### Purpose

The study aims to capture burden of disease experiences from persons with spinocerebellar ataxias (SCA) and their caregivers, to identify disease aspects that are most meaningful to them, and to augment the spinocerebellar ataxia (SCA) Burden of Illness (BOI) study incorporating the patient’s voice.

### Background

- **SCA** is a group of ultra-rare neurodegenerative diseases of the cerebellum and spinal cord that is characterized by progressively by unsteady gait, imbalance, and weakness of the arms and legs. (source: National Institute of Neurological Disorders and Stroke (NINDS), NIH, 2022)

### Methods

#### Participants & Recruitment

- Eligible participants were asked to complete the inclusion criteria and structured interviews in English, French, German, or Italian. A total of 80 participants, including 20 for each of the 4 SCA types, were recruited to complete Phase 2 of the study. (source: Schmahmann JD, Pierce S, MacMore J, L’Italien GJ. Development and validation of a patient-reported outcome measure for spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120).

#### Design

- **Phase 1**: SS QoL and modified Klockgether questionnaire for patient and caregiver. (source: Descriptive Human Subject Research: Application of Research: Intent to Treat (ITT) and the entire cohort of patients. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- **Phase 2**: SF-36® and modified Klockgether questionnaire for caregivers (source: Descriptive Human Subject Research: Application of Research: Intent to Treat (ITT) and the entire cohort of patients. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120).

#### Analysis

- **Phase 1**: Data analysis was performed on descriptive statistics and correlation analysis. (source: Descriptive Human Subject Research: Application of Research: Intent to Treat (ITT) and the entire cohort of patients. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- **Phase 2**: Data analysis was performed using descriptive statistics and correlation analysis. (source: Descriptive Human Subject Research: Application of Research: Intent to Treat (ITT) and the entire cohort of patients. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)

### Results

#### Descriptive Data of Study Participants

<table>
<thead>
<tr>
<th>SCA Type</th>
<th>Age (years)</th>
<th>Gender</th>
<th>Total Participants</th>
<th>Caregivers</th>
</tr>
</thead>
<tbody>
<tr>
<td>SCA1</td>
<td>45.5</td>
<td>72.3%</td>
<td>20</td>
<td>5</td>
</tr>
<tr>
<td>SCA2</td>
<td>52.6</td>
<td>73.5%</td>
<td>20</td>
<td>6</td>
</tr>
<tr>
<td>SCA3</td>
<td>64.5</td>
<td>76.7%</td>
<td>20</td>
<td>7</td>
</tr>
<tr>
<td>SCA6</td>
<td>74.3</td>
<td>82.4%</td>
<td>20</td>
<td>9</td>
</tr>
</tbody>
</table>

#### Desired Therapeutic Outcomes

- **Most Important Disease Impact**: Speech (42.2% of points), gross motor function (13.9% of points), and independent living (12.3% of points). (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- **Most Important Disease Burden Category Among Persons With SCA**: Communication (42.2% of points), gross motor function (13.9% of points), and independent living (12.3% of points). (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)

### Acknowledgments

This research was supported by the National Institute of Neurological Disorders and Stroke (NINDS), the Ataxia Foundation, the International Fibrodysplasia Ossificans Progressiva Association, KrabbeConnect, National Niemann-Pick Disease Foundation, and the International Ataxia Research Organization. (source: Presented at World Orphan Drug Congress, USA • May 23-26, 2022 International Congress of Ataxia Research.)

### Conclusions

- SCAs are a dominantly inherited group of ultra-rare diseases in which individual experience progressive cerebellar ataxia and associated symptoms that worsen over time. (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- Data from the global, cross-sectional, mixed-methods study involving persons with SCA highlights the significant burden that gross motor challenges (including loss of ambulatory ability), impaired mobility, and lack of balance, issues related to independent living, and lack of support, are on all types of SCA. (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- In all SCA types and in both phases of the study, mean summary scores on physical components of SF-36® were numerically lower than the general population and worst in SCA3. (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- This study captures burden of disease experiences of PWSCA and a small number of caregivers to identify disease aspects that are most meaningful to PWSCA, with the goal of capturing their voices. (source: Jackson SJ, AL, PE: Pharmacological and non-pharmacological interventions for the treatment of spinocerebellar ataxia. J Neurol Neurosurg Psychiatry. 2011;82(10):1114-1120)
- Many participants indicated they could accept their current limitations if a remedy capable of stopping disease progression was available. (source: The study was designed to capture burden of disease experiences from persons with spinocerebellar ataxias (PWSCA) and their caregivers).