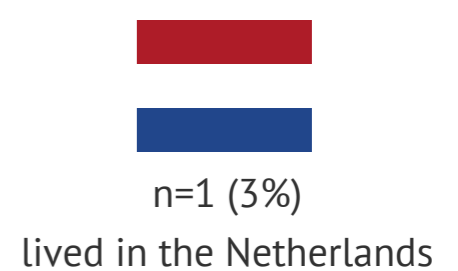
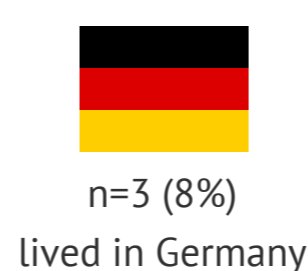
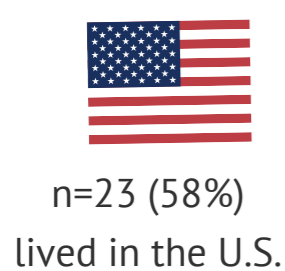


RESULTS FROM ONLINE SURVEY WITH CAREGIVERS OF CHILDREN AND ADULTS WITH CREATINE TRANSPORTER DEFICIENCY (CTD)

- 37 caregivers completed the survey for 40 children with CTD between May 2019 and July 2020.
- The majority of children (n=37, 93%) were male.
- Children were on average around 10 years old (range: 1 - 24 years old).



DIAGNOSIS AND DISEASE MANAGEMENT

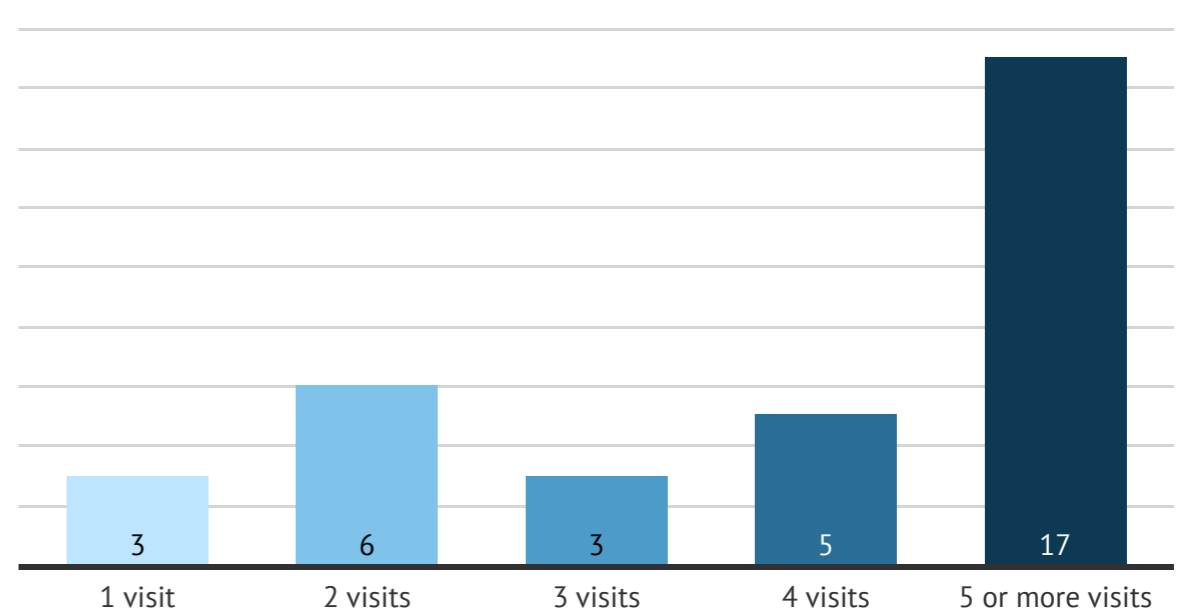
- 6 specialists were typically seen before patients received a CTD diagnosis.
- Most children were diagnosed at around 6 years old.
- Patients were most often diagnosed via:
 1. Gene mutation sequencing (to confirm mutation in SL6A8 gene) (75%)
 2. Urine test of creatine/creatinine (60%)
 3. Magnetic resonance spectroscopy (MRS) for brain image (35%)

Most children began displaying symptoms at around 1 year old.

Most frequently reported initial symptoms:

1. Delayed crawling or walking
2. Speech delay or impairments
3. Global developmental delay

Total Number of ER/Hospital Visits



*Total number of visits per patient; majority of hospitalizations were for seizures.

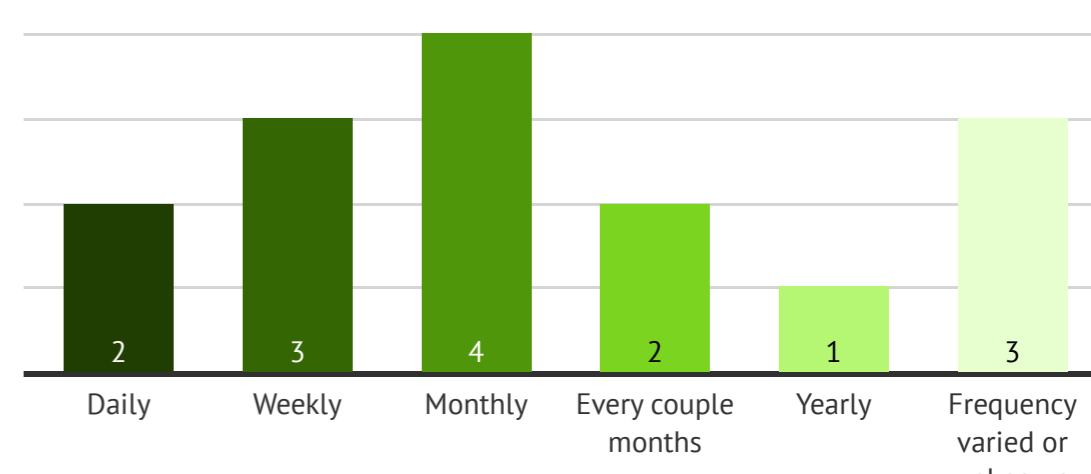
- Approximately half of the patients (n=21, 53%) were taking supplements for CTD management at the time of survey completion; Creatine and Arginine were the most common supplements reported.
- The most commonly reported medications were for the treatment of seizures, sleep issues, constipation, allergies,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

SYMPTOMS AND IMPACTS

Seizures

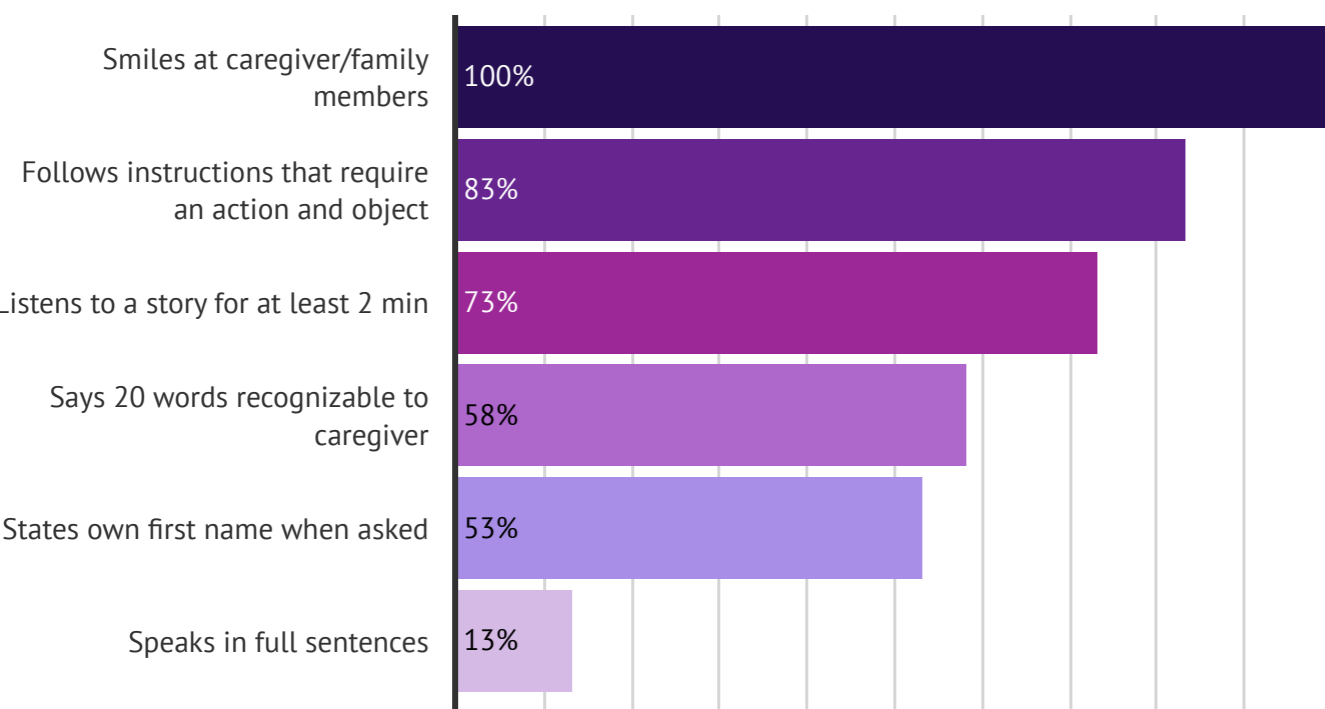
- More than half of the caregivers reported that their child had experienced at least 1 seizure (n=28, 70%).
- Caregivers reported that their child had experienced their first seizure at approximately 4.5 years old.
- 15 caregivers (54%) responded that their child currently experiences seizures.
- Many caregivers (n=20, 71%) responded that their child was currently taking AEDs, of which most (n=16, 80%) reported that these were effective at stopping seizures.

Seizure Frequency



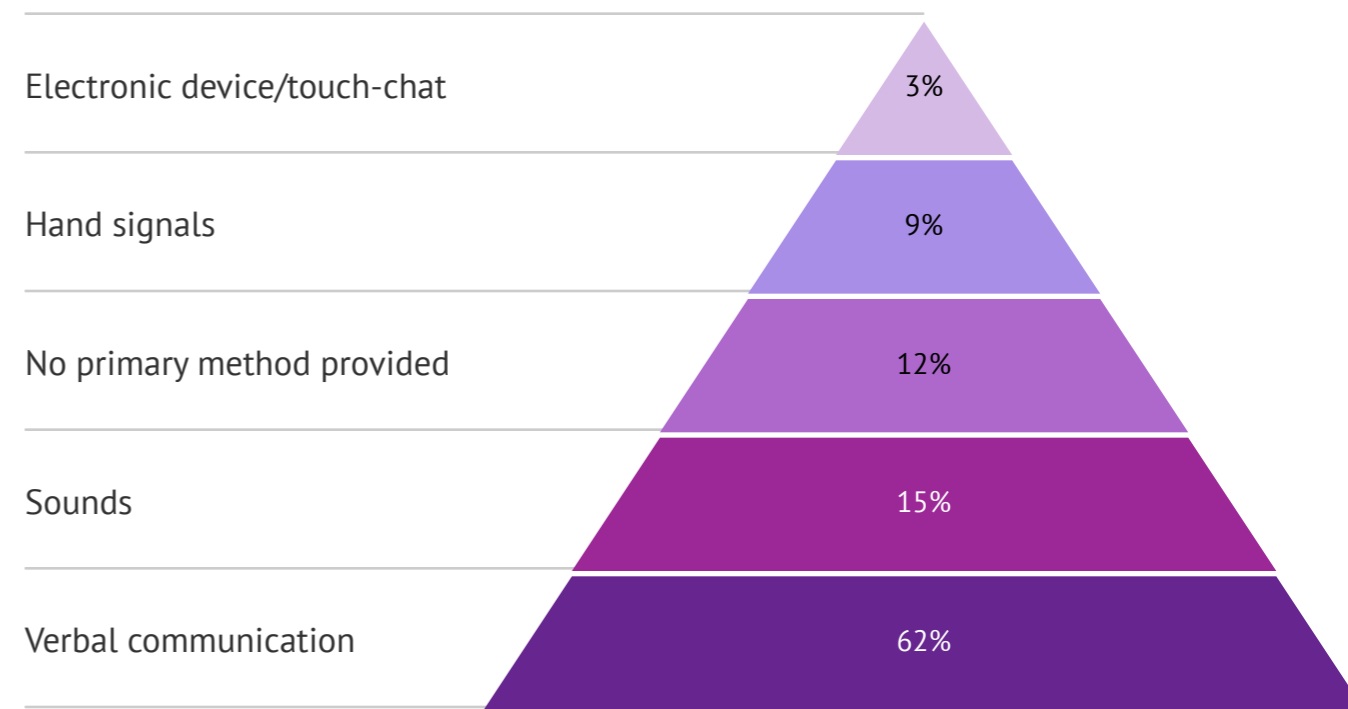
Communication

Communication Milestones Reached



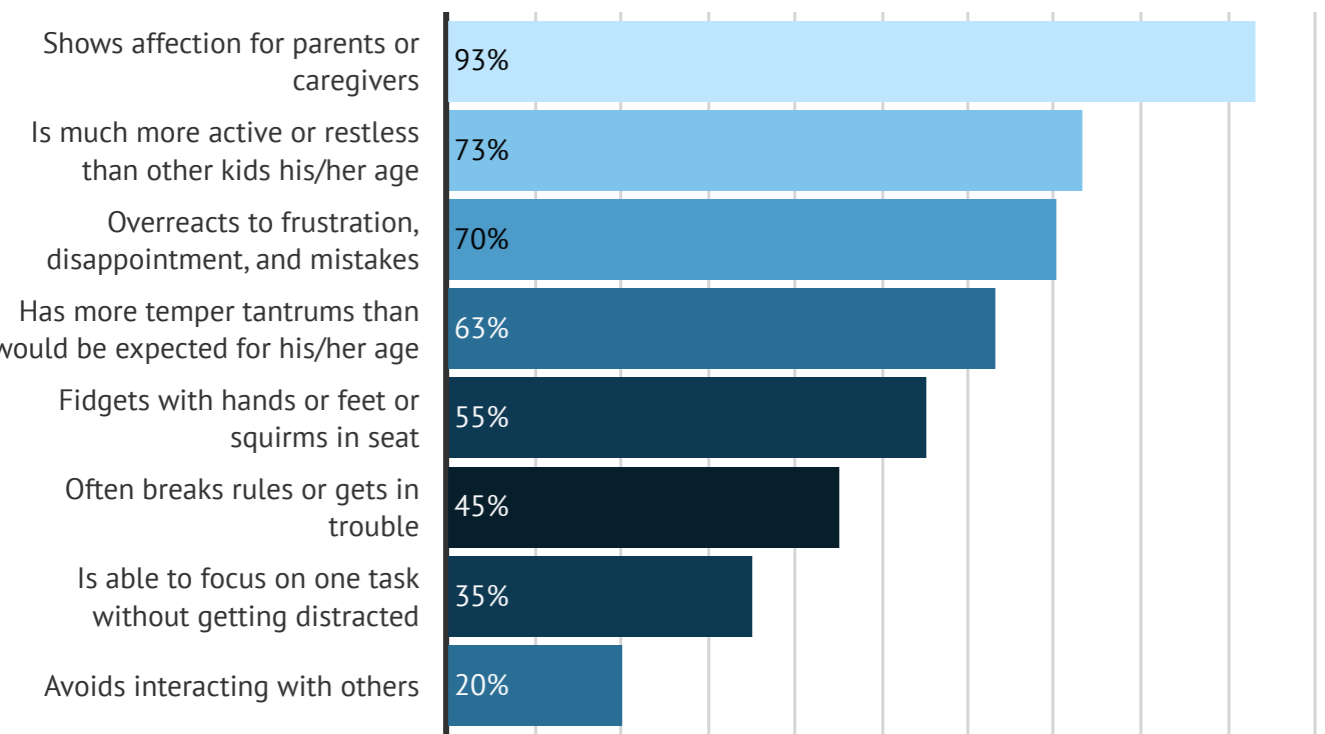
On average, children said their first word at around 3 years old.

Primary Communication Methods



Behavior

Caregiver-Observed Behaviors



Caregiver-reported aspects of CTD that interfere most with child's life

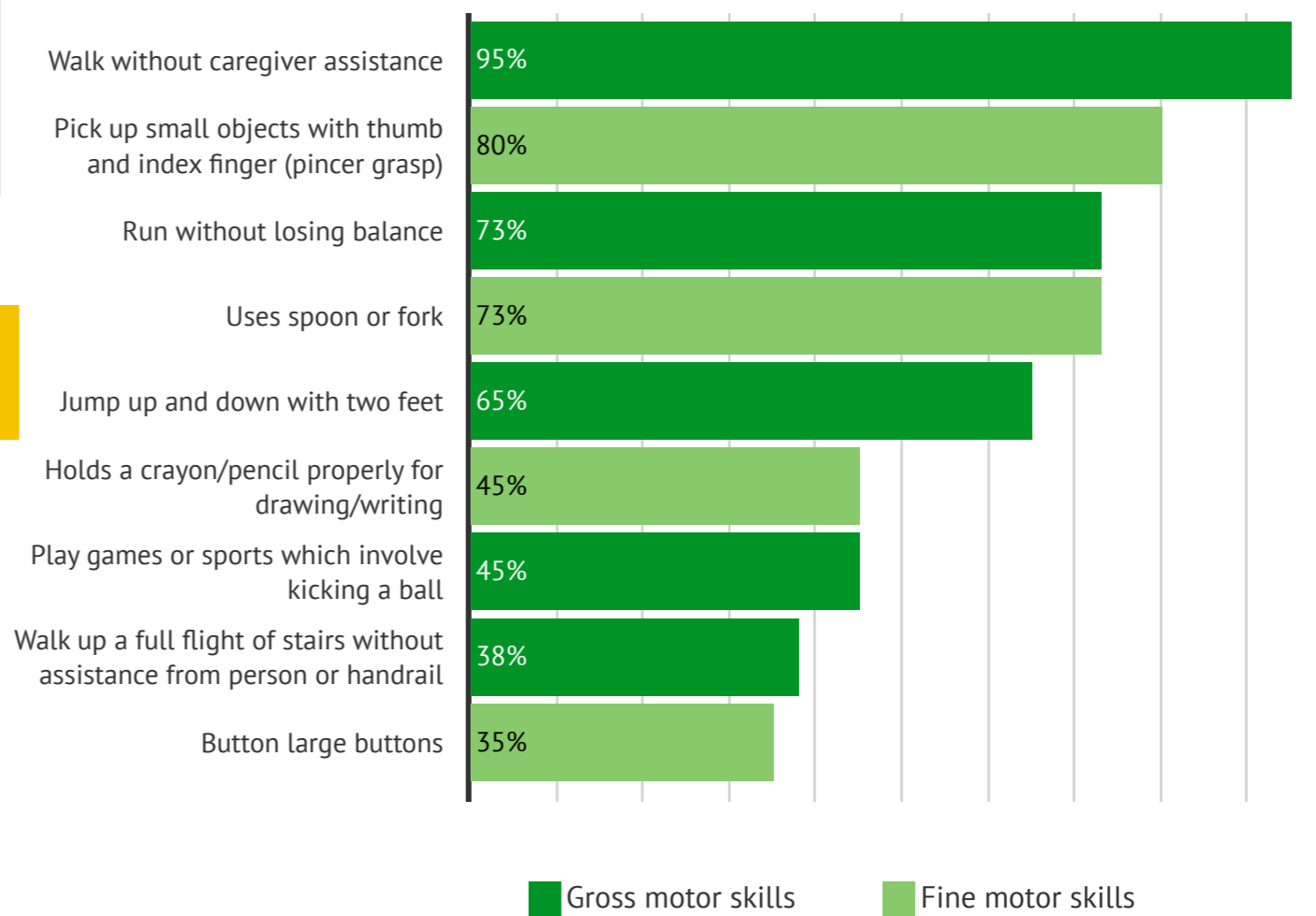
- Speech issues (35%)
- Non-verbal communication issues (18%)
- Behavior issues (e.g., hyperactivity, impulsivity, aggression) (13%)
- Seizures (13%)

Motor functioning

Average age caregivers reported their child reaching motor milestones:

- First sat up without support - 1 year old
- First started crawling - 1 year old
- First unassisted steps - 2 years old

Gross and Fine Motor Skills Achieved

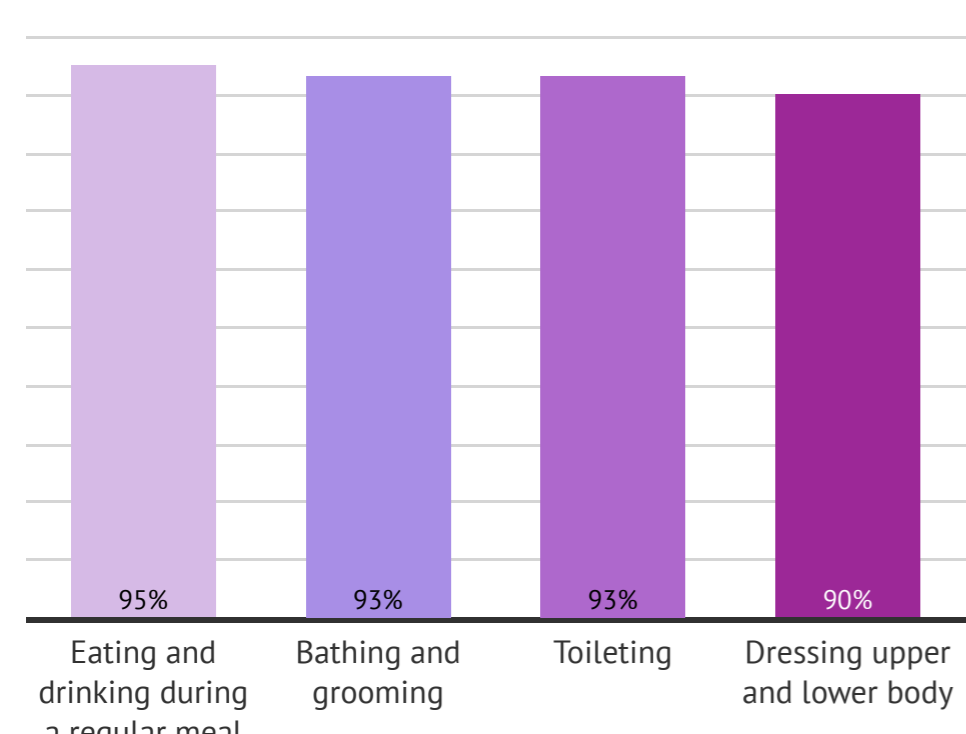


CAREGIVER ASSISTANCE

Most frequently reported self-care activities that children could complete:

- Drink from cup unassisted (73%)
- Demonstrate an understanding that hot things are dangerous (65%)
- Ask to use the toilet (45%)

Activities Requiring Assistance



Impact of CTD on Caregiver Employment

- 15 caregivers (38%) reported that their employment status changed as a result of their child's health.
 - 7 caregivers reported quitting their job
 - 6 caregivers reported having decreased hours
 - 2 caregivers reported changing jobs due to their child's health.

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.