Social and Demographic Determinants of Disease Burden Among Adults with Spinal Muscular Atrophy

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Background

- Spinal muscular atrophy (SMA) is a rare genetic neuromuscular disease characterized by progressive muscle atrophy and weakness that historically was the most common genetic cause of infant mortality[1].
  - SMA is classified into five main types based on age of symptom onset and highest physical milestones achieved.
- Since 2016, three disease modifying therapies (DMTs) have been FDA-approved for the treatment of SMA in adults[2-4], leading to improved rates of survival, motor function, and respiratory function for SMA patients, however, social determinants of health (SDOH) may also influence overall well-being and health outcomes.
- Understanding how SDOH contribute to differences in symptoms and health related quality of life is essential for developing comprehensive care strategies for this population.
- The objective of this analysis was to examine the relationship between SDOH and patient-reported disease burden in adults with SMA Types 2 and 3.

Methods

- Since 2017, Cure SMA, a patient advocacy organization that provides support and funding for care and treatment for those with SMA, has hosted an annual community update survey (CUS) for caregivers and affected individuals.
  - The CUS captures data such as demographics, treatment experience and patient reported outcome measures (PROMs), including the Spinal Muscular Atrophy Health Index (SMA-HI) short-form.
  - The SMA-HI was designed to assess symptoms and health-related quality of life from the perspective of the patient with SMA. It is measured on a scale of 0-100, with 0 representing no disease burden, and 100 representing the maximum level of disease burden[5].
- Data from the 2025 CUS was utilized for this analysis.
  - Survey responses were fielded from 4/24/2025 – 5/27/2025
- The final analysis consisted of living adults 18 years and older at time of survey completion, with type 2 or type 3 SMA, who self-completed the survey.
  - Individuals who did not complete the full SMA-HI short form or who were missing data on household income, employment status, race/ethnicity, treatment status, or ZIP code were excluded.
  - A linear regression model was used to measure the impact of SDOH factors on disease burden, measured by the SMA-HI.

Results

- The final sample included 140 adults with SMA.
- **Table 1** describes the demographic and clinical characteristics of the sample.
  - Most individuals were female (72%) and White (76%), with an average age of 39.7 years.
  - The sample was split between Type 2 (51%) and Type 3 (49%) SMA, with most being non-ambulatory (33% non-sitter; 44% sitter), and most (84%) were currently receiving treatment.
  - The majority (86%) lived in an urban focused area, half (50%) were employed either full-time or part-time, and 35% had an annual household income less than $40,000.

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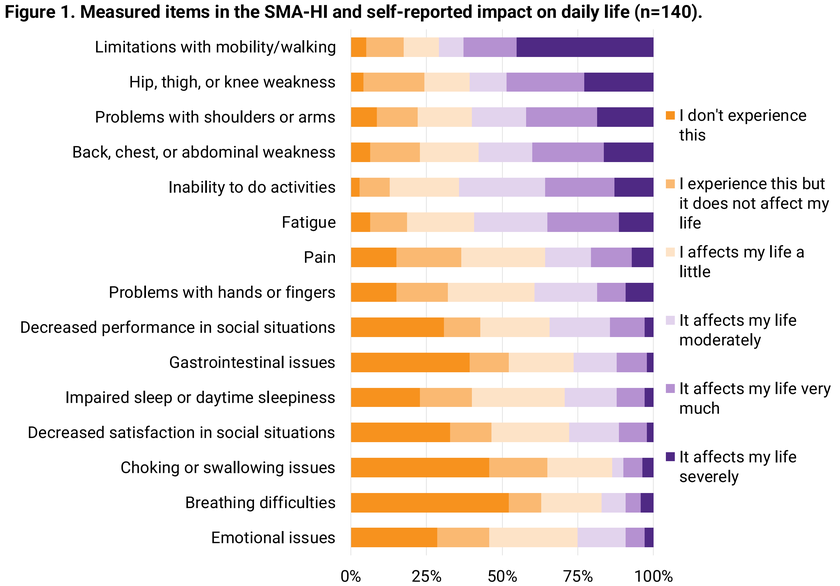
The Cure SMA Industry Collaboration (SMA-IC) was established in 2016 to leverage the experience, expertise, and resources of pharmaceutical and biotechnology companies, as well as other nonprofit organizations involved in the development of spinal muscular atrophy (SMA) therapeutics to more effectively address a range of scientific, clinical, and regulatory challenges. Funding for this research was provided by the 2025 SMA-IC; members include Cure SMA, Biogen, Novartis, Scholar Rock, Genentech/Roche Pharmaceuticals, argenx, NMD Pharma, and SMA Europe.

Results

Table 1. Demographics and SMA-HI scores among respondent sample.

	Overall Sample n (%)	SMA-HI Score mean (sd)
Overall	140 (100%)	39.0 (21.0)
Age at Survey (mean, SD)	39.7 (13.5) years	n/a
Race/Ethnicity		
White	106 (76%)	41.1 (19.9)
Non-White	34 (24%)	32.5 (23.2)
Gender		
Male	37 (26%)	38.2 (20.4)
Female	101 (72%)	29.4 (21.3)
Type of SMA		
Type 2	72 (51%)	42.5 (20.5)
Type 3	68 (49%)	35.4 (21.0)
SMN2 Copy Number		
1-2 copies	12 (9%)	44.4 (22.4)
3 copies	66 (47%)	43.1 (21.1)
4 or more copies	29 (21%)	32 (19.0)
Don't know	33 (24%)	35 (20.3)
Ambulation Status		
Non-sitter	46 (33%)	42.6 (21.9)
Sitter	61 (44%)	41.1 (19.6)
Walker	33 (24%)	30.1 (20.3)
SMA Treatment Status		
Currently on treatment	117 (84%)	38.5 (21)
Discontinued treatment	17 (12%)	45.1 (21.1)
Never treated	6 (4%)	32.2 (20.5)
Residence		
Urban focused area	120 (86%)	39.4 (21.0)
Large rural city/town	11 (8%)	42 (13.4)
Small/isolated small rural town	9 (6%)	30.3 (28.0)
Employment Status		
Full-time employment	49 (35%)	36 (18.7)
Part-time employment	21 (15%)	49.1 (21.6)
Not employed	70 (50%)	48.1 (21.6)
Household Income		
< $40,000	49 (35%)	43.9 (23.1)
$41,000-$70,000	16 (11%)	30.8 (15.8)
$71,000-$100,000	23 (16%)	37 (18.8)
> $100,000	36 (26%)	37.3 (22.0)
Prefer not to answer	16 (11%)	39.1 (18.4)

Figure 1. Measured items in the SMA-HI and self-reported impact on daily life (n=140).



Factors Impacting Disease Burden

- The mean SMA-HI score was 39.0 (SD 21.0). **(Table 1).**
- Items reported as having the greatest impact on the lives of individuals with SMA were "Limitations with mobility or walking", "Hip, thigh, or knee weakness", "Problems with shoulders or arms", and "Back, chest, or abdominal weakness" **(Figure 1).**
- **Table 2** shows findings from the linear regression model. Key findings from the model include:
  - **Household Income:** Individuals with an annual household income of $41,000-$70,000 had significantly lower SMA-HI scores compared to those earning less than $41,000.
  - **Employment status:** Part-time employment was associated with significantly higher SMA-HI scores relative to full-time employment.
  - **Race/Ethnicity:** Non-White individuals had significantly lower SMA-HI scores compared with White individuals.

Table 2. Predictors of SMA-HI scores.

Covariate	Estimate	Std. Error	t-value	Pr(>|t|)
Intercept	47.55	7.01	6.78	<0.001
Age at Survey				
Continuous	0.02	0.14	0.13	0.893
Race/Ethnicity				
White	{ref}			
Non-White	-9.72	4.08	-2.68	**0.019**
Type of SMA				
Type 2	{ref}			
Type 3	-5.39	3.78	-1.42	0.156
SMA Treatment Status				
Currently on treatment	{ref}			
Discontinued treatment	3.67	5.45	0.67	0.501
Never treated	-9.37	9.08	-1.03	0.304
Residence				
Urban focused area	{ref}			
Large rural city/town	1.50	6.47	0.23	0.817
Small/isolated small rural town	-10.76	7.18	-1.50	0.136
Employment Status				
Full-time employment	{ref}			
Part-time employment	12.14	6.04	2.01	**0.047**
Not employed	-2.41	4.77	-0.51	0.614
Household Income				
< $40,000	{ref}			
$41,000-$70,000	-16.75	6.35	-2.64	**0.009**
$71,000-$100,000	-6.79	5.85	-1.16	0.248
> $100,000	-3.78	5.77	-.065	0.514
Prefer not to answer	-3.46	6.10	-0.57	0.572

Conclusions

- A few of the SDOH evaluated were modestly associated with disease burden in adults with SMA.
- These findings suggest that socioeconomic and demographic factors may contribute to disparities in SMA-related quality of life outcomes.
- SDOH factors should be considered when assessing and supporting patients with SMA.
- Further research with larger and more clinically diverse SMA populations is warranted to better understand the impact of SDOH across the full spectrum of disease severity.

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