

# Assessment of Caregiver Impact in SLC13A5 citrate transporter disorder

Sydney Cooper, MS<sup>1,2</sup>, Tanya Brown, PhD<sup>2</sup>, Kimberly Nye<sup>2</sup>, Kimberly Goodspeed, MD<sup>1</sup>

1 Department of Pediatrics, University of Texas Southwestern Medical Center, Dallas, TX, USA; 2 TESS Research Foundation, CA, USA



## Background

An online survey was administered to caregivers of individuals with SLC13A5 citrate transporter disorder to assess its impact on affected patients and their loved ones. The results of these assessments show that SLC13A5 citrate transporter disorder has a significant negative and dynamic impact on caregivers.

## Rationale

- Caring for a loved one affected by a rare disease is known to negatively affect sleep, mental health, physical health, and overall quality of life<sup>1,2</sup>.
- This disease is caused by biallelic loss of function variants in *SLC13A5*, which encodes a citrate transporter<sup>3,4</sup>.
- Patients present with severe infantile epilepsy with multifocal convulsive and subclinical seizures in the first week of life, followed by variable seizure burden<sup>5</sup>, developmental disability<sup>3</sup>, and tooth abnormalities<sup>6</sup>.
- The quality of life of affected patients and their caregivers is of interest for the design of treatments and their clinical trials<sup>7</sup>.

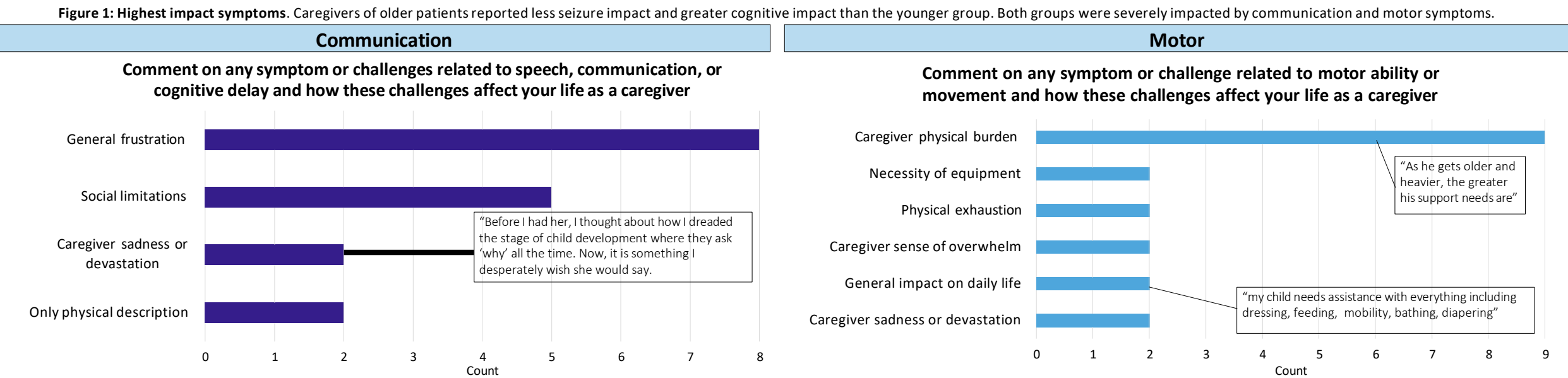
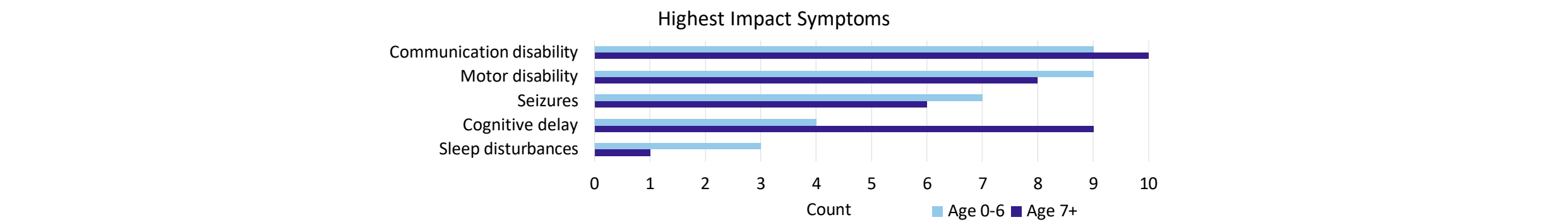
## Study Objectives

- To identify the most impactful symptoms of SLC13A5 citrate transporter disorder from the caregiver perspective
- To characterize disease burden for caregivers

## Methods

- Cross-sectional data was collected using an online survey distributed to the TESS Research Foundation Family Network for caregivers to complete.
- A total of 27 responses were recorded by 25 caregivers.
- Historical data and demographics, symptom prevalence and severity, disease management details, and the severity of impact directly from symptoms and indirectly from disease management were assessed.
- Qualitative themes were determined and each free response answer had themes assigned by two independent reviewers.
- A consensus of themes for each response was determined by the two reviewers together.

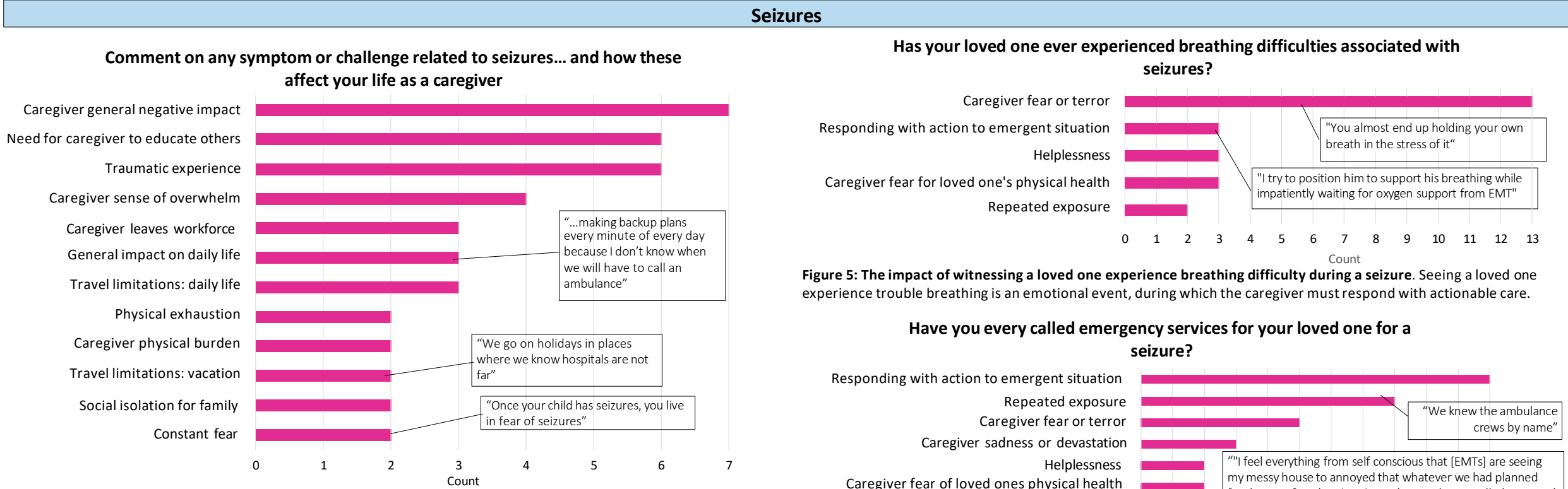
## Results



**Figure 1: Highest impact symptoms.** Caregivers of older patients reported less seizure impact and greater cognitive impact than the younger group. Both groups were severely impacted by communication and motor symptoms.

**Figure 2: The effects of cognitive delay and communication symptoms.** Caregivers share a general sense of frustration in response to cognitive and communication challenges. This symptom leads to social limitations for both the affected loved one and their caregiver.

**Figure 3: The effects of motor challenges.** The impact of motor symptoms on caregivers is extensive in its physical effects (i.e. necessity of equipment and support in daily activities) as well as emotional effects.



**Figure 4: The effects of seizures on caregivers.** Caregivers most frequently share that symptoms related to seizures have a general negative impact. Caregivers share multiple areas of life that are impacted, including limitations on the ability to work or leaving the workforce, travel limitations for daily activities or vacations, and a general sense of social isolation for whole families. Caregivers share a general sense of overwhelm.

**Figure 5: The impact of witnessing a loved one experience breathing difficulty during a seizure.** Seeing a loved one experience trouble breathing is an emotional event, during which the caregiver must respond with actionable care.

**Figure 6: The impact of a seizure severe that requires emergency services..** Such an event requires an urgent response and is a frequent occurrence for many caregivers. The resulting feeling is negative, including helplessness, sadness, and fear that can last well after the event.

## Summary and next steps

- Caregivers report significant negative emotional impact from the communication, cognition, motor, and seizure symptoms.
- The highest impact symptoms change as patients age.
- Motor, communication, and cognitive disabilities negatively impact families' QoL.
- Lived experience experts value a broad spectrum of clinical outcomes, demonstrating the importance of including their perspective in research.
- Next steps: analyze free responses and pair with quantitative responses to demonstrate how SLC13A5 citrate transporter disorder impacts families over time.

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- For any questions or comments, please contact [Sydney@tessfoundation.org](mailto:Sydney@tessfoundation.org).