Functional and social burden of spinal muscular atrophy type 3 in Brazil: a mixed-methods patient-centered study

Cristina Guimarães Rodrigues[1], **Vanessa Nicolao**[2], **Flávia Sauer Tobaruella**[3], **Juliana Coleridge**[3], **Nayara Gerez**[3], **Gustavo Magno Baldin Tiguman**[3]

1. Grupo de Economia da Saúde e Criminalidade (GEESC), Cedeplar, Federal University of Minas Gerais, Minas Gerais, Brazil; 2. Proclame - Instituto de Apoio e Cuidados às Pessoas com Atrofia Muscular Espinhal, Brazil; 3. Produtos Roche Químicos e Farmacêuticos S.A., São Paulo, Brazil.

PCR64

Introduction

Spinal Muscular Atrophy (SMA) is a progressive neuromuscular disease characterized by cumulative loss of strength and escalating limitations and is classified by type (0-4), according to the motor milestones achieved[1,2]. In Brazil, reimbursement of disease modifying treatments is unavailable for patients with SMA type 3 (SMA3), which highlights access inequalities and underestimates the disease burden[3].

Objective

This study assessed the functional and social burdens of SMA3 from the perspective of adult patients and pediatric patients (represented by their caregivers).

Methods

A patient-centered, mixed-methods observational study utilized the Nominal Group Technique (NGT), adapted for virtual application[4], involving 13 participants (8 adults, 5 caregivers). Disease-related challenges were defined through consensus and analyzed using the WHO International Classification of Functioning, Disability and Health (ICF)[5] framework and descriptive statistics.

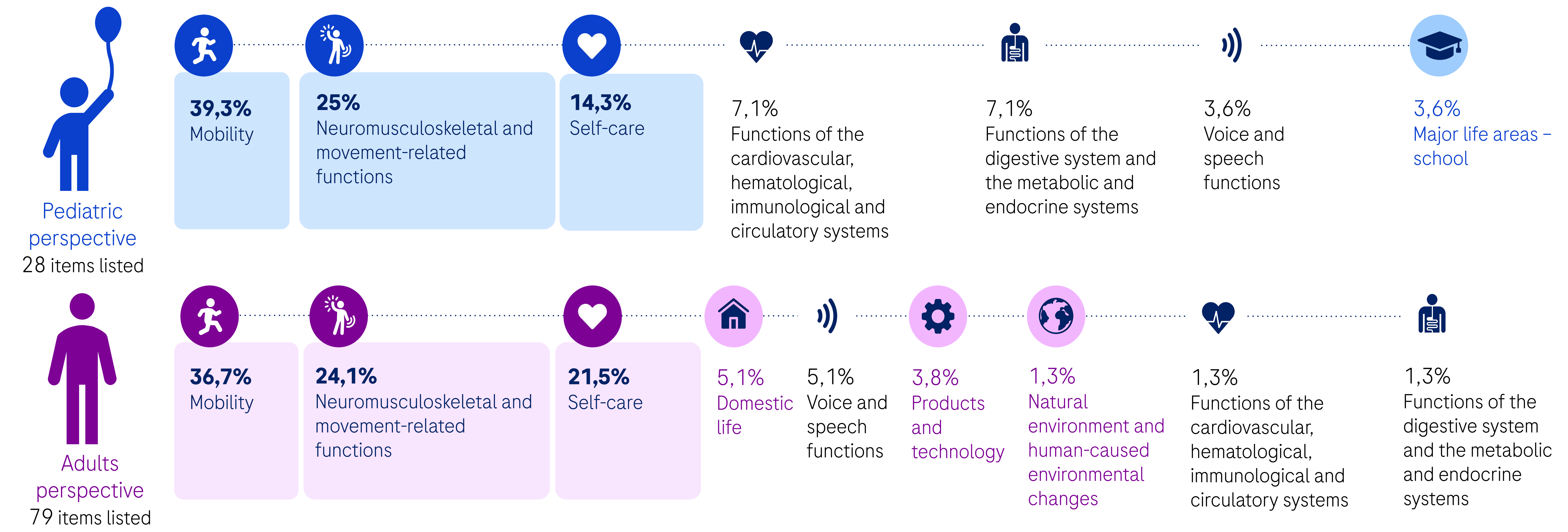
Results

Functional analysis (Figure 1) revealed that 'Mobility' (39.3% pediatric; 36.7% adult) and 'Neuromusculoskeletal Functions' (25% pediatric; 24.1% adult) were the most frequently reported difficulties. Furthermore, 'Self-care' represented 21.5% of adult-reported items, significantly exceeding the 14.3% reported by caregivers. Notably, three-quarters (75%) of adult participants required a wheelchair. Social impacts (Figure 2) on pediatric patients were heavily concentrated in the psychosocial dimension, with 'Mental Functions' (Emotional) comprising 50% of concerns, followed by 'Interactions/Relationships' (25%). In contrast, adult social impacts were mostly related to 'Community, Social and Civic Life' (14.1%), followed by environmental factors related to 'Products and Technology' (12.5%) such as accessibility barriers in public spaces, transportation, and healthcare services, 'Interactions and Relationships' (10.9%), and 'Work and Employment' (9.4%).

Conclusions

SMA3 patients face a substantial functional and social burden in Brazil. While motor impairment remains the hallmark of the disease, it also translates into severe emotional barriers for children and adds profound structural and socioeconomic hurdles for adults. These results may guide health policy decision makers in resource planning toward comprehensive support that addresses the unmet needs faced by Brazilian patients with SMA3.

Figure 1. Results of adults and pediatric patients perspectives of ICF groups representing functional and daily life limitations of SMA3.



Pediatric perspective
28 items listed

39,3%
Mobility

25%
Neuromusculoskeletal and movement-related functions

14,3%
Self-care

7,1%
Functions of the cardiovascular, hematological, immunological and circulatory systems

7,1%
Functions of the digestive system and the metabolic and endocrine systems

3,6%
Voice and speech functions

3,6%
Major life areas – school

Adults perspective
79 items listed

36,7%
Mobility

24,1%
Neuromusculoskeletal and movement-related functions

21,5%
Self-care

5,1%
Domestic life

5,1%
Voice and speech functions

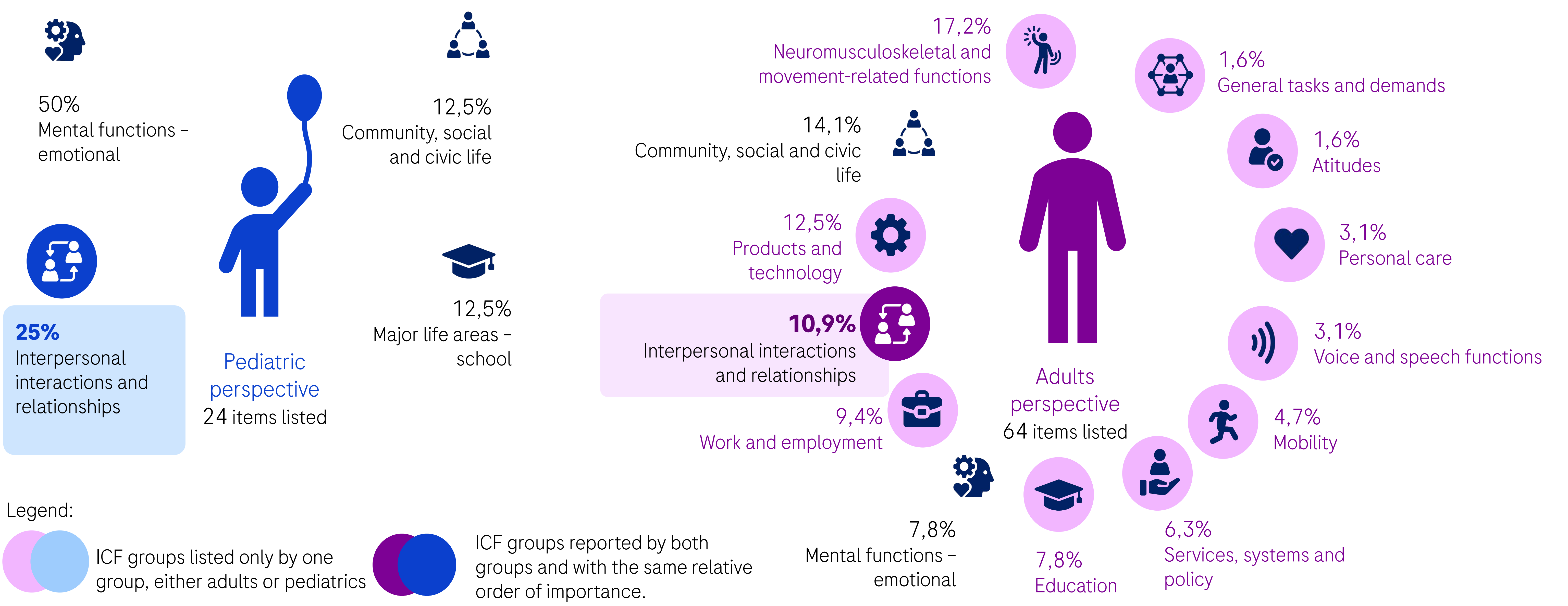
3,8%
Products and technology

1,3%
Natural environment and human-caused environmental changes

1,3%
Functions of the cardiovascular, hematological, immunological and circulatory systems

1,3%
Functions of the digestive system and the metabolic and endocrine systems

Figure 2. Results of adults and padiatric patients perspectives of ICF groups representing social and different areas of life impact of SMA3.



50%
Mental functions – emotional

12,5%
Community, social and civic life

25%
Interpersonal interactions and relationships

Pediatric perspective
24 items listed

12,5%
Major life areas – school

17,2%
Neuromusculoskeletal and movement-related functions

14,1%
Community, social and civic life

12,5%
Products and technology

10,9%
Interpersonal interactions and relationships

9,4%
Work and employment

7,8%
Mental functions – emotional

7,8%
Education

6,3%
Services, systems and policy

4,7%
Mobility

3,1%
Voice and speech functions

3,1%
Personal care

1,6%
Atitudes

1,6%
General tasks and demands

Adults perspective
64 items listed

Legend:

ICF groups listed only by one group, either adults or pediatrics

ICF groups reported by both groups and with the same relative order of importance.

References: 1. D'Amico A, et al. Spinal muscular atrophy. Orphanet J Rare Dis. 2011 Nov 2;6(1):71.2. Butchbach MER. Copy Number Variations in the Survival Motor Neuron Genes: Implications for Spinal Muscular Atrophy and Other Neurodegenerative Diseases. Front Mol Biosci [Internet]. 2016 Mar 10 [cited 2025 Jan 6]. Available from: https://www.frontiersin.org/journals/molecular-biosciences/articles/10.3389/fmolb.2016.00007/full; 3. Protocolo Clínico e Diretrizes Terapêuticas de Atrofia Muscular Espinhal AME 5q Tipos I e II. – Comissão Nacional de Incorporação de Tecnologias no Sistema Único de Saúde - CONITEC [Internet]. [cited 2025 Jan 6]. Available from: https://www.gov.br/conitec/pt-br/midias/consultas/relatorios/2023/protocolo-clinico-e-diretrizes-terapeuticas-de-atrofia-muscular-espinhal-ame-5q-tipos-i-e-ii/view. 4. Gusmão VCDL, et al. Adaptação da técnica de grupo nominal para versão virtual: relato de experiência. Rev Esc Enferm USP. 2024;58:e20230298. 5. World Health Organization. International classification of functioning, disability and health: ICF. Geneva: WHO. 2001.