RESULTS FROM ONLINE SURVEY WITH CAREGIVERS OF CHILDREN AND ADULTS WITH CREATINE TRANSPORTER DEFICIENCY (CTD)

- 37 caregivers completed the survey for 40 children with CTD between May 2019 and July 2020.
- The majority of children (n=27, 66%) were male.
- Children were on average around 10 years old (range: 1 - 24 years old).

DIAGNOSIS AND DISEASE MANAGEMENT

- 8 specialists were typically seen before patients received a CTD diagnosis.
- Label children were diagnosed at around 6 years old.
- Patients were most often diagnosed at or before age 1 year.
- Gene mutation sequencing (to confirm mutation in GLUT9 gene) (76%) and Urine test of creatine/creatinine (50%) were the most commonly reported methods.
- Gene mutation sequencing (to confirm mutation in SL6A8 gene) (75%) was the most frequently reported initial symptom.

SYMPTOMS AND IMPACTS

- More than half of the caregivers reported that their child had experienced at least 1 seizure (n=21, 53%).
- Caregivers reported that their child had experienced their first seizure at approximately 4.5 years old.
- 15 caregivers (54%) reported that their child currently experiences seizures.
- Many caregivers (n=20, 71%) reported that their child was currently taking AEDs, of which most (n=16, 80%) reported that these were effective at stopping seizures.

- On average, children said their first word at around 3 years old.
- Children were on average around 10 years old (range: 1 - 24 years old).
- 37 caregivers completed the survey for 40 children with CTD between May 2019 and July 2020.

- Approximately half of the patients (n=21, 53%) were taking supplements for CTD management at the time of survey completion; Creatine and Arginine were the most common supplements reported.
- The most commonly reported medications were for the treatment of seizures, sleep issues, constipation, allergies, appetite, gastroesophageal reflux disease (GERD), and nausea.

Communication

- Most common communication milestone reached by children (n=23, 58%): Electronic device/touch-chat.
- Communication milestones reached:
  - First words: 2 years old
  - First sat up without support: 1 year old
  - First unassisted steps: 2 years old

Behavior

- Caregiver-reported aspects of CTD that interfere most with child’s life:
  - Speech issues (33%)
  - Non-verbal communication issues (62%)
  - Behavior issues (e.g., hyperactivity, impulsivity, aggression) (35%)
  - Seizures (55%)

Motor functioning

- Average age of caregivers who reported their child reaching motor milestones:
  - First setup without support: 0.5 years old
  - First crawling: 1 year old
  - First unassisted steps: 2 years old

- Gross and Fine Motor Skills Achieved:
  - Toes touch floor
  - Pull up onto forearms
  - Sit independently
  - Stand with support

Caregiver assistance

- Impact of CTD on Caregiver Employment
  - 2 caregivers reported changing jobs due to their child’s health.
  - 6 caregivers reported having decreased hours.
  - 7 caregivers reported quitting their job.

Caregivers most commonly agreed with the following statement regarding emotional impacts:

- Feel stressed between caring for your child and trying to meet other responsibilities for your family/work.

Thank you to the Association for Creatine Deficiencies (ACD) for assisting with recruitment for this study.

We would also like to recognize and thank all of the caregivers who participated in this study.

**Impact of CTD on Caregiver Employment**

- 13 caregivers (34%) reported that their employment status changed as a result of their child’s health.
- 1 caregiver reported changing their job.
- 6 caregivers reported having decreased hours.
- 7 caregivers reported quitting their job due to their child’s health.