

# Impact of the motor function of individuals with spinal muscular atrophy on caregiver burden

*Impacto da função motora de indivíduos com atrofia muscular espinhal na sobrecarga de seus cuidadores*

*Impacto de la función motora de los individuos con atrofia muscular espinal en la carga de sus cuidadores*

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**ABSTRACT** | Spinal Muscular Atrophy (SMA) is a neurodegenerative disease that impairs motor function, justifying the help of a caregiver. This study aimed to analyze the impact of the motor function of individuals with SMA on caregiver burden. This is a cross-sectional study of 32 individuals with SMA and 27 caregivers, carried out in a rehabilitation center, using the identification questionnaire, anamnesis and sociodemographic profile, Motor Function Measurement Scale and Burden Interview. Motor function and caregiver burden were compared between SMA types using the analysis of covariance (ANCOVA) and correlated using Pearson's correlation test. Individuals with SMA type I had greater impairment of motor function when compared to types II and III, and individuals with type III had better scores in all domains of motor function. No correlation was observed between motor function and caregiver burden: however, most caregivers presented some level of burden, especially those from mild to moderate. Moderate to severe burden was found in caregivers with SMA in a considerable prevalence, which requires care and attention from health professionals.

**Keywords** | Neuromuscular Disease; Rare Diseases; Caregiver Burden.

**RESUMO** | Atrofia muscular espinhal (AME) é uma doença neurodegenerativa que provoca comprometimento na função motora, justificando o auxílio de cuidador. O objetivo deste estudo foi analisar o impacto da função motora de indivíduos com AME na sobrecarga de seus cuidadores. Trata-se de um estudo transversal realizado em um centro de reabilitação com 32 indivíduos portadores de AME e 27 cuidadores, utilizando como metodologia o questionário de identificação, anamnese e perfil sociodemográfico, a escala da medida da função motora e *Burden Interview*. A função motora e a sobrecarga do cuidador foram comparadas entre os tipos de AME por meio da Análise da Covariância (Ancova) e correlacionadas pelo teste de correlação de Pearson. Os indivíduos com AME tipo I apresentaram maior comprometimento da função motora quando comparado entre os tipos II e III, e indivíduos tipo III apresentaram melhores escores em todos os domínios

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da função motora. Não foi percebida correlação entre função motora e sobrecarga de cuidador, entretanto encontramos a maior parte dos cuidadores apresentando algum nível de sobrecarga, com destaque ao de leve a moderado. A sobrecarga moderada a severa foi encontrada nos cuidadores com AME em uma prevalência considerável, o que requer cuidado e atenção dos profissionais de saúde.

**Descritores** | Doenças Neuromusculares; Doenças Raras; Fardo do Cuidador.

**RESUMEN** | La atrofia muscular espinal (AME) es una enfermedad neurodegenerativa que provoca un deterioro de la función motora, por lo que requiere la asistencia de cuidadores. El objetivo de este estudio fue analizar el impacto de la función motora de los individuos con AME en la carga de sus cuidadores. Se trata de un estudio transversal, realizado en un centro de rehabilitación con 32 individuos con AME y 27 cuidadores, utilizando como metodología el cuestionario de identificación, anamnesis y perfil

sociodemográfico, la escala de medida de la función motora y la entrevista de carga. La función motora y la carga de los cuidadores se compararon entre los tipos de AME mediante el análisis de covarianza (Ancova) y se correlacionaron mediante la prueba de correlación de Pearson. Los individuos con AME tipo I tuvieron un mayor deterioro de la función motora en comparación con los tipos II y III, y los individuos de tipo III obtuvieron mejores puntuaciones en todos los dominios de la función motora. No se encontró correlación entre la función motora y la carga del cuidador, pero la mayoría de los cuidadores tenían algún nivel de carga, en particular de leve a moderada. Se encontró una carga de moderada a grave en los cuidadores con AME con una prevalencia considerable, lo que requiere cuidados y atención por parte de los profesionales de la salud.

**Palabras clave** | Enfermedades Neuromusculares; Enfermedades Raras; Carga del Cuidador.

## INTRODUCTION

Spinal muscular atrophy (SMA) is defined as a neurodegenerative disease of dominant recessive genetic character, marked by a progressive impairment of the motor neuron, having as main characteristics muscle weakness and atrophy<sup>1</sup>. Its incidence is approximately 1:11,000 live births<sup>2</sup>.

Individuals diagnosed with SMA are divided into four types according to age of symptom onset, motor milestones achieved, and clinical characteristics. Individuals classified as type I are those in which the onset of symptoms occurs before six months of age and who present symmetrical muscle weakness, severe hypotonia, inability to sit, in addition to changes in respiratory pattern. Individuals classified as type II are those in which symptoms occur from seven to 18 months of age, are able to sit, but cannot walk, and are prone to develop musculoskeletal and respiratory complications<sup>3</sup>.

Types III and IV are milder forms, and individuals with type III can develop the ability to stand and walk. In type IV, they can maintain the ability to walk in adulthood and the course of the disease is the mildest<sup>3</sup>.

Due to the differences between the types and changes in phenotypes after interventions, especially medication, it is necessary to evaluate motor function and the ability to perform daily activities<sup>4</sup>. In this sense, several scales have

been developed and, in Brazil, the Portuguese version of the motor function measurement scale has been shown to be reliable and highly reproducible in people with neuromuscular diseases<sup>5</sup>.

The complexity of the clinical condition and its impact on functionality makes the individual need help in performing some basic daily tasks. Caregivers—who may be an informal caregiver or a family member—come to assume responsibility for care in the home setting, directly assisting in the routine of this individual. They can present difficulties and negative implications for care, involving physical and emotional exhaustion<sup>6</sup>, mainly because they are people who were not prepared and who start to play a new role involving a series of daily challenges<sup>7</sup>. When these caregivers are the parents, who generally prioritize the needs of their children and dedicate many hours to their care, the difficulties and implications may be more significant, harming their own health and increasing the level of burden<sup>8</sup>.

In this context, aspects related to caregivers, especially the level of burden, are relevant and should be studied, and this analysis is possible by employing specific questionnaires<sup>9</sup>, thus enabling professionals who deal directly with this population to perceive the difficulties of these caregivers and conduct strategies to provide qualified support, reducing the burden and seeking to improve their well-being<sup>7</sup>.

After reviewing the literature, it was possible to observe that studies that correlate motor function and the level of burden of caregivers of individuals with SMA are scarce, but of fundamental importance to know the profile of them and their caregivers, understand how deficits in motor function arising from the clinical condition can impact the level of burden of the caregiver, and enable the development and conduct of strategies and interventions to improve the well-being of caregivers, preventing or reducing the level of burden.

Accordingly, this study aims to analyze the impact of motor function of individuals with SMA on the level of burden of their caregivers.

## METHODOLOGY

### Study specifications

This is an observational, cross-sectional, and analytical study carried out in a rehabilitation center in the city of Goiânia, GO, Brazil. All participants included in the study, aged over 18 years, signed an informed consent form. Participants aged under 18 years had the consent form signed by their guardian and assented to their participation through the informed assent form. The study followed the Regulatory Standards for Research Involving Human Beings. Data collection was carried out after approval by the Ethics Committee, from March to August 2022.

### Sample

The sample consisted of 32 participants with SMA and 27 caregivers. Individuals diagnosed with SMA of both sexes and in any age group were included, provided the disease had been diagnosed by a physician, confirmed through clinical findings and laboratory examinations, and who were under medical or outpatient follow-up at the rehabilitation center. The study also included the main family caregivers of these individuals, of both sexes and aged over 18 years. Caregivers or participants with SMA who had difficulties understanding the questions or refused to answer any of the instruments were excluded from the study.

Sample size was calculated according to motor function and caregiver burden scores using G.Power® 3.1 software. To this end, a 5% significance level, a 95% confidence interval, and a 80% sampling power were adopted. Thus, a minimum sample of 19 participants with

SMA was estimated, so the sample size of 32 participants was adequate for the inferences presented.

### Evaluation instruments and procedures

The evaluation was carried out in person, after signing the two forms. In the first stage, an interview was conducted with the participation of individuals with SMA and their caregivers, aiming to answer the questionnaire of identification, anamnesis, and sociodemographic profile. Individually and in a private place, without the presence of the participant with SMA, the primary family caregiver answered the Burden Interview (BI) scale.

Motor function was assessed using the motor function measurement (MFM-32) scale or motor function measurement – reduced version (MFM-20) scale according to age group and in the presence of the caregiver.

### Burden Interview (BI)

The BI is an instrument used to assess the level of burden of caregivers, and its Brazilian version contains 22 items. Each item is scored from 0 to 4, and the total score is obtained by adding all items, ranging from 0 to 88. The higher the score, the higher the level of burden<sup>10</sup>. The level of burden is classified according to the total score as follows: little or no burden ( $\leq 21$  points), mild to moderate burden (21–40 points), moderate to severe burden (41–60 points), and severe burden ( $\geq 61$  points)<sup>11</sup>.

### Motor function measurement scale

Motor function was assessed according to age group. Participants under six years of age were evaluated using the MFM-20, a reduced version of the MFM-32, which presents 20 items selected from the original scale<sup>12</sup>. Those older than six years were evaluated by MFM-32, a scale composed of 32 items divided into three dimensions, the first referring to the standing position and transfers (D1), the second to the axial and proximal motor function (D2), and the third to the distal motor function (D3). The total score of the two versions is expressed by accounting for the percentage of the maximum score obtained. The higher the score, the better the motor function<sup>5</sup>.

### Statistical analysis

The profile of the individuals was characterized by absolute and relative frequency for the categorical

variables and mean and standard deviation for the continuous variables. Data parametricity was verified by a normalized Q-Q plot and histogram of standardized residuals<sup>13</sup>. The distribution of the demographic and clinical profile was compared according to the type of SMA by applying the analysis of standardized residuals "Post hoc"<sup>14</sup>. Motor function and caregiver burden were compared between the types of SMA through the analysis of covariance (ANCOVA), controlling for the effect of the variables that presented  $p < 0.05$  in the initial exploratory analyses and Pearson's chi-square test. The motor function and caregiver burden variables were correlated using Pearson's correlation test, and the correlation coefficient was classified as low, moderate, strong and very strong<sup>15</sup>. The data were analyzed using

the Statistical Package for Social Science (SPSS), version 26.0 and a 5% significance level was adopted ( $p < 0.05$ ).

## RESULTS

The sample consisted of 32 participants with SMA, the age ranged from 1 to 59 years, 17 (53.1%) were male. Individuals were categorized into three groups according to the type of disease, with type III being the most frequent, with 17 participants, followed by type II, with nine, and type I, with six. Table 1 shows the sociodemographic characterization and clinical profile data.

Table 1. Sociodemographic and clinical profile of participants with spinal muscular atrophy (n=32)

Parameter	Type of spinal muscular atrophy			Total	p*
	Type I (n=6)	Type II (n=9)	Type III (n=17)		
<b>Sex</b>	n (%)	n (%)	n (%)	n (%)	
Female	2 (33.3)	7 (77.8)	6 (35.3)	15 (46.9)	0.09
Male	4 (66.7)	2 (22.2)	11 (64.7)	17 (53.1)	
<b>Age group</b>					
1-5 years	4 (66.7) <sup>†</sup>	1 (11.1)	1 (5.9)	6 (18.8)	<0.01
6-18 years	2 (33.3)	6 (66.7) <sup>†</sup>	3 (17.6)	11 (34.4)	
>18 years	0 (0.0)	2 (22.2)	13 (76.5) <sup>†</sup>	15 (46.9)	
<b>Funcional capacity</b>					
Unable to sit	6 (100.0) <sup>†</sup>	2 (22.2)	2 (11.8)	10 (31.3)	<0.01
Remains seated	0 (0.0)	7 (77.8) <sup>†</sup>	10 (58.8)	17 (53.1)	
Walks	0 (0.0)	0 (0.0)	5 (29.4)	5 (15.6)	
<b>Feeding route</b>					
Gastrostomy	5 (83.3) <sup>†</sup>	1 (11.1)	0 (0.0)	6 (18.8)	<0.01
Oral	0 (0.0)	7 (77.8) <sup>†</sup>	17 (100.0) <sup>†</sup>	24 (75.0)	
Oral - Modified consistency	1 (16.7)	1 (11.1)	0 (0.0)	2 (6.3)	
<b>Ventilatory support</b>	6 (100.0) <sup>†</sup>	8 (88.9) <sup>†</sup>	5 (29.4)	19 (59.4)	0.01
<b>Presence of caregiver</b>	6 (100.0)	9 (100.0)	12 (70.6)	27 (84.4)	0.07

Data expressed as frequency (n) and percentage (%). \* Pearson's chi-square; † Post hoc. Considering the statistical significance level of  $p < 0.05$ . NA (not applicable)

We observed a statistical difference in the age group between individuals younger than 5 years (type I) and older than 18 years (type III). When analyzing functional capacity, all type I participants (n=6) were unable to remain seated independently, in type II seven (77.8%) were able to remain seated independently and only five (29.4%) participants with type III SMA remained able to walk.

Considering the clinical aspects, the most frequent feeding route was oral for type II and III, and gastrostomy for type I. The need for ventilatory support was prevalent in type I and II.

We also assessed 27 caregivers, 26 (96.3%) female, 20 (74.1%) mothers and 24 (88.9%) dedicated to the care of the home and of the person with SMA. Table 2 shows the sociodemographic characteristics of these caregivers.

Table 2. Caregiver profile according to the type of spinal muscular atrophy (n=27)

	Type of spinal muscular atrophy			Total	p <sup>*</sup>
	Type I (n=6)	Type II (n=9)	Type III (n=12)		
	<i>Mean±SD</i>	<i>Mean±SD</i>	<i>Mean±SD</i>	<i>Mean±SD</i>	
<b>Hours dedicated to care</b>	22±4.9	24±0.0	18.0±6.6	20.9±5.5	<b>0.04</b>
Age	31.3±5.4	42.8±10.8	41.8±11.5	39.8±10.9	0.09
<b>Sex</b>	<i>n (%)</i>	<i>n (%)</i>	<i>n (%)</i>	<i>n (%)</i>	
Female	6 (100.0)	9 (100.0)	11 (91.7)	26 (96.3)	0.52
Male	0 (0.0)	0 (0.0)	1 (8.3)	1 (3.7)	
<b>Relationship</b>					
Mother	5 (83.3)	9 (100.0)	6 (50.0)	20 (74.1)	
Father	0 (0.0)	0 (0.0)	1 (8.3)	1 (3.7)	
Aunt	1 (16.7)	0 (0.0)	0 (0.0)	1 (3.7)	0.12
Wife	0 (0.0)	0 (0.0)	3 (25.0)	3 (11.1)	
Sister	0 (0.0)	0 (0.0)	2 (16.7)	2 (7.4)	
<b>Marital status</b>					
With partner	5 (83.3)	7 (77.8)	9 (75.0)	21 (77.8)	0.92
Without partner	1 (16.7)	2 (22.2)	3 (25.0)	6 (22.2)	
<b>Profession</b>					
Homemaker	6 (100.0)	9 (100.0)	9 (75.0)	24 (88.9)	0.12
Other	0 (0.0)	0 (0.0)	3 (25.0)	3 (11.1)	
<b>Education level</b>					
Elementary school	2 (33.3)	2 (22.2)	3 (25.0)	7 (25.9)	0.96
High school	3 (50.0)	4 (44.4)	6 (50.0)	13 (48.1)	
Higher education	1 (16.7)	3 (33.3)	3 (25.0)	7 (25.9)	
<b>Household income</b>					
1 minimum wage	3 (50.0)	2 (22.2)	3 (17.6)	8 (25.0)	0.49
2 to 3 minimum wages	2 (33.3)	5 (55.6)	12 (70.6)	19 (59.4)	
4 or more minimum wages	1 (16.7)	2 (22.2)	2 (11.8)	5 (15.6)	

Data expressed as mean, standard deviation (±SD), frequency (n) and percentage (%). \* Pearson's chi-square; † Post hoc; †† ANCOVA. Considering the statistical significance level of p<0.05.

The sample of caregivers was smaller than that of participants with SMA, since five patients with the disease reported living alone or not requiring help for performing daily activities.

Regarding the motor function of participants with SMA and level of burden of caregivers, these variables

were also analyzed according to the type of SMA, based on the dimensions of the motor function instrument and the total score, and the total score and classifications for the caregiver burden instrument. These data are described in Table 3.

Table 3. Motor function and caregiver burden according to the type of spinal muscular atrophy

	Type I n=6	Type II n=9	Type III n=17	Total n=32	p <sup>*</sup>
<b>Motor function (MFM)</b>					
D1 - Stand and transfer	1.4±2.2	2.4±2.7	19.1±28.4 <sup>†</sup>	11.1±22.2	<b>&lt;0.001</b>
D2 - Axial and proximal	22.7±12.1	53.1±22.5 <sup>†</sup>	67.6±28.4 <sup>†</sup>	55.1±29.4	<b>0.009</b>
D3 - Distal motor	26.8±19.1	67.5±19.2 <sup>†</sup>	79.9±28.1 <sup>†</sup>	66.4±31.1	<b>0.028</b>
Total score	15.0±8.3	35.6±13.2	50.6±25.3 <sup>†</sup>	39.7±24.0	<b>0.002</b>
<b>Burden Interview</b>					
Little or none (≤21)	1 (16.7)	0 (0.0)	1 (8.3)	2 (7.4)	0.47
Mild to moderate (21 to 40)	4 (66.7)	7 (77.8)	5 (41.7)	16 (59.3)	
Moderate to severe (41 to 60)	1 (16.7)	2 (22.2)	4 (33.3)	7 (25.9)	
Severe (>60)	0 (0.0)	0 (0.0)	2 (16.7)	2 (7.4)	
Total score	36.0±13.3	38.2±12.7	42.1±17.1	39.4±14.6	0.34

Data expressed as mean and standard deviation (±SD) and frequency (n) and percentage (%). \* ANCOVA and Pearson's chi-square; † Tukey's test. Considering the statistical significance level of p<0.05.

In the analysis of motor function, participants with type I showed greater impairment and those with type III lower impairment, considering all dimensions and the total score. Dimension three of MFM related to distal motor function presented higher scores in the three types of SMA when compared to the other dimensions of the instrument.

Caregiver burden showed no significant difference between the types of SMA: however, most caregivers were classified with some level of burden, with 25.9% presenting moderate to severe burden and 59.3% presenting mild to moderate burden.

We observed no correlation between motor function by MFM and caregiver burden assessed through the Burden Interview. These data are shown in Table 4

Table 4. Correlation between motor function and caregiver burden according to the type of spinal muscular atrophy

	Burden Interview		
	Type I	Type II	Type III
D1 - Stand and transfer	r=-0.15 p=0.78	r=-0.27 p=0.48	r=0.25 p=0.44
D2 - Axial and proximal motor function	r=0.62 p=0.19	r=-0.30 p=0.43	r=0.54 p=0.07
D3 - Distal motor function	r=0.40 p=0.44	r=-0.64 p=0.06	r=0.39 p=0.21
Total score	r=0.53 p=0.28	r=-0.42 p=0.26	r=0.46 p=0.13

D1, D2 and D3: Dimension 1, 2 and 3, respectively. MFM: Motor Function Measurement. r: Pearson correlation. A statistical significance level of  $p < 0.05$  was considered.

## DISCUSSION

When analyzing the sociodemographic and clinical profile of participants with SMA, it was found that among the 32 participants evaluated, 53.1% were male and 46.9% were older than 18 years. Sample distribution by types of SMA showed a lower frequency in participants of type I ( $n=6$ ), higher for type III ( $n=17$ ) and males. The variation in the age group (5 to 59 years) and the smaller number of participants with type-I SMA was also verified in the study by Vuillerot et al.<sup>16</sup>

Functional classification was also used in a study with 101 participants with SMA, which verified a large part of the sample unable to sit (45.5%) and only 15.8% with the ability to walk<sup>17</sup>, partially corroborating our study, in which 15.6% also had the ability to walk, but most of the sample in our study had the ability to sit (53.1%).

The frequent use of gastrostomy and the need for ventilatory support in type I patients were also found in a study that evaluated 49 individuals with SMA, 15 of whom were type I, and observed that all required ventilatory support and 93.3% used gastrostomy<sup>18</sup>.

When analyzing the caregiver profile, most were female, with a mean age of 39.8 ( $\pm 0.9$ ) years, 77.8% with a partner, mothers and who were fully dedicated to care, of both family and home, corroborating Aranda-Reneo I et al.,<sup>19</sup> who analyzed the burden of 68 informal caregivers

of individuals with SMA, 81% female, with a mean age of 39.9 ( $\pm 9.1$ ) years, 73% of whom were married. The average Burden Interview score found in our study also corroborates Aranda-Reneo I et al.<sup>19</sup>, who verified an average of 31.9 ( $\pm 16.5$ ) points and found no association between this variable and the types of SMA.

In Europe, researchers also evaluated the level of burden of caregivers to individuals with SMA using the same instrument, but the findings were not stratified by type. Eleven caregivers were evaluated in the United Kingdom, 16 in France, 25 in Germany, resulting in 26.6 ( $\pm 13.3$ ), 40.3 ( $\pm 16.1$ ) and 21.3 ( $\pm 18.3$ ) points, respectively<sup>20</sup>. The values of this study were similar to those found in France.

Our hypothesis was refuted, as we believed that caregivers of individuals with SMA types I and II had higher levels of burden when compared with type III. Burden may not be associated only with the type of disease, but also with other factors such as: lack of information, financial support, difficulty in accessing services or adequate support. Parents in the position of caregivers often prioritize the needs of their children, which may result in damage to their own health and increased level of burden<sup>8</sup>. We found in this study that most caregivers are mothers who dedicate 20.9 ( $\pm 5.5$ ) hours a day to care, 88.9% main occupation is homemaking, 96.3% live with the participant and 59.4% have a monthly income of two to three minimum wages

Most caregivers evaluated presented some level of burden, with 25.9% having moderate to severe burden, in addition to dedicating more than 20 hours to care. These findings highlight the reality of these caregivers and the importance of thinking about strategies to improve their well-being. Sociocultural factors are modifiable aspects that can be considered in the health care strategy, thus making health care professionals protagonists due to their fundamental role in helping improve the quality of life through awareness, intervention and evaluation strategies to determine the need for support, which may be physical, social, financial, or psychological support<sup>21</sup>.

The study by Vuillerot et al.<sup>16</sup> found higher values when analyzing motor function through MFM in 112 participants with SMA, aged 5 to 59 years, contrasting with our findings. However, there was a similarity in the results when verifying that the values for participants with SMA type III are higher when compared to the other types and that dimension three of MFM presents higher values when compared to the other dimensions of the scale.

There was no correlation between motor function and the level of caregiver burden in our findings. A study carried out in Chile<sup>22</sup> with 50 caregivers of individuals with SMA sought to highlight the relationship between caregiver burden and functional capacity using the classification unable to sit, able to sit, and able to walk, concluding that individuals with greater motor impairment had more burdened caregivers, disagreeing with our findings; however, this study did not use an assessment scale such as MFM, and did not highlight caregiver characteristics that could influence the level of burden, as well as the participant's profile.

In the literature, there are few studies that correlate these characteristics for individuals with SMA, mainly using the same instruments, but in other populations these characteristics have been studied. Two studies<sup>23,24</sup> evaluated MFM and Burden Interview as assessment instruments to analyze the relation of motor function with the level of caregiver burden in individuals with Duchenne Muscular Dystrophy, but no correlations were found in any of them. Therefore, it is possible to question whether the motor function instrument is relevant to infer caregiver burden; thus, further studies are required to elucidate this issue.

The limitations of this study involve a variable age group, as we sought to include as many participants as possible with this rare clinical condition. However, it is known that the time since the onset of the disease can

influence burden, just as care in adulthood is different from care in childhood or adolescence. Due to the variation in age group, it was also necessary to use MFM-32 and MFM-20 to assess motor function—studies with this combination are still scarce in the literature.

## CONCLUSION

Individuals with type III SMA have the best performance in motor function, and the dimension referring to distal motor function was better scored in the three types of SMA. There was no correlation between caregiver burden and motor function. However, most caregivers evaluated presented some level of burden, ranging in a lower proportion from moderate to severe and in a higher prevalence from mild to moderate, which implies special attention from professionals who deal directly with these caregivers.

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