2
- **Difficulty walking**: 1
- **Epilepsy**: 1
- **Uncontrolled movements**: 1
- **Pain or discomfort**: 1

Note: People may be shown in more than one bar if they reported multiple symptoms.
**Type of Pain or Discomfort**

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

**Symptoms with the Greatest Impact**

Caregivers were asked to rank 3 symptoms that have the most impact on them or their family members with a 1q21.1 duplication. Below, we show the 3 symptoms that were reported to have the most impact.

1. Intellectual disability / delay
2. Limited or absent speech
3. Picky eating

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

Caregivers were asked to rate the impact for every symptom experienced by their family members with a 1q21.1 duplication. They rated the impact for each symptom from 'no impact' to 'very high impact.'

This table shows the number of people with a 1q21.1 duplication who are either highly impacted or very highly impacted by a given symptom.

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Impact</th>
</tr>
</thead>
<tbody>
<tr>
<td>Attention issues</td>
<td>8</td>
</tr>
<tr>
<td>Intellectual disability/delays</td>
<td>7</td>
</tr>
<tr>
<td>Sensory issues</td>
<td>6</td>
</tr>
<tr>
<td>Limited or absent speech</td>
<td>6</td>
</tr>
<tr>
<td>Anxiety</td>
<td>6</td>
</tr>
<tr>
<td>Picky eating, food aversions or refusal</td>
<td>6</td>
</tr>
<tr>
<td>Hyperactivity</td>
<td>5</td>
</tr>
</tbody>
</table>

**Notes:**
- People may be shown in more than one row if they reported multiple symptoms.
- Symptoms that affected at least 5 or more people were included.
Caregivers selected all the daily activities that are most challenging for their family members with a 1q21.1 duplication. Bars in the graph show the number of people with A 1q21.1 duplication that have trouble with each daily activity.

### Most Challenging Daily Activities

- **Engaging in learning**: 13
- **Social interaction and participation**: 9
- **Nonverbal communication**: 7
- **Independence in everyday care**: 7
- **Regular sleep**: 7
- **Using their hands**: 6
- **Using speech/language to communicate**: 6
- **Attending school or having a job**: 6
- **Participating in community outings**: 6
- **Using the toilet**: 4
- **Walking**: 1
- **Sitting up**: 1
- **Feeding/eating**: 1

### Barriers to Participating in Community Outings

- **Anxiety**: 1
- **Sensory sensitivity**: 2
- **Behavior**: 3

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 a 1q21.1 duplication. This graph shows the 3 treatment targets ranked as most important.

1. Improve learning and cognitive ability
2. Improve eating or feeding
3. Improve sleep

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!