MEASURING THE EXPERIENCE OF LIVING WITH SPINAL MUSCULAR ATROPHY: THE ROLE OF THE SMA INDEPENDENCE SCALE

Vazquez-Costa JF[1], Brañas Pampillon M[2], Pitarch I[1], Lopez M[3], Medina J[4], Povedano M[5], Fernandez Ramos JA[6], García MC[7], Rojas Garcia R[8], Pascual SI[9], Málaga I[10], Eiris J[11], De Lemus M[12], Cattinari MG[12], Cabello R[2], Diaz P[2], Terrancle M[2], Maurino J[2], Rebollo P[13], Madruga M[14]

[1]Hospital La Fe, Valencia, Spain, [2]Medical Department, Roche Farma, Madrid, Spain, [3]Hospital Universitario Virgen del Rocio, Sevilla, Spain, [4]Hospital Sant Joan de Déu, Barcelona, Spain, [5]Functional Unit of Amyotrophic Lateral Sclerosis (UFELA), Service of Neurology, Bellvitge University Hospital, L'Hospitalet de Llobregat, Spain, [6]Hospital Reina Sofía, Córdoba, Spain, [7]Hospital Miguel Servet, Zaragoza, Spain, [8]Hospital Sant Pau, Barcelona, Spain, [9]Hospital Universitario La Paz, Madrid, Spain, [10]Hospital Universitario Central de Asturias, Oviedo, Spain, [11]Hospital Universitario de Santiago, Santiago de Compostela, Spain, [12]Fundacion Atrofia Muscular Espinal España, Madrid, Spain, [13]Iqvia, Madrid, Spain, [14]Hospital Viamed Santa Ángela de la Cruz, Sevilla, Spain

BACKGROUND

- Spinal Muscular Atrophy (SMA) is genetic disorder characterized by motor neuron loss in the spinal cord and lower brainstem, leading to progressive proximal muscle weakness and atrophy.[1,2]
- SMA is classified in 4 different phenotypes (1-4) according to the age of symptoms onset and the highest motor milestone achived.[3,4]
- Although several scales evaluate the motor function of SMA patients, the experience of living with SMA has been insufficiently explored.[5,6,7]

OBJECTIVE

- This study aims to assess the disease impact from the patients´ perspective and to analyze the psychometric characteristics of the SMAIS.

MATERIALS AND METHODS

Study design and patients

- This is an ongoing multicenter, non-interventional, prospective study conducted in 12 Spanish hospitals specialized in the management of patients with SMA.[9]
- Patients ≥2 years with genetic confirmation of SMA were included.
- An interim analysis is described here.

Procedures and endpoints

- Baseline sociodemographic and clinical characteristics were described in two groups according to subjects' age: over 12 years (G1) and younger patients (G2).
- The Clinical Global Impressions Severity (CGI-S) scale of illness is used to overall assess the current severity of the patient's symptoms.
- The SMA Independence Scale (SMAIS) is a new self-reported questionnaire to assess the amount of assistance patients require to perform daily activities.[8]
- Higher SMAIS scores indicate greater independence (range: 0-44). Analysis of reliability, construct validity and concept validity of the SMAIS was performed.
- Patients over 12 years (G1) answered patient version and parents of younger patients (G2) answered caregiver's version of the SMAIS.

RESULTS

Baseline sociodemographic and clinical characteristics

- 119 patients were recruited until April 14, 2021. (Table 1). Mean age was 20.4 years and 28.6% were ambulant.
- 34 patients (28.6%) could walk independently and 4 (3.4%) had swallowing or feeding difficulties.

Table 1. Baseline characteristics

Characteristic	≥12 years (n=76)	<12 years (n=43)	Total (n=119)
Age, years, mean (SD)	28.4 (14.6)	6.2 (2.8)	20.4 (15.9)
Sex, male, n (%)	42 (55.3%)	26 (60.5%)	68 (57.1%)
Time since SMA diagnosis, years, mean (SD)	16.0 (11.3)	5.0 (2.7)	12.0 (10.6)
Type of SMA, n (%)			
Ib	-	4 (9.3%)	4 (3.4%)
Ic	-	5 (11.6%)	5 (4.2%)
II	34 (44.7%)	26 (60.5%)	60 (50.4%)
IIIa	15 (19.7%)	8 (18.6%)	23 (19.3%)
IIIb	23 (30.3%)	-	23 (19.3%)
IV	4 (5.3%)	-	4 (3.4%)
Patients receiving ventilatory support, n (%)	18 (23.7%)	17 (39.5%)	35 (29.4%)
Time since ventilatory support, years, mean (SD)	12.1 (6.0)	4.2 (2.3)	8.3 (6.1)
History of scoliosis surgery, n (%)	32 (42.1%)	2 (4.7%)	34 (28.6%)
Patients receiving active drug for SMA, n (%)	56 (73.7%)	42 (97.7%)	98 (82.4%)
Time since initiated active drug, years, mean (SD)	1.6 (0.8)	2.6 (1.0)	2.0 (1.1)
Clinical Global Impressions-Severity of Illness, n (%)			
1-Normal, not at all ill	6 (7.9%)	5 (11.6%)	11 (9.2%)
2-Borderline ill	8 (10.5%)	5 (11.6%)	13 (10.9%)
3-Mildly ill	14 (18.4%)	12 (27.9%)	26 (21.8%)
4-Moderately ill	27 (35.5%)	17 (39.5%)	44 (37.0%)
5-Markedly ill	17 (22.4%)	4 (9.3%)	21 (17.6%)
6-Severely ill	4 (5.3%)	-	4 (3.4%)

SD, standard deviation; SMA, Spinal Muscular Atrophy

SMAIS and CGI-S scores

- Mean (SD) SMAIS G1 and G2 total scores were 31.0 (12.8) and 28.2 (8.3).
- As expected, higher SMAIS scores were found in patients with SMA types IIIb and IV vs type II or IIIa, as well as with the increasing severity of the CGI-S scores.

Figure 1. SMAIS - Patient version scores (0-100) according to CGI-S and type of SMA

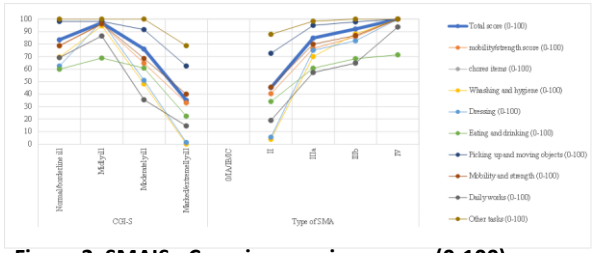
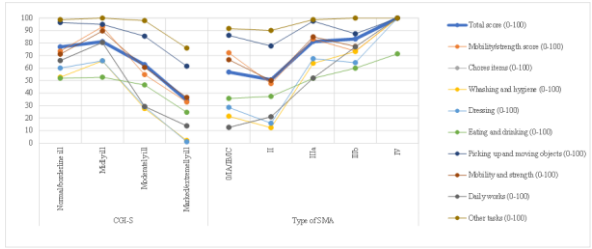


Figure 2. SMAIS - Caregiver version scores (0-100) according to CGI-S and type of SMA



Psychometric characteristics of the SMAIS

- Reliability of SMAIS was high (Cronbach's alpha 0.96).
- A factorial analysis showed a unidimensional structure, except for the mobility/strength item.

CONCLUSIONS

- This study provided insights about the impact of SMA in physical, psychological and social areas of patient's life.
- SMAIS showed adequate psychometric properties and may constitute a complementary tool to assess patients with SMA.

REFERENCES

1. Farrar MA, et al. *Neurotherapeutics*. 2015;12(2):290–302.
2. Kolb SJ, et al. *Neurol Clin* 2015;33:831-46
3. Finkel RS, et al. *Neuromuscul Disord.* 2017;27(6):596–605.
4. Talbot K, et al. *Gene Ther*. 2017;24(9):529–33.
5. McGraw S, et al. *BMC Neurol*. 2017;17(1):68.
6. Mongiovi P, et al. *Neurology*. 2018;91(13):e1206-e14.
7. de Lemus M, Rebollo P, Cattinari MG, et al. *SMA Europe Second International Congress*, Paris-Evry 2020. Abstract 00122
8. Trundell D, Skalicky A, Hareendran O, et al. *2019 Annual SMA Conference*, June 28–July 1, 2019, Anaheim, USA. USA: Anaheim; 2019.
9. Madruga-Garrido M, et al. *Neurol Ther*. 2021; 10(1):361-373.

ACKNOWLEDGEMENTS

The authors would like to thank parents and children with SMA whose support and collaboration are making the SMA-TOOL study possible.

DISCLOSURES

This study was funded by Roche Farma Spain. MB, RC, PD, MT and JM are employees of Roche Farma Spain. None of the other authors report any conflict of interest

ISPOR 2021, 30 November - 3 December 2021, Virtual

Copyright © 2021 juan.vazquez.neuro@gmail.com