The Burden of Spinal Muscular Atrophy Type 1 on Caregivers in Argentina: First Results of a Global Survey Launched in Latin America

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Introduction

- SMA is a rare, genetic, progressive neuromuscular disorder caused by biallelic deletion or mutation of the *SMN1* gene[1]
- SMA type 1 is the most common and most severe form of SMA and accounts for >50% of new SMA cases. It often results in death or the need for permanent ventilation before 2 years of age if untreated.[1,2]
- Symptoms of SMA type 1 usually appear at birth or within the first months of life[1]
  - Most infants with SMA type 1 are unable to support their heads or sit without assistance
  - They also develop breathing and swallowing problems[3]
- SMA is associated with substantial costs,[4] as well as physical and emotional burdens for patients, families, caregivers, and society[5]
  - 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[6–8]
  - Caregiver burden associated with SMA type 1 around the world has not been well-characterized

Objectives

- We sought to describe sociodemographic and time costs for the families and caregivers of patients with SMA type 1
  - The goal of this ongoing study is to assess burdens globally
  - Data presented here reflect caregiver burden in Argentina

Methods

- An online survey was disseminated through a patient advocacy group (FAME) from February 2022 through May 2022
- 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

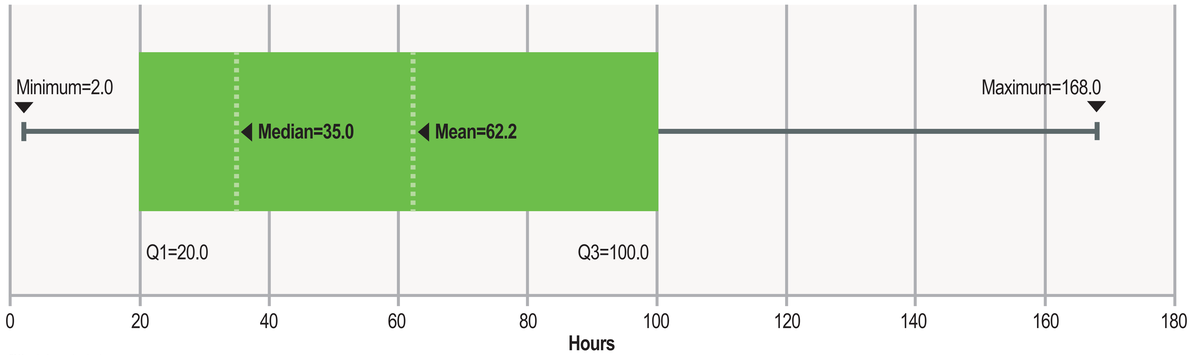
Caregiver demographics and characteristics

- A total of 22 caregivers managing 23 patients with SMA type 1 responded to the survey
  - Most patients (21/23 [91.3%]) had caregivers who cared for one patient; two (8.7%) had caregivers who cared for two patients
  - The mean (SD; median [range]) age of the patients was 6.7 (5.3; 4.8 [1.8–17]) years
  - The mean (SD; median [range]) age of the patients at diagnosis was 0.5 (0.3; 0.4 [0.1–1.4]) years
- The mean (SD) age of the caregivers was 36.6 (7) years
- Most patients (15/23 [65.2%]) had caregivers who were female, and for most patients (14/23 [60.9%]), the caregiver was the patient's mother
- Thirteen patients (56.5%) had caregivers who considered themselves to be the primary caregiver, and ten (43.5%) had caregivers who considered themselves to be a co-primary caregiver
- Ten patients (43.5%) had a caregiver whose highest level of education was a high school diploma. Six patients (26.1%) had a caregiver who completed an Associate's degree, three patients (13.0%) had a caregiver who completed a Bachelor's degree, and three (13.0%) had a caregiver who completed a Master's degree or higher.

Impact on caregiver time

- The mean (SD) patient care time was 62.2 (54.7) hours/week (**Figure 1**)
- The activities described by caregivers that required the most time were daily activities, physical therapy, and support for bathing

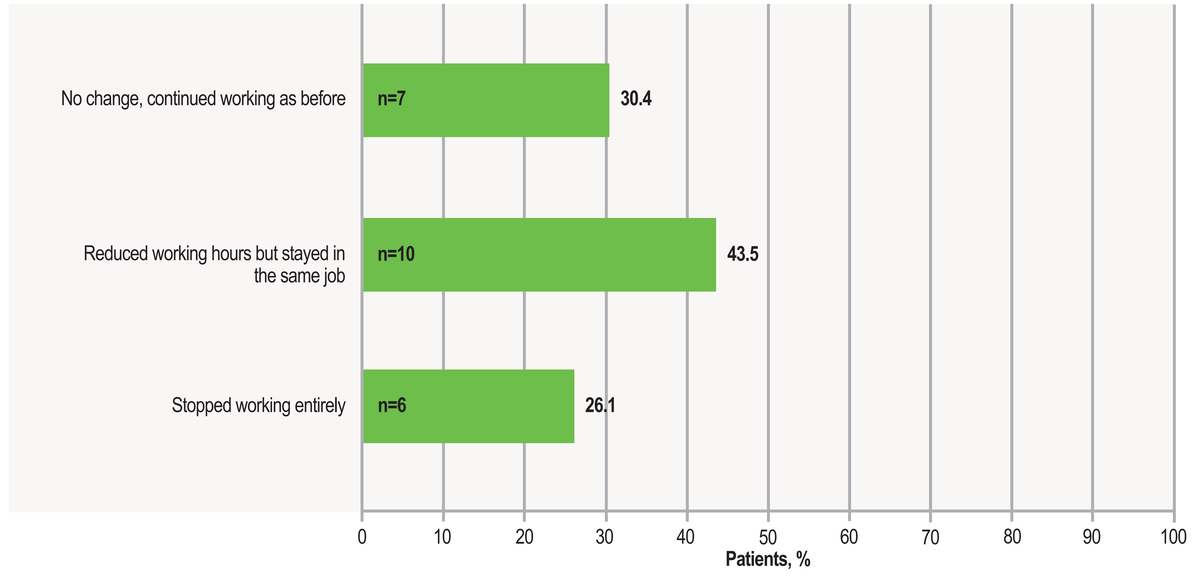
Figure 1. Hours of patient care time per week for caregivers of patients with SMA type 1



SMA, spinal muscular atrophy.

- Of the 23 patients, six (26.1%) had a caregiver who stopped working to provide care and ten (43.5%) had a caregiver who reduced working hours (mean reduction, 18.7 hours/week) (**Figure 2**)
- Within the last 6 months, 13 of 17 (76.5%) employed caregivers took days off from work for caregiving (mean [SD], 12.1 [20.9] days)
- A total of 15 (65.2%) patients had caregivers who reported the impact of caregiving on net income. The mean (SD) monthly reduction was 48.3% (29.6).

Figure 2. Changes to employment in the past 6 months for caregivers of patients with SMA type 1



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Limitations

- 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. This concern is greater because answers to several questions are not mandatory. Consequently, the participant may become encouraged to excessively rely on the "skip" button whenever this option is allowed.

Conclusions

- Caregivers of patients with SMA type 1 in Argentina reported a substantial burden on time, employment status, and income
- The high degree of care that is required and the financial pressure that is created can have a substantial impact on caregiver health-related quality of life
- Lost productivity associated with caregiving for a patient with SMA type 1 imposes direct and indirect societal burdens
- Early identification of SMA via newborn screening and early access to disease-modifying treatments may reduce these burdens and associated costs
- Work is ongoing to describe caregiver burden globally

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Abbreviations

FAME, Families of SMA, Argentina; IQR, interquartile range; SD, standard deviation; SMA, spinal muscular atrophy; *SMN1*, *survival motor neuron 1* gene.

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