The Impact of Spinal Muscular Atrophy Type 1 on Caregivers in Brazil: Results of a Global Survey

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Introduction

- SMA is a rare, progressive, genetic neurodegenerative disease that causes loss of both voluntary motor and bulbar functions[1,2]
- SMA1, accounting for >50% of SMA cases, is the most severe form, with death often occurring prior to 2 years of age[1,2]
- Patients with SMA1 develop symptoms within the first 6 months of life, including limb weakness, respiratory insufficiency, and poor feeding.[1] Historically, patients with SMA1 did not achieve the ability to sit independently.[1]
- A few studies, mostly conducted in the United States and Europe, demonstrated that caregivers of patients with SMA1 are greatly impacted by time-consuming daily care tasks, financial problems, and work adjustments owing to care tasks[3]
- Overall, data are limited on the impact of the time and costs associated with care for patients with SMA1, particularly in Brazil, where an estimated 250–300 newborns per year are affected by SMA[4]

Objective

- We sought to describe the sociodemographics, HCRU, and time and out-of-pocket costs for caregivers of patients with SMA1 in Brazil

Methods

- An online survey of caregivers in Argentina, Brazil, Japan, Taiwan, the United Kingdom, and the United States was conducted to collect data on time spent on and economic impact of caring for a patient with SMA1–3 (**Figure 1**). Here, we report the results for caregivers of patients with SMA1 in Brazil.

Figure 1. Caregiver survey study design

Study Design

Cross-sectional, non-interventional, online survey of voluntary non-HCP caregivers in Brazil. The survey was disseminated to caregivers through patient advocacy groups and principal investigators

Survey start
September 2022

Survey end
March 2023

Data Collected in Survey

- Caregiver sociodemographics
- Characteristics of patients with SMA cared for by caregivers
- Time spent as a caregiver
- Impact of caregiving on employment and income
- HCRU
- Direct medical care: HCP visits, surgeries, or hospitalizations
- Indirect medical care: out-of-pocket expenses

Participants

Adult, unpaid primary or secondary caregivers of a child or adult with SMA1 at the time of survey completion

- HCP caregivers
- Paid caregivers (e.g., home health aides or registered nurses)
- Caregivers of patients residing in stationary facilities (e.g., nursing homes or hospice centers)

Ethics

- All participants provided informed consent electronically
- Study documents and protocols were approved by an international IRB and local ethics committees

HCP, health care provider; HCRU, health care resource utilization; IRB, institutional review board; SMA1, spinal muscular atrophy type 1.

Results

Caregiver demographics and patient characteristics

- Fourteen caregivers, managing 14 patients with SMA1, responded to the survey
- Caregivers were a mean age of 34.4 years and were either the patient's mother (71.4%) or father (28.6%; **Table 1**)

Table 1. Sociodemographics for caregivers of patients with SMA1

Characteristic	Caregivers (N=14)
Sex, n (%)	
Female	10 (71.4)
Male	4 (28.6)
Relationship to the patient, n (%)	
Mother	10 (71.4)
Father	4 (28.6)
Age, years	
Mean (SD)	34.4 (8.3)
Median (range)	32.5 (20.0–52.0)
Highest education level completed, n (%)	
Some primary or secondary school	4 (28.6)
GCSEs/O-levels/standard grade	4 (28.6)
Undergraduate degree	3 (21.4)
Graduate degree	3 (21.4)
Marital status, n (%)	
Single	1 (7.1)
Married or in a domestic partnership	9 (64.3)
Living with a partner (not married or in a domestic partnership)	4 (28.6)
Geographic entity, n (%)	
Rural (countryside or village)	1 (7.1)
Urban (town or city)	13 (92.9)
Number of children in the household younger than 18 years[a]	
Mean (SD)	1.6 (0.9)
Median (range)	1.0 (1.0–3.0)
Current employment status, n (%)[b]	
Employed full time	4 (26.7)
Self-employed/business owner	2 (13.3)
Unemployed	1 (6.7)
Full-time caregiver of a person with SMA	8 (53.3)
Current gross income before taxes, n (%)	
<R$2,090.00/month	4 (28.6)
R$2,090.01–R$4,180.00/month	2 (14.3)
R$4,180.01–R$10,450.00/month	4 (28.6)
R$10,450.01–R$20,900.00/month	2 (14.3)
Prefer not to answer	2 (14.3)

R$, Brazilian real; GCSE, General Certificate of Secondary Education; SD, standard deviation; SMA, spinal muscular atrophy.
[a]Including the patient with SMA1. [b]Caregivers may select more than one option.

- The mean age of patients with SMA1 was 3.5 years, and the mean age at SMA diagnosis was 0.7 years (**Table 2**)
  - Patients achieved their greatest level of motor function at a mean age of 0.7 years
  - All patients required the use of respiratory equipment within the last 6 months, whereas most patients (92.9%; 13/14) required mobility equipment, and only six of 14 patients (42.9%) required nutrition equipment

Table 2. Characteristics of patients with SMA1

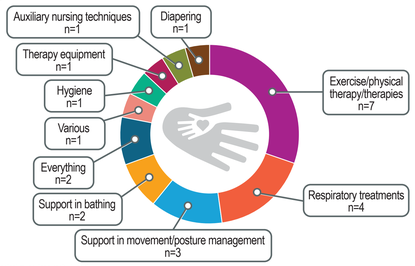
Characteristic	Patients[a] (N=14)
Age, years	
Mean (SD)	3.5 (4.0)
Median (range)	2.5 (0.9–15.9)
Age at SMA diagnosis, years	
Mean (SD)	0.7 (1.0)
Median (range)	0.5 (0.1–4.0)
Time from first signs/symptoms to diagnosis of SMA, months	
Mean (SD)	6.2 (10.9)
Median (range)	2.5 (0–40)
Highest level of motor function achieved, n (%)	
Head control	5 (35.7)
Rolling	0
Sitting independently for >10 seconds	3 (21.4)
Standing with support	0
Standing without support	0
Walking with support	1 (7.1)
Walking without support	0
None of the above	5 (35.7)
Age patient maintained highest level of motor function achieved, years[b]	
Mean (SD)	0.7 (0.7)
Median (range)	0.5 (0–1.8)
Current level of motor function, n (%)[b]	
Head control	2 (22.2)
Rolling	0
Sitting independently for >10 seconds	4 (44.4)
Standing with support	0
Standing without support	0
Walking with support	0
Walking without support	0
None of the above	3 (33.3)
Respiratory equipment used within the last 6 months, responses (% of responses)[c]	
Cough assist machine	13 (23.6)
Non-invasive ventilation	12 (21.8)
Nebulizer	11 (20.0)
Pulse oximeter	10 (18.2)
Suction machine	5 (9.1)
BiPAP machine	1 (1.8)
Breathing machine	1 (1.8)
High-frequency chest wall oscillation	1 (1.8)
None	0
Mobility equipment used in the last 6 months, responses (% of responses)[c]	
Ankle-foot orthoses	12 (38.7)
Wheelchair	4 (12.9)
Adaptive	2/4 (50.0)
Manual	1/4 (25.0)
Manual with power assist wheels	1/4 (25.0)
Corset or spinal jacket	4 (12.9)
Special bed	3 (9.7)
Hand braces	3 (9.7)
None	1 (3.2)
Nutrition equipment used in the last 6 months, responses (% of responses)[c]	
NG tube	1 (6.7)
G-tube	3 (20.0)
None	8 (53.3)

BiPAP, bilevel positive airway pressure; G-tube, gastric feeding tube; NG, naso-gastric; SD, standard deviation; SMA1, spinal muscular atrophy type 1.
[a]Responses provided by 14 caregivers for a total of 14 patients with SMA1. [b]Nine of 14 caregivers provided responses. [c]Caregivers may select more than one option.

Level of care provided

- All caregivers were either the patient's primary (57.1%; 8/14) or co-primary (42.9%; 6/14) caregiver
- The mean (SD; median [range]) patient care time reported by caregivers (N=14) was 62.8 (60.0; 24.0 [8.0–168.0]) hours/week
- Caregivers reported that respiratory treatments, therapies, and physical therapy were the most time-consuming activities in caring for their patients with SMA1 (**Figure 2**)

Figure 2. Most time-consuming tasks reported by caregivers of patient with SMA1



SMA1, spinal muscular atrophy type 1.

Employment and income

- Of the 14 caregiver respondents, nine stopped working and two reduced their working hours to provide care for their patients with SMA1 (**Figure 3**)
- Caregivers (n=2 respondents) reduced their working hours by a mean (SD; median [range]) of 4.0 (0; 4.0 [4.0–4.0]) hours/week

Figure 3. Employment changes for caregivers of patients with SMA1

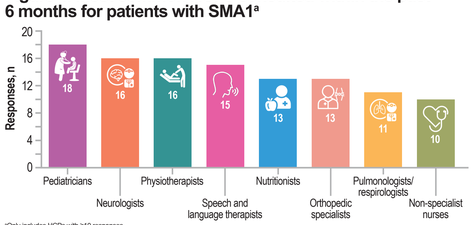
SMA1, spinal muscular atrophy type 1.

- Within the last 6 months, four of five caregivers (80.0%) took days off from work related to caregiving, and the mean (SD; median [range]) number of days these four caregivers took off from work was 22.5 (15.5; 17.5 [10.0–45.0]) days
- Eleven of 14 caregivers (78.6%) reported an impact on income related to caregiving with an estimated mean (SD; median [range]) income reduction of R$2,833.30 (816.50; 3,000 [1,500–4,000])/month for six caregiver respondents

HCRU

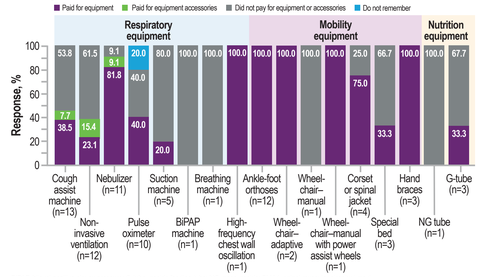
- All caregivers consulted at least one HCP during the last 6 months for their patient with SMA1. Patients required multidisciplinary care from several types of HCPs (**Figure 4**).
- For patients with SMA1 who had surgeries, surgery types included gastrostomies (n=8 responses), gastrostomies with Nissan fundoplication (n=2 responses), tracheostomies (n=2 responses), and orchiopexies or shunting (n=1 response each)
- Seven of 14 patients (50.0%) had ≥1 overnight hospitalizations, excluding SMA-related surgeries, within the last 6 months
  - The primary reasons for these hospitalizations were chest infection/breathing difficulties (n=4), and COVID-19, infection, or feeding difficulties (n=1 each)
  - The mean (SD; median [range]) duration of these hospitalizations for these seven patients was 7.9 (7.2; 5.0 [1.0–21.0]) days
- Caregivers reported paying out-of-pocket expenses for most respiratory and mobility, but not nutritional, equipment needed for their patients with SMA1 (**Figure 5**)

Figure 4. Most common HCPs consulted within the past 6 months for patients with SMA1[a]



[a]Only includes HCPs with ≥10 responses.
HCP, health care provider; SMA1 spinal muscular atrophy type 1.

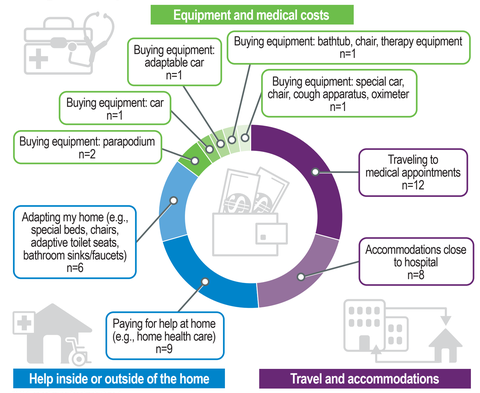
Figure 5. Out-of-pocket expenses caregivers reported for respiratory, mobility, and nutrition equipment needed for their patients with SMA1



BiPAP, bilevel positive airway pressure; G-tube, gastric feeding tube; NG, naso-gastric; SMA1, spinal muscular atrophy type 1.

- Caregivers also reported paying out-of-pocket expenses for travel and accommodations, equipment, and help inside or outside of the home to care for their patients with SMA1 (**Figure 6**)

Figure 6. Additional out-of-pocket expenses reported by caregivers of patients with SMA1



SMA1, spinal muscular atrophy type 1.

Limitations

- A small number of participants completed the survey, and not all participants responded to all survey questions
- The accuracy of caregiver survey responses could not be verified
- Several survey questions targeted intimate and discreet behaviors of everyday life. This type of information is often hard to validate, 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. In addition, because answers to several questions were not mandatory, participants may have become encouraged to excessively rely on the "skip" button whenever this option was allowed.

Conclusions

- In Brazil, caregivers were parents of children with SMA1 who often required the use of respiratory and mobility equipment
- Caregivers reported a substantial impact on their time for providing care, particularly because of respiratory treatments, therapies, and physical therapy
- Often caregivers reduced their working hours or stopped working, leading to a reduction in income
- Caregivers reported out-of-pocket costs for equipment, travel and accommodations, and help inside or outside of the home
- Early SMA identification and early access to disease-modifying treatments may reduce the impact on caregivers and associated costs, because early treatment can improve patients' clinical outcomes and health-related quality of life

References

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Abbreviations

BiPAP, bilevel positive airway pressure; COVID-19, coronavirus disease 2019; G-tube, gastric feeding tube; GCSE, General Certificate of Secondary Education; HCP, health care provider; HCRU, health care resource utilization; IRB, institutional review board; NG, naso-gastric; R$, Brazilian real; SD, standard deviation; SMA, spinal muscular atrophy; SMA1, spinal muscular atrophy type 1.

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