Assessing the impact of "any point differences" or "stability" in the Hammersmith Functional Motor Scale–Expanded on activities of daily living in adult patients with SMA

Virtual Poster

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INTRODUCTION

- Spinal muscular atrophy (SMA) is a neuromuscular disease characterized by the irreversible loss of spinal motor neurons and progressive skeletal muscle atrophy, leading to weakness and motor function decline[1]
- Motor function assessments such as the Hammersmith Functional Motor Scale–Expanded (HFMSE) are well-established tools for evaluating treatment efficacy in SMA[2]; however, these measures may not fully capture the broader, clinically meaningful impacts of treatment on daily life[3]
- Insights from patients or their caregivers can contextualize HFMSE changes and enhance the interpretation of what is clinically meaningful[3]
- To better understand how changes in HFMSE scores are perceived in terms of real-world impact, we conducted 60-minute semistructured interviews with adult patients with SMA

OBJECTIVE

- To gain perspectives from adult patients with SMA on how improvement or stability in specific HFMSE tasks would lead to beneficial changes in activities of daily living (ADLs)

METHODS

- 60-minute, web-based, in-depth, qualitative interviews were conducted with adult patients with SMA
- Patients meeting the inclusion criteria were eligible for study enrollment (**Table 1**)
- A semistructured discussion guide was informed by a focused targeted literature review (**Figure 1**)

Table 1. Key eligibility criteria

Key inclusion criteria	Self-reported SMA diagnosis
	Current self-reported motor function ability categorized as a sitter (can sit independently but cannot walk without help/support) or walker (can walk without using any help/support)
	Age of majority in state of residence[a]
	English language proficient
	United States resident
	Had access to technology (eg, smartphone, computer) to participate in interview
	Completed informed consent to participate in the study and to audio recording of the interview discussion

[a]≥18 years of age, except if a Mississippi resident (≥21 years of age) or a resident of Alabama or Nebraska (≥19 years of age).
SMA, spinal muscular atrophy.

Figure 1. Study approach overview

- Performed using PubMed and Google Scholar databases
- Informed the development of study documents (eg, interview discussion guide)
- Conducted to understand clinical meaningfulness in SMA and of select outcome measures

- Developed study documents and submitted to IRB for approval/exemption
  - Study protocol
  - Interview discussion guides

- Conducted semistructured, in-depth interviews with patients
- Data collected during the interviews were coded and synthesized
- Study findings were contextualized using information from the fTLR

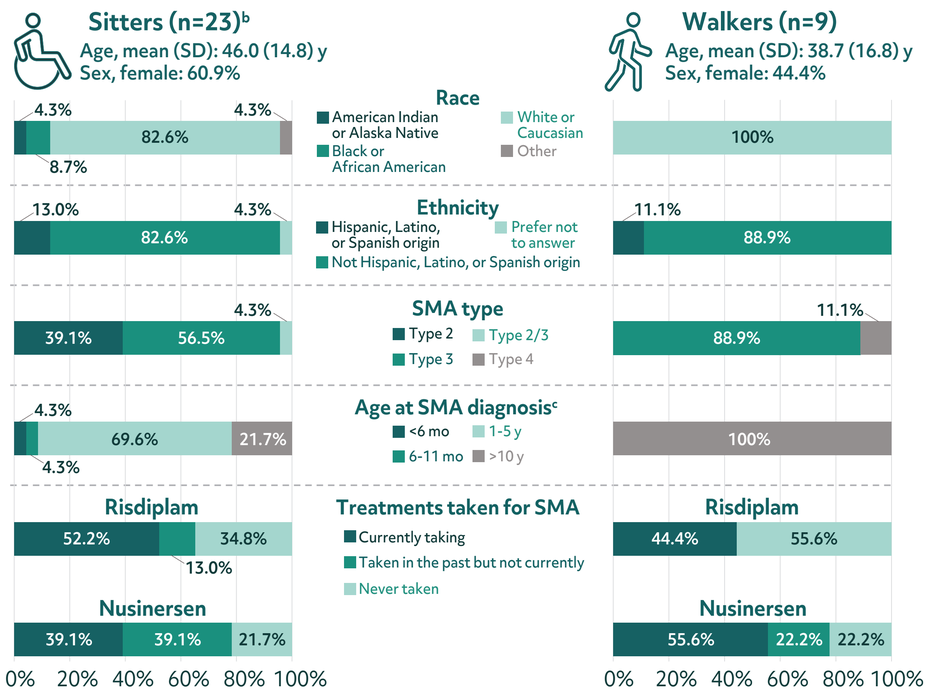
IRB, institutional review board; SMA, spinal muscular atrophy.

- The discussion guide included open-ended questions to elicit and examine perspectives and experiences on:
  - How clinical meaningfulness is interpreted
  - What constitutes meaningful change at the individual item level on the HFMSE
  - How meaningful change in specific motor function abilities may impact the patient's ability to perform ADLs in addition to their psychosocial well-being, independence, and quality of life
- Interview transcripts were analyzed using the constant comparative method, an iterative approach to aggregate and synthesize qualitative data

RESULTS

- Overall, 32 adult patients participated; mean (range) age was 44 (18-72) years, and 56.3% were female (**Figure 2**; **Table S1**)
- Patients' responses generally focused on HFMSE tasks they were familiar with; some patients found it challenging to envision unfamiliar changes (**Table 2**)

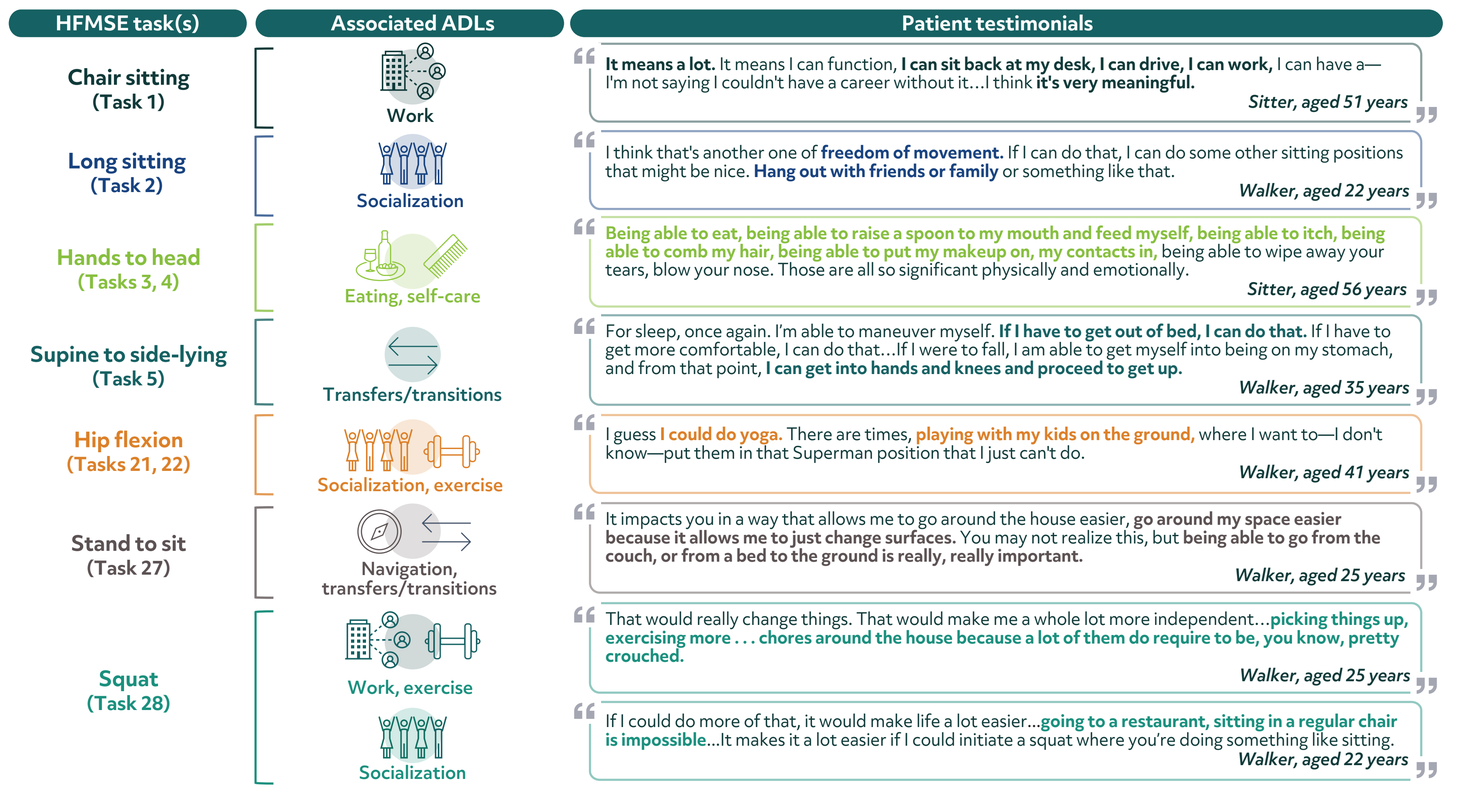
Figure 2. Adult patient demographics and clinical characteristics[a]



[a]All demographic information and clinical characteristics, including SMA type and current motor function ability (eg, sitter or walker) were self-reported by the patients. [b]14/23 (61%) of sitters reported having previously been able to walk without help/support but could not walk without any help/support at the time of the study. [c]None of the patients were diagnosed with SMA at age 6-10 y.
mo, months; SD, standard deviation; SMA, spinal muscular atrophy; y, years.

- Key areas patients associated with improved motor function included ADLs, psychosocial aspects, independence, safety, and quality of life
- Patient responses related to ADLs generally indicated that any point improvements or stabilization within HFMSE tasks can lead to meaningful benefits in daily life
- Patients generally described that being able to gain or maintain the ability to eat, practice self-care, work, socialize, and navigate or transfer would augment their day-to-day activities (**Figure 3**; **Table 3**)
- Please also see Poster 8: "Understanding adult patient perspectives on the clinical meaningfulness of 'any point differences' on the Hammersmith Functional Motor Scale–Expanded in SMA" presented by our group

Figure 3. Example patient perspectives on meaningful changes in ADLs reflected in HFMSE tasks



ADLs, activities of daily living; HFMSE, Hammersmith Functional Motor Scale–Expanded.

Table 2. Number of patients who provided commentary on each HFMSE task grouping

HFMSE item number	Task	Sitters (n=23)	Walkers (n=9)
1	1 Plinth/chair sitting	23	1
2	2 Long sitting, legs straight	23	3
3, 4	3 One hand to head in sitting 4 Two hands to head in sitting	23	4
5-9	5 Supine to side-lying 6, 7 Rolls prone to supine over R/L 8, 9 Rolls supine to prone over R/L	23	7
10, 14	10 Sitting to lying 14 Lying to sitting	21	3
11	11 Props on forearms	9	1
13	13 Props on extended arms	13	7
12, 17	12 Lifts head from prone 17 Lifts head from supine	18	9
15, 16	15 Four-point kneeling 16 Crawling	15	9
18-20	18 Supported standing 19 Unsupported standing 20 Stepping	7	8
21-27	21, 22 Hip flexion in supine (R/L) 23, 24 High kneeling to half kneel (R/L) 25, 26 High kneeling to stand leading with R/L leg 27 Stand to sit	19	9
28, 29	28 Squat 29 Jump 12"	2	9
30-33	30 Ascends stairs with rail 31 Descends stairs with rail 32 Ascend stairs without rail 33 Descends stairs without rail	0	9

HFMSE, Hammersmith Functional Motor Scale–Expanded; R/L, right/left.

Table 3. Mapping some HFMSE tasks to commonly associated patient-reported ADLs

ADL domain	Chair/long sitting (Tasks 1, 2)	Hands to head (Tasks 3, 4)	Rolling (Tasks 5-9)	Transitioning (Tasks 10, 14)	Propping (Tasks 11, 13)	Lifting head (Tasks 12, 17)	Crawling (Tasks 15, 16)	Standing/ stepping (Tasks 18-20)	Transitions/ kneeling (Tasks 21-27)	Squat/jump (Tasks 28, 29)	Stairs (Tasks 30-33)
Eating	✓	✓		✓							
Self-care	✓	✓	✓	✓			✓	✓	✓		
Work/ household tasks	✓	✓					✓	✓	✓	✓	✓
Socializing	✓					✓	✓		✓	✓	✓
Transfers/ transitions	✓		✓	✓	✓	✓	✓	✓	✓	✓	

✓Checkmarks represent ADL domains that patients described as being influenced by improvements or stability in each HFMSE task. This matrix reflects thematic associations from qualitative data rather than any quantification.
ADLs, activities of daily living; HFMSE, Hammersmith Functional Motor Scale–Expanded.

CONCLUSIONS

- There was general consensus among adult patients surveyed that any point improvements or stabilization across HFMSE tasks are generally viewed as meaningful, as they correspond to improvements in daily functioning
- Movements and ADLs that become possible with increased motor function will be perceived as more meaningful as they become part of patients' daily experience
- These findings highlight the value of incorporating patient perspectives into the interpretation of treatment efficacy to provide a more comprehensive definition of what is clinically meaningful

References

1. Mercuri E, et al. *Nat Rev Dis Primers*. 2022;8(1):52.
2. Ramsey D, et al. *PLoS One*. 2017;12(2):e0172346.
3. McGraw S, et al. *BMC Neurol*. 2017;17:68.

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Disclosures

AH has served on advisory boards and/or as a consultant for Biogen, Catalyst, J&J, Sarepta, and Scholar Rock, Inc. **SD** has served on advisory boards for Alexion, argenx, Biogen, CSL Behring, Genetech, Immunovant, and Sarepta. **TD** has served on advisory boards and/or as a consultant for Biogen, Cure SMA, Duchenne UK, Dyne, Genentech, Roche, Scholar Rock Inc., and Trinds. **MCM** is an employee of Precision AQ and owns an equity interest in Precision Medicine Group, the parent company of Precision AQ. **CCagle** is an employee of Precision AQ. **TB**, **CCherubino**, and **MG** are employees and stockholders of Scholar Rock, Inc. **AS-C** has served on an advisory board and/or received consulting fees from Catalyst, Novartis, and Scholar Rock, Inc. **LN** has served as a consultant for Avexis, Biogen, F. Hoffman-La Roche, Novartis, and Scholar Rock, Inc.

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SUPPLEMENTARY MATERIAL

Table S1. Adult patient demographics and baseline characteristics[a]

Characteristic	Full sample (N=32)
Motor function status, n (%)	
Sitter	23 (71.9)[b]
Walker	9 (28.1)
Age, years	
Mean (SD)	44.0 (15.5)
Range	18-72
Sex, n (%)	
Female	18 (56.3)
Male	14 (43.8)
Race, n (%)	
American Indian or Alaska Native	1 (3.1)
Black or African American	2 (6.3)
White or Caucasian	28 (87.5)
Other	1 (3.1)
Ethnicity, n (%)	
Hispanic, Latino, or Spanish origin	4 (12.5)
Not Hispanic, Latino, or Spanish origin	27 (84.4)
Prefer not to answer	1 (3.1)
SMA type, n (%)	
Type 2	9 (28.1)
Type 3	21 (65.6)
Type 4	1 (3.1)
Other: Type 2/3	1 (3.1)
Age at SMA diagnosis, n (%)	
<6 months	1 (3.1)
6 to 11 months	1 (3.1)
1 to 5 years	16 (50.0)
6 to 10 years	0 (0.0)
>10 years	14 (43.8)
Treatments taken for SMA, n (%)	
Risdiplam	
Currently taking	16 (50.0)
Taken in the past but not currently	3 (9.4)
Never taken	13 (40.6)
Nusinersen	
Currently taking	14 (43.8)
Taken in the past but not currently	11 (34.4)
Never taken	7 (21.9)

[a]All demographic information and clinical characteristics, including SMA type and current motor function ability (eg, sitter or walker) were self-reported by the patients. [b]14/23 (61%) of sitters reported having previously been able to walk without help/support but could not walk without any help/support at the time of the study.
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