Impacts of Disruptive Seizures, Sleep, and Behaviors on Activities of Daily Living and Communication in Developmental and Epileptic Encephalopathies: Interim Results of a Caregiver Survey

Poster 2.318

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Background

- Developmental and epileptic encephalopathies (DEEs) are characterized by severe and frequent seizures and developmental delays[1]
- People who live with DEE also have difficulty with communication and primarily depend on caregivers for activities of daily living (ADLs), which include feeding, bathing and hygiene, dressing, and toileting[2-5]
- Available literature on the impact of disruptive seizures, sleep, and/or behavior on the ability to communicate and to independently perform ADLs is lacking

Objective

- To assess caregiver-reported ADL support needs and methods of communication in people with DEE
- To explore observations of "normal" and "disruptive" seizures, sleep, and behavior and their perceived effects on ADLs and communication

Methods

- An internet-based 63-question anonymous survey was co-developed in consultation with the Dravet syndrome (DS) and Lennox-Gastaut Syndrome (LGS) communities
  - The survey was actively distributed by the CACNA1A Foundation, Dravet Syndrome (DS) Foundation, Dup15q Alliance, International Foundation for CDKL5 Research, KCNT1 Epilepsy Foundation, Lennox-Gastaut Syndrome (LGS) Foundation, PCDH19 Alliance, SLC6A1 Connect, STXBP1 Foundation, CURE SYNGAP1, and Tuberous Sclerosis Complex (TSC) Alliance to caregivers of people with DEE
- Caregivers responded to questions about 5 key domains (seizures, sleep, behavior, ADLs, and communication) and quality of life (QoL) to:
  - Define "normal" (typical daily experience during the current phase of the DEE journey) and disruptive (deviations from the typical experience) seizures, sleep, and behavior
  - Explore their perceived effect on ADLs and communication, and overall impact on QoL
- The current analysis summarizes data specific to ADLs and communication, including the impact of disruptive seizures, sleep, and behavior on these domains

Results

- In total, 489/524 survey responses were included (**Table 1**); 35 were excluded for various reasons

Table 1. Caregiver-Reported Demographics of People With DEE

	People With DEE N=489
Age at diagnosis (y), median (range)	3 (0–64)
Age at time of survey (y), median (range)	8 (0–67)
Most common primary DEE diagnoses, n (%)	
LGS	67 (13.7)
SLC6A1	67 (13.7)
STXBP1	64 (13.1)
CACNA1A	54 (11.0)
DS	54 (11.0)
SYNGAP1	48 (9.8)
KCNT1	43 (8.8)
Secondary DEE diagnoses[a], n (%)	84 (17.2)
LGS	58/84 (69.0)
DS	2/84 (2.4)
Gastrostomy tube, n (%)	92 (18.8)
Age, y; median (range)	9 (0.8–30.0)
Diapers used at all times[b], n (%)	302 (61.8)
Age (y); median (range)	7 (0.2–67.0)
Age >2 y; n (%)	252/302 (83.4)

[a]Secondary DEE diagnoses are defined as an additional DEE diagnosis as reported by the caregiver. Most common secondary DEE diagnoses are listed; all other secondary diagnoses were reported once.
[b]Diapers used at all times was defined in the survey as "24/7".
DEE, developmental and epileptic encephalopathy; DS, Dravet syndrome; LGS, Lennox-Gastaut syndrome.

Overview

QUESTION

- To what extent do people with DEE require support in performing ADLs (feeding, bathing/hygiene, dressing, and toileting)?
- What methods of communication are used by people with DEE?
- Is there a relationship between caregiver-defined disruptive symptoms experienced by a person with DEE and changes in their ability to perform ADLs and/or communicate?

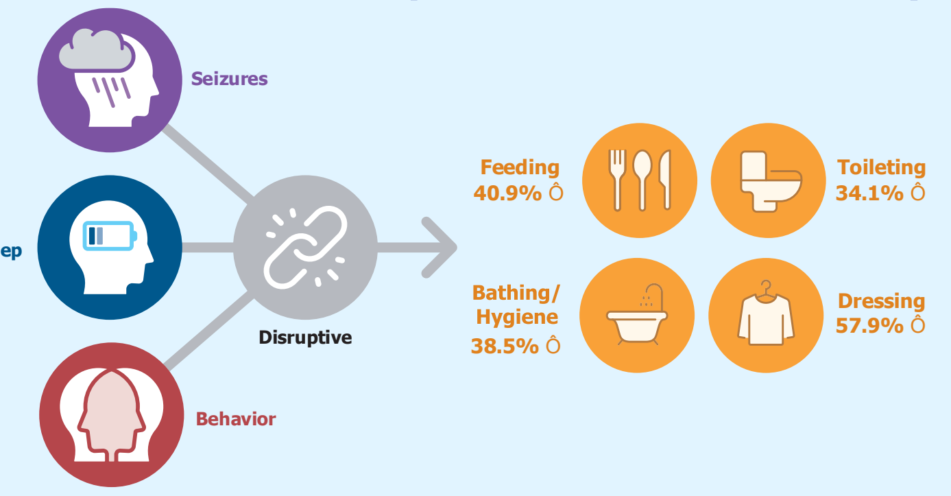
INVESTIGATION

- An internet-based anonymous survey was distributed to primary caregivers of a person diagnosed with DEE
- Caregivers defined "normal" (typical) and disruptive symptoms for seizures, sleep, and/or behavior and which, if any, affect the ability of the person with DEE to perform ADLs and/or communicate

RESULTS

Disruptive seizures, sleep, and/or behavior reportedly impacted the ability to perform an ADL in 22.1%–60.1% of people with DEE with any level of ability to perform the ADLs independently (N=489)

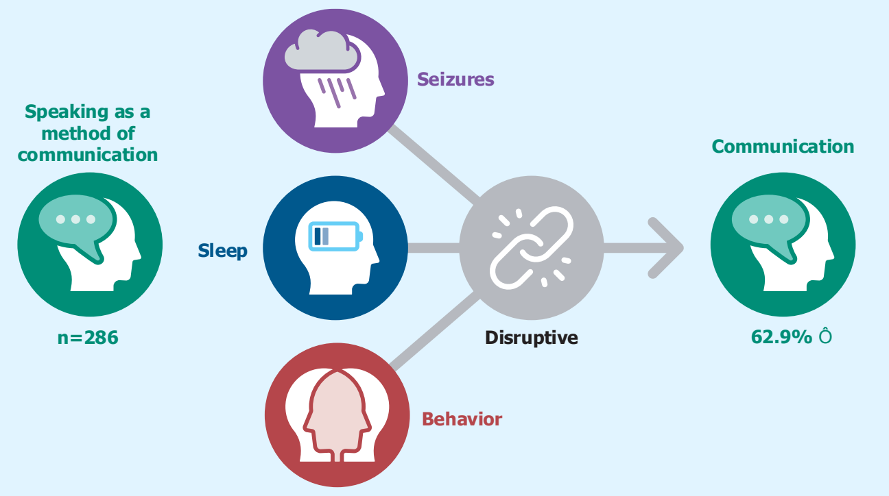
Temporary Loss in Ability to Perform an ADL in People With DEE Who Normally Have Some Independence (N=489)



People with DEE who use a g-tube are not included in "feeding"; people with DEE who use a diaper 24/7 are not included in "toileting". Feeding: 106/268; Bathing/Hygiene: 37/96; Dressing: 61/145; Toileting: 47/138.

The ability to communicate by any reported method was temporarily affected by disruptive seizures, sleep, and/or behavior in 297 (60.7%) people with DEE (N=489)

Temporary Loss in Ability to Communicate in People With DEE Who Speak as a Method of Communication (n=286)



CONCLUSIONS

- Disruptions in daily life in people living with DEE affect abilities to perform ADLs to various degrees
  - Disruptive seizures, sleep, and/or behavior were reported as impacting at least one of the 4 ADLs and/or communication by 344 (70.3%) caregivers
  - Within each ADL, impacts were reported at similar rates regardless of typical ability to independently perform that ADL
- These disruptions likely affect quality of life for people with DEE, their caregivers, and their families
- Further assessments of the impact of disruptive seizure, sleep and behavior symptoms on quality of life, via their effect on ADLs and communication, are warranted

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Abbreviations: ADL, activity of daily living; DEE, developmental and epileptic encephalopathy.

- Most people with DEE require support on at least one ADL (**Table 2**)
- The median age of people with DEE who never require support on ADLs was greater than the median age of those who always require support

Table 2. Median Age of People With DEE Always or Never Requiring Support on ADLs

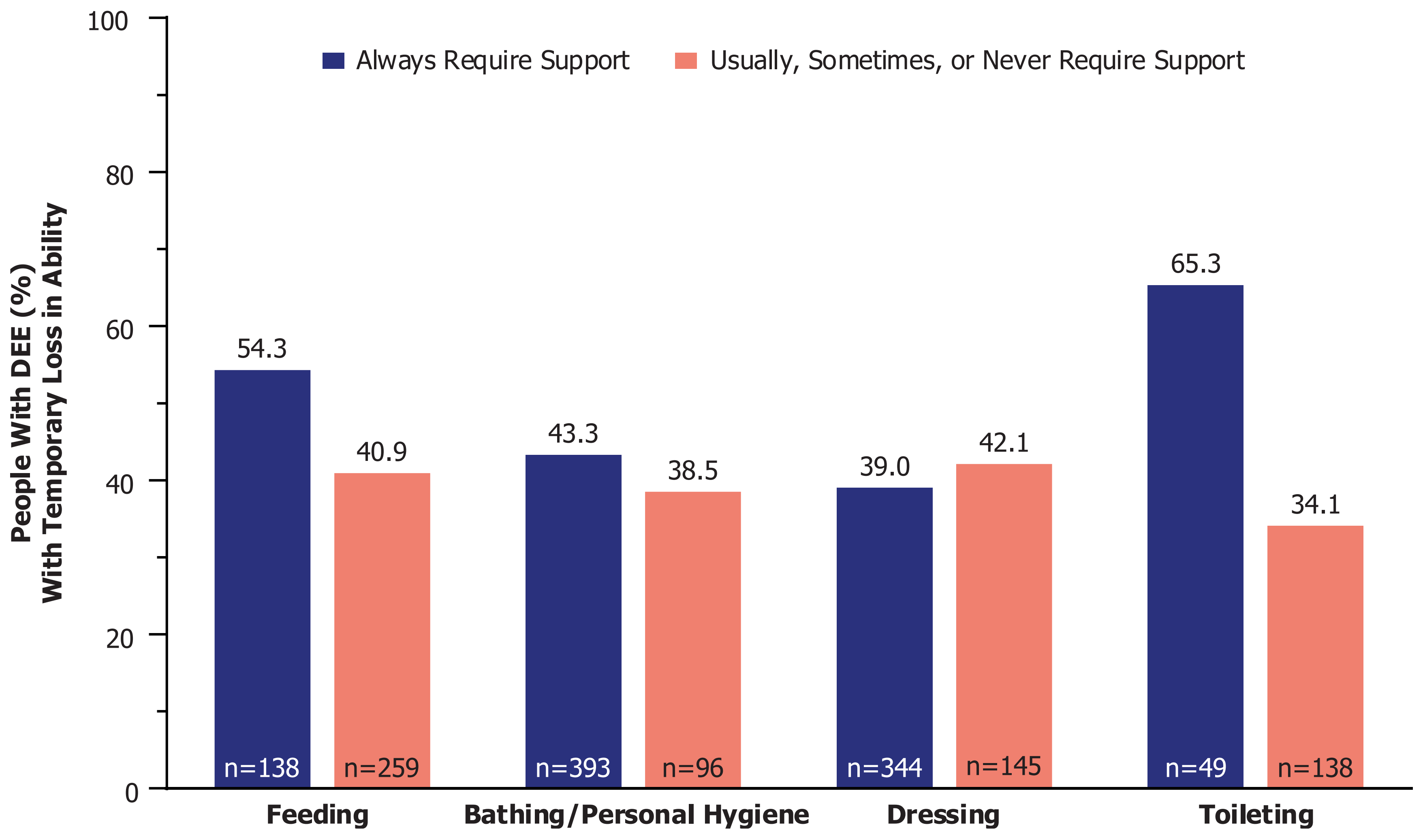
	n (%)	Age, years Median (range)
Always Require Support		
All ADLs	210 (42.9)	7 (0.2–67.0)
Feeding[a]	138/397 (34.8)	6 (0.2–67.0)
Bathing/Hygiene	393 (80.4)	8 (0.2–67.0)
Dressing	344 (70.3)	7 (0.2–67.0)
Toileting[b]	49/187 (26.2)	11 (0.2–44.0)
Never Require Support		
All ADLs	14 (2.9)	15 (0.8–26.0)
Feeding[a]	97/397 (24.4)	11 (0.7–54.0)
Bathing/Hygiene	18 (3.7)	16 (0.8–35.0)
Dressing	31 (6.3)	17 (0.8–54.0)
Toileting[b]	50/187 (26.7)	14 (0.8–37.0)

[a]Includes only people with DEE who do not use a G-tube. [b]Includes only people with DEE who do not require diapers 24/7.
ADL, activity of daily living; DEE, developmental and epileptic encephalopathy; G-tube, gastrostomy tube.

ACTIVITIES OF DAILY LIVING

- Of caregivers to people with DEE who did not always require support, 47/153 (30.7% [toileting, with or without diapering 24/7] to 61/145 (42.1% [dressing]) reported a temporary loss in ability to perform ADLs due to disruptive seizures, sleep, and/or behavior (**Figure 1**)
- Among people with DEE who did not use a G-tube, 181/397 (45.6%) experienced a temporary loss in feeding ability related to disruptive symptoms
- Among people with DEE who did not require diapers, 79/187 (42.2%) experienced temporary loss in toileting ability related to disruptive symptoms

Figure 1. Temporary Loss in Ability to Perform ADLs Due to Disruptive Seizures, Sleep, and/or Behavior in People With DEE By Level of Support Required



The percentage of people with temporary loss in ability within each subgroup is presented above each bar. The total number of people within each subgroup is presented at the bottom of each bar.
People with DEE who use a G-tube were not included in the feeding analyses. People with DEE who require diapers 24/7 were not included in the toileting analyses.
ADL, activity of daily living; DEE, developmental and epileptic encephalopathy; G-tube, gastrostomy tube.

COMMUNICATION

- In total, 286 (58.5%) people with DEE communicate by speaking (**Table 3**)
  - There were 45 caregivers (9.2%) who reported that they were unsure of how the person with DEE communicates; 21 of these did not report any method of communication

Table 3. Caregiver-Reported Age for Methods of Communication in People With DEE and Milestones in Neurotypical Children

	People With DEE, n (%)	Age (y), Median (range)	Typical Age Range, (y)[6,7]
Speaks	286 (58.5)	10 (0.9–67)	
Single words	64 (13.1)	7 (1–27)	1–2
3–5 words	44 (9.0)	7.5 (2–43)	3–4
Short sentences	92 (18.8)	12 (3–67)	4–5
Long sentences or fluently	86 (17.6)	11 (0.9–42)	4–5
Eye contact, facial expressions[a]	142 (29.0)	7 (0.2–54)	0–0.6
Gestures, signs, points, uses hands[a]	84 (17.2)	8 (0.2–54)	0.6–1
Writes, draws, uses equipment[a]	54 (11.0)	10 (0.5–34)	N/A

[a]Caregivers were able to select more than one option on the survey. Categories are inclusive of each other; Speaking methods are exclusive of all other categories.

- Communication was affected by disruptive seizures, sleep, and/or behavior in 297 (60.7%) people with DEE, overall
  - 180/286 (62.9%) people with DEE who communicate by speaking were affected

Conclusions

- People with DEE often require support in performing ADLs and have severe communication challenges, regardless of age
  - Additional loss in ability to perform ADLs due to disruptive symptoms was reported by over a quarter of caregivers for each ADL
  - While approximately half of caregivers reported that speaking was at least one method of communication, 62.9% of these caregivers reported a temporary loss in ability to communicate due to disruptive symptoms
- Improving QoL remains a top priority for individuals with DEEs and their families
  - Higher QoL has been reported when symptoms are predictable[8]
  - Disruptions are challenging, affecting not only their ability to perform daily tasks and/or communicate, but also reducing stability
- Further examinations of normal and disruptive seizures, sleep, and behavior and their relationships with ADLs and communication in people with DEE are warranted

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