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 9 participants with a 16p11.2 distal duplication. 4 females and 5 males are represented.

Participants were between 5 and 16 years old. The average age was 11 years.

### Symptoms Reported for People with a 16p11.2 Distal Duplication

Caregivers were asked to report all the current symptoms experienced by their family members with a 16p11.2 distal duplication. A list of 32 symptoms was provided. Bars in the graph show the number of people who currently experience the symptom.

- Anxiety: 6
- Attention issues: 6
- Intellectual disability/delay: 5
- Difficulty with fine motor control: 5
- Hyperactivity: 5
- Picky eating, food aversions or refusal: 5
- Difficulty with social interaction: 4
- Sensory issues: 4
- Visual impairment: 3
- Self-injurious behavior: 3
- Irritability: 3
- Sleep problems: 2
- Low muscle tone: 2
- Mood swings: 2
- Tantrums: 2
- Repetitive or rigid behavior: 2
- Hearing impairment: 1
- Limited or absent speech: 1
- Limited means of communication: 1
- Aggression toward others: 1
- Gastrointestinal problems: 1
- Not using toilet for bowel movements: 1
- Not using toilet for urinating: 1
- Feeding problems: 1
- Overeating or sneaking food: 1
- Respiratory/breathing problems: 1

Note: People may be shown in more than one bar if they reported multiple symptoms.
Caregivers were asked to rank 3 symptoms that have the most impact on them or their family members with a 16p11.2 distal duplication. Below, we show the 3 symptoms that were reported to have the most impact.

1. Attention issues
2. Picky eating
3. Anxiety

Caregivers selected all the daily activities that are most challenging for their family members with a 16p11.2 distal duplication. Bars in the graph show the number of people that have trouble with each daily activity.

- Engaging in learning: 5
- Social interaction and participation: 4
- Feeding/eating: 3
- Using their hands: 2
- Using speech/language to communicate: 2
- Independence in everyday care: 2
- Regular sleep: 2
- Nonverbal communication: 1
- Using the toilet: 1
- Attending school or having a job: 1
- Participating in community outings: 1

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 be shown in more than one category if they reported more than one type of pain.
Important Targets for Drug Treatments

Caregivers were asked to rank 3 treatment targets that are most important to their family members with a 16p11.2 distal duplication. This graph shows the 3 treatment targets ranked as most important.

1. Improve learning and cognitive ability
2. Improve mood, irritability, or anxiety
3. Improve eating or feeding

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!