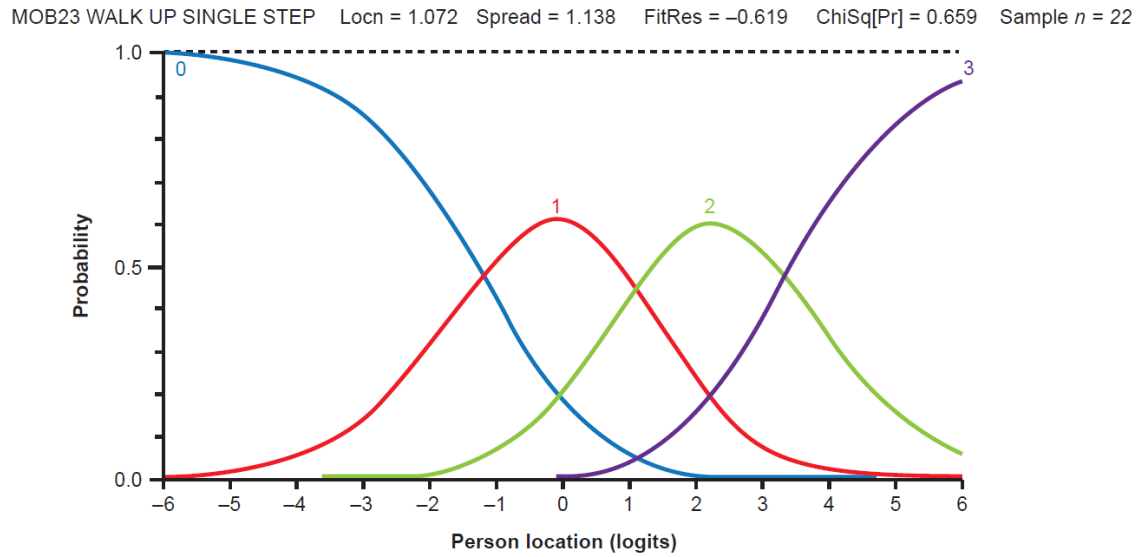


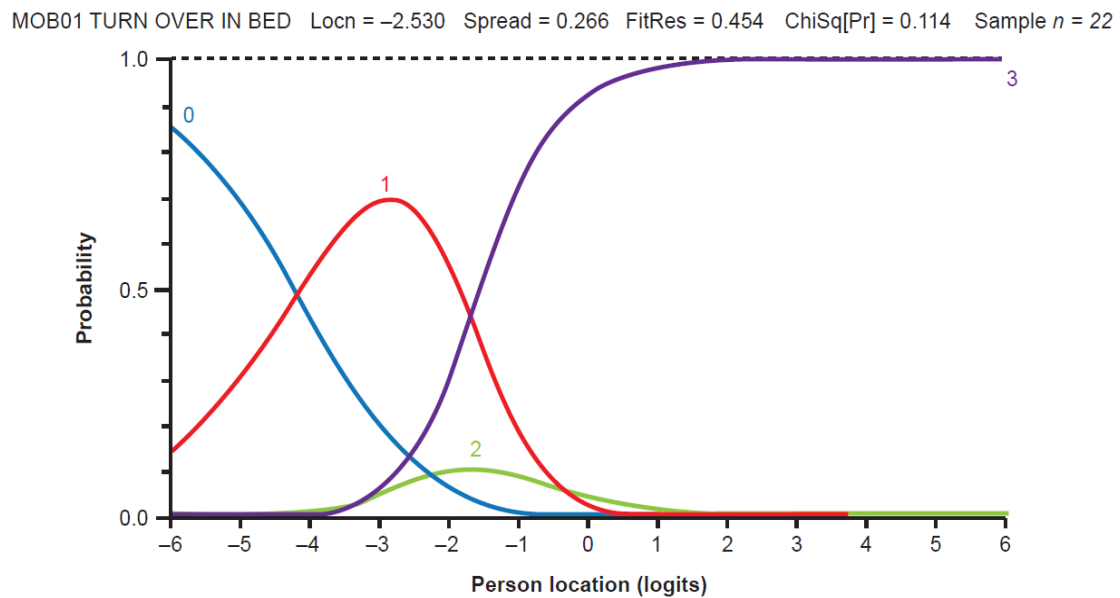
SUPPLEMENTARY DATA

Supplementary Figure 1: Wave 3 mobility items: examples of category probability curves

(A)



(B)



(A) An example of an ordered category probability curve (CPC). (B) An example of a disordered CPC. CPCs show the probability (y axis) of endorsing each response category

(different colored lines) for each level of the construct under measurement on the measurement continuum (x axis). The numbers indicate the degree of independence an individual with SMA has when completing a specific task. 0. 'Cannot do this at all without help'; 1. 'Needs a lot of help/needs a little help'; 2. 'Independent with aids'; 3. 'Independent with no aids'. The 'not applicable' option was counted as missing for the purposes of the RMT analysis. CPC estimation excludes extreme codes, in this case $n = 3$, leaving the total sample of $n = 22$ for this specific analysis.

ChiSq (Pr)=Chi square (probability); FitRes=fit residuals; Locn=location; MOB=mobility item; RMT=Rasch Measurement Theory.

Supplementary Table 1: Demographics of participants included in the RMT analyses

Patients with SMA	RMT sample (<i>n</i> = 10)
Patient age (years)	
Mean age (SD), range	47 (12), 30–69
Mean age at diagnosis (SD), range	17 (12), 1–31
Diagnosis duration (SD), range	30 (13), 7–50
Gender, <i>n</i> (%)	
Male	2 (20)
Female	8 (80)
Ethnicity, <i>n</i> (%)	
White Caucasian	10 (100)
Hispanic	-
Biracial	-
Employment, <i>n</i> (%)	
Part time	1 (10)
Retired	2 (20)
Full time	1 (10)
Unemployed	-
School	-
Unable to work due to disability	2 (20)
Homemaker	2 (20)
Disabled and able to work part time	2 (20)
Student	-
Education, <i>n</i> (%)	
Secondary/High school	2 (20)
Technical or Vocational degree	1 (10)

Some college	1 (10)	
College degree	3 (30)	
Some graduate work	-	
Graduate degree	3 (30)	
Self-reported Ambulatory status		
Ambulant	7	
Near-ambulant	3	
Self-reported Level of assistance needed, <i>n</i> (%)	In the home	Out of the home
I cannot do any activities without help	1 (10)	1 (10)
I need a lot of help	-	3 (30)
I need some help	4 (40)	4 (40)
I need a little bit of help	4 (40)	1 (10)
I do not need any help	1 (10)	1 (10)

Caregivers of patients with SMA	RMT sample (<i>n</i> = 15)
Caregiver age	
Mean age (SD), range	36 (4), 30–44
Caregiver gender, <i>n</i> (%)	
Male	2 (13)
Female	13 (87)
Caregiver relationship, <i>n</i> (%)	
Mother	13 (87)
Father	2 (13)
Grandparent	-
Caregiver ethnicity	

White Caucasian	12 (81)
Latino	-
Asian	2 (13)
Hispanic	-
Biracial/Mixed	1 (6)
Caregiver employment	
Full time	8 (54)
Part time	1 (6)
Retired	-
Homemaker	6 (40)
Caregiver education	
Secondary/High school	-
Technical or Vocational degree	1 (6)
Some college	-
College degree	4 (27)
Some graduate work	-
Graduate degree	10 (67)
Children with SMA (years)	
Mean age (SD), range	4 (2), 2–8
Mean age at diagnosis (SD), range	1 (1), 0–2
Diagnosis duration (SD), range	4 (2), 1–8
Patient gender, <i>n</i> (%)	

Male	10 (66)	
Female	5 (34)	
Caregiver-reported Ambulatory status		
Ambulant	9	
Near-ambulant	6	
Patient level of assistance needed as reported by the caregiver, <i>n</i> (%)	In the home	Out of the home
They cannot do any activities without help	-	2 (13)
They need a lot of help	4 (27)	5 (34)
They need some help	6 (40)	4 (27)
They need a little bit of help	3 (20)	2 (13)
They do not need any help	2 (13)	2 (13)

RMT=Rasch Measurement Theory; SD=standard deviation; SMA=spinal muscular atrophy.

Supplementary Table 2: Tracking matrix following Waves 2 and 3 on the final mobility items

Item number	First draft of mobility items based on CE data	Wave 2 decision	Wave 3 debriefing	Short-term changes post Wave 3 ^a	Final mobility item
1.	Turn over in bed	No change	No change	No change	Turn over in bed
2.	Sit up from lying down	No change	No change	No change	Sit up from lying down
3.	Stand up from a low chair	No change	No change	No change	Stand up from a low chair
4.	Stand/Get up from the floor	No change	No change	No change	Stand/Get up from the floor
5.	Bend over to pick something up from the floor	No change	No change	No change	Bend over to pick something up from the floor
6.	Getting up from a squatting position	No change	No change	No change	Getting up from a squatting position
7.	Getting into bed	No change	No change	No change	Getting into bed
8.	Getting in the bath	No change	No change	No change	Getting in the bath
9.	Getting in the shower	No change	No change	No change	Getting in the shower
10.	Getting onto the toilet	No change	No change	No change	Getting onto the toilet
11.	Step up on a curb	No change	No change	No change	Step up on a curb
12.	Walk up a single step indoors	No change	No change	Modify – item formatting	Walk up a single step indoors
13.	Walk up a flight of stairs	No change	No change	Modify – item formatting	Walk up a flight of stairs
14.	Walk across a room	No change	No change	Modify – item formatting	Walk across a room

15.	Walk from room to room	No change	No change	Modify – item formatting	Walk from room to room
16.	Walk a short distance outside, e.g. from house to car (10–50 feet)	No change	No change	Modify – item formatting	Walk a short distance outside, e.g. from house to car (10–50 feet)
17.	Walk a short distance around your community, e.g. shop to shop	No change	No change	Modify – item formatting	Walk a short distance around your community, e.g. shop to shop
18.	Walk quickly if needed, e.g. to cross a road	No change	No change	Modify – item formatting	Walk quickly if needed, e.g. to cross a road
19.	Walk in a busy, crowded place	No change	No change	Modify – item formatting	Walk in a busy, crowded place
20.	Walk on a flat surface outside	No change	No change	Modify – item formatting	Walk on a flat surface outside
21.	Walk on an uneven surface outside	No change	No change	Modify – item formatting	Walk on an uneven surface outside
22.	Walk on an unstable surface outside, e.g. sand, gravel	No change	No change	Modify – item formatting	Walk on an unstable surface outside, e.g. sand, gravel
23.	Walk on a slippery surface	No change	No change	Modify – item formatting	Walk on a slippery surface
24.	-	Standing in the shower: Flagged as missing concept – new item added	No change	No change	Standing in the shower
25.	-	Walk up an incline: Flagged as missing	No change	Modify – item formatting	Walk up an incline

		concept – new item added			
26.	Stand up from a chair	Modify – item wording to reduce ambiguity	No change	No change	Stand up from a regular height chair
27.	Walk for a mile or more	Modify – item wording to better match the remaining items (i.e. walk short distance items)	No change	Modify – item formatting	Walk a long distance around your community e.g. a mile, a kilometer, or more
28.	-	Walking down an incline: Flagged as missing concept – new item added	Remove – to reduce overlap with inverse item ^b	-	-
29.	Walk on a flat surface indoors	Remove – target concept adequately captured by the item distinction of ‘ in the home’ and ‘out of the home’	-	-	-
30.	Walk in an unfamiliar place/environment	Remove – ambiguity issues	-	-	-
31.	Step onto a bus/train	Remove – not relevant to participants in some geographical areas	-	-	-
32.	Step off a bus/train	Remove – not relevant to participants in some geographical areas	-	-	-
33.	Stand up from a high/raised chair	No change	Remove – to reduce overlap	-	-

			with inverse item ^b		
34.	Getting out of bed	No change	Remove – to reduce overlap with inverse item ^b	-	-
35.	Sit down into a chair	Modify – item wording due to ambiguity regarding chair height	Remove – to reduce overlap with inverse item ^b	-	-
36.	Sit down into a high/raised chair	No change	Remove – to reduce overlap with inverse item ^b	-	-
37.	Sit down into a low chair	No change	Remove – to reduce overlap with inverse item ^b	-	-
38.	Getting out of the bath	No change	Remove – to reduce overlap with inverse item ^b	-	-
39.	Getting out of the shower	No change	Remove – to reduce overlap with inverse item ^b	-	-
40.	Getting off the toilet	No change	Remove – to reduce overlap with inverse item ^b	-	-

41.	Getting out of a car	No change	Remove – to reduce overlap with inverse item ^b	-	-
42.	Step down off a curb	No change	Remove – to reduce overlap with inverse item ^b	-	-
43.	Walk down a single step indoors	No change	Remove – to reduce overlap with inverse item ^b	-	-
44.	Walk down a flight of stairs	No change	Remove – to reduce overlap with inverse item ^b	-	-
45.	Getting into a car	No change	-	Remove – issues with item clarity and quality/fit	-
iADL items					
1.	Wash dishes while standing	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
2.	Standing while preparing meals/cooking	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-

3.	Cleaning the house	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
4.	Mopping/sweeping the floor	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
5.	Making your bed	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
6.	Mowing the lawn	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
7.	Vacuuming the floor	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
8.	Setting the table	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-
9.	Tidying up or putting things/toys away	Remove – lack of conceptual relevance/endorsement frequency information	-	-	-

		ent frequency information			
10.	Unloading the dishwasher	Remove – lack of conceptual relevance/endorsem ent frequency information	-	-	-
11.	Going shopping for groceries	Remove – lack of conceptual relevance/endorsem ent frequency information	-	-	-

No change = item was not modified; New item added = item was added following TAE endorsement in Wave 2; Modify – item wording = item modified based on feedback; Modify – item formatting = the word “walk” was bolded to reduce ambiguity; Remove = item removed based on participant feedback.

^aInitial changes included deletion of 14 items from the previous 41-item set and bolding of the ‘walking’ items.

^bConsidering the qualitative feedback on conceptual overlap, as well as the very strong quantitative evidence on item dependency between mirroring (up/down, in/out) item pairs, it was decided to remove the item considered to be the ‘easiest’ by both participants and TAEs.

CE=concept elicitation; iADL=instrumental activities of daily living; TAE=therapeutic area expert.

Supplementary Table 3: Tracking matrix on the final response options and instructions

Original response options for PRO^a following CE interviews	Wave 2 decision	Wave 3 decision^b	Post Wave 3 item refinement	Final response options
I do not need help	Modified: 'independent with no aids; Independent with aids'	No change	No change	Independent with no aids; Independent with aids
I need some help	No change	Modified: 'I need a little help; I need a lot of help'	No change	I need a little help; I need a lot of help
I cannot do this at all without help	Modified: remove 'without help'	No change	No change	I cannot do this at all
Not applicable	No change	No change	No change	Not applicable
Original instructions following CE interviews (PRO only)^c	Wave 2 decision	Wave 3 decision	Post Wave 3 item refinement	Final instructions
We would like to learn about your level of independence for doing daily activities. Please select the option that best describes the amount of assistance you need from another person or from a supportive aid (e.g. a handrail, walking stick, mobility scooter, etc.) to perform each activity.	Modified: 'the amount of assistance you need from another person or from a supportive aid (e.g. a handrail, walking stick, mobility scooter, etc.)' to 'the amount of assistance you need' and added 'If the question is not relevant (e.g. you do not have a bath in your house),	No change	No change	We would like to learn about your level of independence. Please select the option that best describes the amount of assistance you need to perform each activity. Please think about the past 7 days and pick only one answer per activity. If the question is not relevant (e.g. you do not have a bath

Please think about the past 7 days and pick only one answer per activity.	please choose the not applicable response option.'			in your house), please choose the not applicable response option.
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No change = text was not modified; Modify = text was modified based on feedback.

^aCaregiver response options were identical. ^bResponse options were debriefed in Wave 3 by *n* = 11. ^cCaregiver instructions were identical.

CE=concept elicitation; PRO=patient-reported outcome.

Supplementary Table 4: Final list of supportive aids

Final list of supportive aids
Grabbers
Braces/Orthotics
Walking stick
Walking frame
Wheelchair/mobility scooter
Help of furniture/handrails
Handrail for stairs, shower, bath
Raised chair
Bench in the shower
Non-slip mat
Raised toilet seat
Lift or ramps to get in wheelchair/vehicle
Other