The Burden of Spinal Muscular Atrophy Type 2 on Caregivers in Japan, the United Kingdom, and the United States: Results of a Global Survey

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Introduction

- SMA is a rare, genetic, progressive neuromuscular disorder caused by biallelic deletion or mutation of the *SMN1* gene. The phenotypic presentation of SMA ranges from profound weakness at birth to milder, more slowly progressive symptoms with adult onset, but all SMA types are debilitating if untreated.[1–4]
- Patients with SMA type 2 experience relatively rapid neuromuscular decline until 13 years of age and more gradual decline through adulthood. Patients with SMA type 2 can sit independently but do not stand or walk.[4–6]
- SMA is associated with substantial costs,[7] as well as physical and emotional burdens for patients, families, caregivers, and society[8]
  - Direct costs include inpatient, outpatient, and emergency care; medications; medical devices and mobility aids; respiratory and ventilatory assistance; and transportation[9–11]
  - Indirect costs and societal burdens include loss of productivity, time spent caregiving, anxiety and stress related to caregiving, and changes in employment status for the caregiver[9–11]
  - SMA is associated with greater medical expenses and HCRU than other complex chronic conditions[3]
  - Medical expenses and coverage for SMA vary from country to country, and caregiver burden around the world is largely unknown

Objective

- We sought to describe out-of-pocket expenditures and time costs, as well as HCRU, for the families and caregivers of patients with SMA type 2
  - The goal of this ongoing study is to assess burdens globally
  - Data presented here reflect caregiver burden in Japan, the UK, and the US

Methods

- Online surveys were disseminated through patient advocacy groups (Cure SMA, Japan Network for SMA, Muscular Dystrophy, ACT for SMA, TreatSMA, and SMA UK) and external recruitment companies from July 2020 through December 2021 across Japan, the UK, and the US
- Eligible respondents were voluntary non–health care professional adult caregivers. Caregivers of patients who lived permanently at stationary facilities (e.g., nursing homes or hospice centers) were excluded.
- The study was descriptive in nature, and no hypotheses were tested
  - Continuous variables are described as number and percentage, mean with corresponding SD, or median with corresponding range or IQR
  - Categorical variables are described as frequency and percentage
  - Missing or invalid observations were not included in the calculations

Results

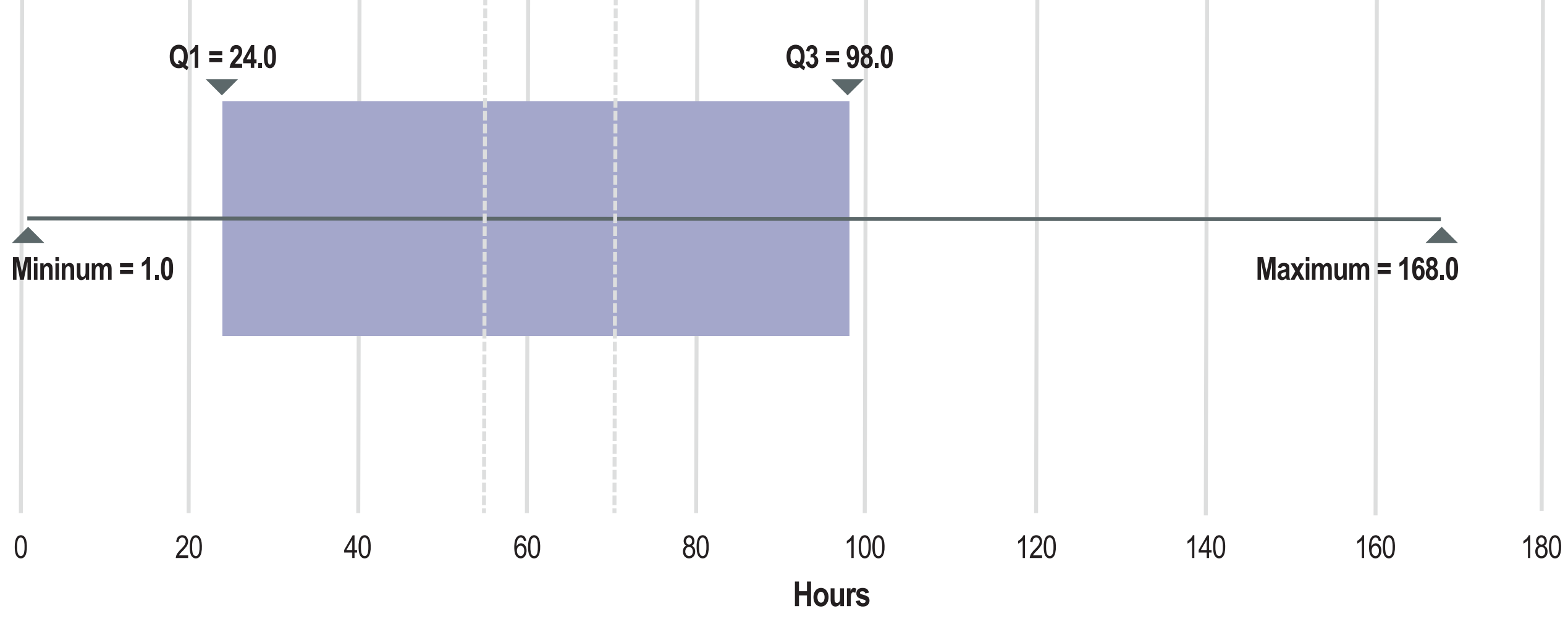
Caregiver demographics and characteristics

- A total of 124 caregivers managing 127 patients with SMA type 2 responded to the survey. Thirty caregivers in Japan cared for 32 patients, 30 caregivers in the UK cared for 30 patients, and 64 caregivers in the US cared for 65 patients.
  - Most patients (119/127 [93.7%]) had caregivers who cared for one patient
  - The mean (SD; median [range]) age of the patients was 14.0 (11.2; 10.4 [5.3–20.0]) years
  - The mean (SD; median [range]) age of the patients at diagnosis was 2.8 (5.2; 1.5 [1.1–2.1]) years
- The mean (SD) age of the caregivers was 43.6 (11.3) years
- Most patients (99/127 [78.0%]) had caregivers who were female, and for most patients (93/127 [73.2%]), the caregiver was the patient's mother
- Sixty-eight patients (53.5%) had caregivers who considered themselves to be the primary caregiver, and 54 (42.5%) had caregivers who considered themselves a co-primary caregiver. Five patients (3.9%) had caregivers who considered themselves to be a secondary caregiver.
- Nearly one-third of patients (41/127 [32.3%]) had a caregiver who completed a bachelor's degree, and 18.1% (23/127) had a caregiver who completed a post-graduate degree

Impact on caregiver time

- The mean (SD) patient care time was 69.6 (52.7) hours/week (**Figure 1**). The activities described by caregivers that required the most time were daily activities, support for bathing, dressing and toileting, posture management, feeding, assistance in moving, and physical therapy.

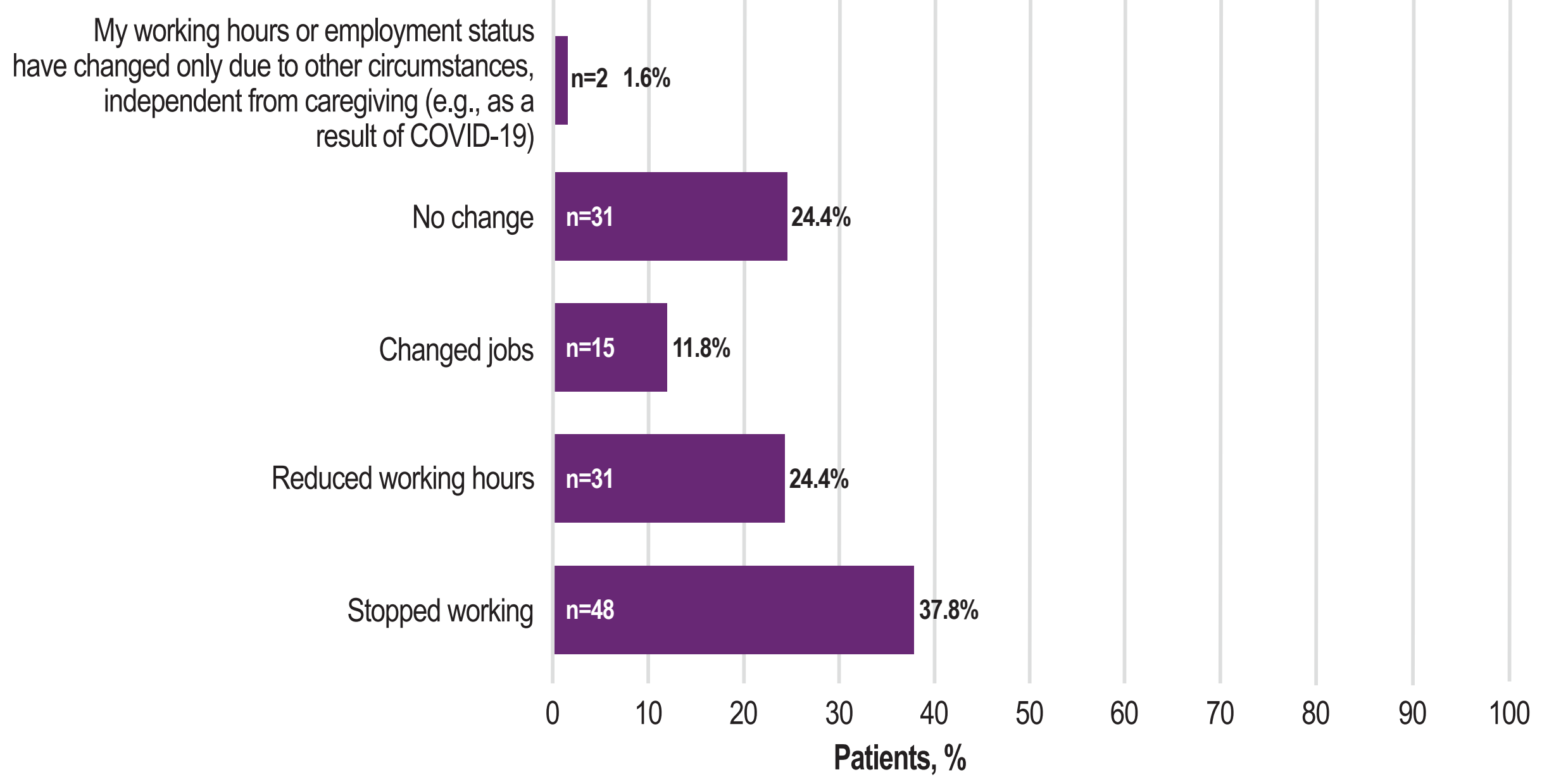
Figure 1. Hours of patient care time per week for caregivers of patients with SMA type 2



Q, quartile; SMA, spinal muscular atrophy.

- Of the 127 patients, 48 (37.8%) had a caregiver who stopped working to provide care; 31 (24.4%) had a caregiver who reduced working hours (mean reduction, 16.2 hours/week); and 15 (11.8%) had a caregiver who changed jobs (**Figure 2**)
- Within the last 6 months, 58/79 (73.4%) employed caregivers took days off from work for caregiving (mean [SD], 21.6 [38.8] days)
- A total of 84 (66.1%) caregivers reported the impact of caregiving on net income. The median (range) monthly reduction was $1,095 ($601–$2,000; USD equivalent as of September 30, 2022).
- In Japan, the median (range) monthly reduction was $1,034.26 ($551.61-$1,379.01); in the UK, $498.25 ($440.33-$782.80); and in the US, $1,500 ($1,000–$2,600)

Figure 2. Changes to employment for caregivers of patients with SMA type 2



SMA, spinal muscular atrophy.

Hospitalizations

- Within the last 6 months, 33 of 122 patients (27.0%) had ≥1 overnight hospitalizations, excluding SMA-related surgeries
  - The mean (SD) number of hospitalizations was 1.4 (0.7; median [range], 1.0 [1.0–2.0])
  - The mean (SD) length of hospitalization was 6.9 (10.6; median [range], 3.0 [2.0–7.0]) days
  - The primary reason for hospitalization was reported to be receiving treatment for SMA (18/33 [54.5%])
- The most frequent surgical interventions for patients with SMA type 2 were gastrostomy (n=24) and spinal fusion (n=36). Sixty patients (47.2%) had no surgical interventions related to SMA.

Out-of-pocket costs

- The most common out-of-pocket expenses reported by caregivers were traveling to medical appointments, adapting the home, commuting to school or work, securing accommodations close to the hospital, and paying for home health care (**Table 1**)
- Respiratory support, mobility equipment and aids, and nutrition support require substantial upfront and/or ongoing expenses for caregivers (**Table 2**)

Table 1. Selected out-of-pocket costs for caregivers of patients with SMA type 2 over the last 6 months

Out-of-pocket expense	Number of caregivers, n/N (%)[a]	Cost, mean (SD)[b]
Travel to/from medical appointments	102/124 (82.3%)	$587 ($1,223)
Home adaptations	74/124 (59.7%)	$8,310 ($24,334)
Commuting to school/workplace	33/124 (26.6%)	$1,596 ($7,271)
Accommodations close to hospital	23/124 (18.5%)	$655 ($1,179)
Home health care	23/124 (18.5%)	$2,890 ($6,293)

SMA, spinal muscular atrophy.
[a]Not all responses for this category were mandatory. Caregivers may have incurred these out-of-pocket costs but were not required to report them.
[b]USD equivalent as of September 30, 2022.

Table 2. Selected upfront and ongoing costs for caregivers of patients with SMA type 2

Out-of-pocket expense		Number of caregivers, n/N (%)[a]	Cost at start of use, mean (SD)[b]	Cost per month, mean (SD)[b]
Respiratory support	Pulse oximeter	24/44 (54.5%)	$77 ($101)[c]	$64 ($124)
	Cough assist machine	11/57 (19.3%)	$284 ($183)[c]	$116 ($190)
	Suction machine	8/31 (25.8%)	$250 ($350)[c]	$30 ($39)
	Nebulizer	8/33 (24.2%)	$960 ($1,047)[c]	$27 ($34)
Mobility equipment	Ankle-foot orthosis	15/60 (25.0%)	$290 ($306)	$13 ($27)
	Special bed	10/35 (28.6%)	$713 ($709)	$11 ($34)
	Special seating or sitting retainer	17/46 (37.0%)	$518 ($1,008)	$45 ($109)
Wheelchair	Adaptive stroller/standard stroller	10/15 (66.7%)	$1,367 ($2,453)	$94 ($174)
	Manual wheelchair, including self-propelled wheelchair with electromotion system	35/59 (59.3%)	$1,282 ($1,446)	$13 ($73)
	Power wheelchair	33/83 (39.8%)	$3,217 ($4,883)	$47 ($152)
Nutrition support	Naso-gastric tube (NG tube)	1/2 (50.0%)	$350 (–)	N/A
	Gastric feeding tube (G-tube)	4/18 (22.2%)	$963 ($1,858)	N/A

N/A, not available; SMA, spinal muscular atrophy.
[a]Denominators represent the number of caregivers who reported the use of a specific device by patients. Numerators represent the number of those who incurred out-of-pocket costs for use of those devices.
[b]USD equivalent as of September 30, 2022.
[c]Not available for Japan.

Limitations

- Caregiver answers were declarative
- Several survey questions targeted intimate and discreet behaviors of everyday life. This type of information is often hard to grasp as the participants' answers tend to be inaccurate or nonspecific.
- The lengthening of the recall period was likely to trigger the telescopic memory effect and cause participants to insert imprecise answers. This concern is greater because answers to numerous questions are not mandatory. Consequently, the participant may become encouraged to excessively rely on the "skip" button whenever this option is allowed. Survey fatigue may have also played a role in response limitations.
- The heterogeneity between the countries limits the interpretation of the results
- Specific information about the treatments that patients received and when they may have received therapy was not collected for patients in Japan, although this was collected for patients in the UK and US
- This study was conducted amid the COVID-19 pandemic in 2020 and 2021

Conclusions

- Caregivers of patients with SMA type 2 in Japan, the UK, and the US reported a substantial burden on time, employment status, income, and out-of-pocket costs
- Lost productivity and HCRU associated with SMA type 2 impose great direct and indirect societal burdens
- Early SMA identification via newborn screening and early access to DMTs may reduce these burdens and associated costs
- Work is ongoing to describe caregiver burden globally

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Abbreviations

DMT, disease-modifying treatment; HCRU, health care resource utilization; IQR, interquartile range; SMA, spinal muscular atrophy; *SMN1*, *survival motor neuron 1* gene; UK, United Kingdom; US, United States.

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