Data in this report are from 26 participants with Okur-Chung Syndrome (OCNDS), a change in the CSN2A1 gene. 18 females and 8 males are represented.

Participants were between 2 and 20 years old. The average age was 9 years.

Symptoms Reported for People with OCNDS

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

- Intellectual disability/delay: 25
- Difficulty with fine motor control: 20
- Low muscle tone: 17
- Attention issues: 16
- Gastrointestinal problems: 16
- Repetitive or rigid behavior: 15
- Sensory issues: 14
- Limited or absent speech: 12
- Anxiety: 12
- Difficulty with social interaction: 11
- Tantrums: 11
- Sleep problems: 11
- Difficulty walking: 10
- Irritability: 9
- Picky eating, food aversions or refusal: 9
- Visual impairment: 8
- Epilepsy: 8
- Mood swings: 8
- Limited means of communication: 7
- Hyperactivity: 7
- Aggression toward others: 6
- Not using toilet for urinating: 6
- Feeding problems: 5
- Self-injurious behavior: 5
- Not using toilet for bowel movements: 5
- Apnea: 4
- Uncontrolled movements: 4
- Hearing impairment: 3
- Respiratory/breathing problems: 3
- Pain or discomfort: 3
- Overeating or sneaking food: 2

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

- **Head**: 1
- **Eyes, ears, nose, throat**: 1
- **Dental**: 1
- **Chest**: 1
- **Muscular**: 2
- **Stomach/GI/Digestive**: 1
- **Limbs/joints**: 3

Note: People may be shown in more than one category if they reported more than one 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 OCNDS. Below, we show the 3 symptoms that were reported to have the most impact.

1. **Intellectual disability / delay**
2. **Limited or absent speech**
3. **Sleep**
Caregivers were asked to rate the impact for every symptom experienced by their family members with OCNDS. They rated the impact for each symptom from 'no impact' to 'very high impact.'

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

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Number of People</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intellectual disability, developmental delays, or learning delays</td>
<td>21</td>
</tr>
<tr>
<td>Attention issues</td>
<td>10</td>
</tr>
<tr>
<td>Sleep problems (getting to sleep or staying asleep)</td>
<td>9</td>
</tr>
<tr>
<td>Limited or absent speech</td>
<td>8</td>
</tr>
<tr>
<td>Anxiety</td>
<td>8</td>
</tr>
<tr>
<td>Sensory issues</td>
<td>7</td>
</tr>
<tr>
<td>Difficulty with fine motor control</td>
<td>7</td>
</tr>
<tr>
<td>Repetitive or rigid behaviors</td>
<td>6</td>
</tr>
<tr>
<td>Difficulty with social interaction</td>
<td>6</td>
</tr>
<tr>
<td>Picky eating, food aversions or refusal</td>
<td>6</td>
</tr>
<tr>
<td>Limited means of communication</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 OCNDS. Bars in the graph show the number of people with OCNDS that have trouble with each daily activity.

### Most Challenging Daily Activities

- Independence in everyday care: 18
- Engaging in learning: 15
- Using speech/language to communicate: 12
- Using the toilet: 9
- Regular sleep: 8
- Using their hands: 7
- Feeding/eating: 6
- Social interaction and participation: 6
- Participating in community outings: 5
- Walking: 4
- Nonverbal communication: 3
- Attending school or having a job: 3

### Barriers to Participating in Community Outings

- Mobility: 1
- Behavior: 1
- Sensory sensitivity: 1

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 OCNDS. This graph shows the 3 treatment targets ranked as most important.

1. Improve learning and cognitive ability
2. Improve speech / language
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!