

Table of Outcomes – SMA Delphi Process Round 1

<i>Type of Measure</i>	<i>Outcome Measure</i>	<i>Age</i>	<i>Timing</i>	<i>Scoring Type</i>	<i>Reliability</i> H – High (0.85+) M – Moderate (0.70-0.84) L – Low (<0.70)	<i>Validity</i> H – High (0.85+) M – Moderate (0.70-0.84) L – Low (<0.70)	<i>Proprietary</i>	<i>Other</i>	<i>References</i>
Functional Motor Assessment	Gross Motor Function Measure (GMFM)	5–18 years	45 to 60 minutes	Total and Sub-scores	Inter-rater: H ^{1,2} Intra-rater: H ³ Internal: M-H ⁵ Test-Retest: H ²	Criterion: QMT/HHD – M (0.63-0.86) ⁴ HFMSE – H (0.98) ⁶	Yes – available with author’s written permission.	Sensitivity shown to be good against QMT ⁴ May need equipment.	¹ Iannaccone ST 2002 ² Iannaccone et al. 2003 ³ O’Hagen et al. 2007 ⁴ Nelson et al. 2006 ⁵ Cano et al. 2013 ⁶ Glanzman et al. 2011 Montes et al. 2009 Russell et al. 1989 TREAT-NMD
Functional Motor Assessment	Expanded Hammersmith Scale (HFMSE)	2–45 years	<15 minutes	Ordinal Scoring of 36 items	Intra-rater: H ¹ Internal:M-H ⁹ Test-Retest: H ¹	Content: H ¹⁰ Criterion: GMFM – H (0.98) ² FVC – H (0.87) ² ULM – H ⁵ 6MWT – M-H ^{6,7} TUG – M ⁴	No	Limitation for use in strong type III who can run and participate in age-appropriate activities.	¹ O’Hagen et al. 2007 ² Glanzman et al. 2011 ³ Mercuri et al. 2018

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								Sensitivity unknown. ² Used in Clinical Trials for SMA and Nusinersen. ³ Combined measure of HFMS and GMFM ⁸ .	⁴ Dunaway et al. 2014 ⁵ Mazzone et al. 2011 ⁶ Young et al. 2016 ⁷ Montes et al. 2010 ⁸ Berard et al. 2008 ⁹ Cano et al. 2013 ¹⁰ Pera et al. 2017 Montes et al. 2009
Functional Motor Assessment	Revised Hammersmith Scale (RHS)	Children & Adults	~15 minutes	Scored and Timed sections (Max Score 69)	Internal: H ¹	Criterion: WHO Walking Milestones – H (0.86) ¹	No	Combination measure of HFMSE, NSAA, CHOP INTEND, and WHO Developmental Milestones. ¹	¹ Ramsey et al. 2017

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Functional Motor Assessment	Motor Function Measure (MFM)	2+	~36 Minutes	32 items assessed via Likert Scale	Inter-rater: M-H ^{1,2} Intra-rater: M-H ¹ Internal: H ¹ Test-Retest: H ¹	Criterion: FIM – H (0.91) ¹ Vignos – H (0.91) ¹ Brooke – H (0.85) ¹	No	Measure not specific to SMA ¹ . Regularly recommended alongside HFMSE and GMFM. ⁴ Responsiveness was shown to be good in SMA ³ .	¹ Berard et al. 2008 ² Ruiz-Cortes et al. 2017 ³ Vuillerot et al. 2013 ⁴ Bertini et al. 2005 Montes et al. 2009 TREAT-NMD
Functional Motor Assessment	Egen Klassifikation Scale Revisited (EK2)	Teens-Adults	10 minutes	Ordinal Scoring of 10 items	Inter-rater: H ¹ Intra-rater: H ³ Internal: M-H ⁴	Content: H⁵ Criterion: FVC – M (0.77) ² MMT – H (0.87) ²	No		¹ Steffensen et al. 2002 ² Steffensen et al. 2008 ³ Werlauff & Steffensen 2014 ⁴ Cano et al. 2013 ⁵ Steffensen et al. 2001 Montes et al. 2009 TREAT-NMD
Functional Motor Assessment	Spinal Muscular Atrophy	2+ years	<30 minutes	Composite score of	No Information Available	No Information Available	No		¹ Montes et al. 2015

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	Functional Composite Scale			HFMSE, ULM, 6MWT					
Functional Motor Assessment	Manual Muscle Testing (MMT)	4+ years	15 Minutes	Qualitative Scale (0-5)	Inter-rater: H ^{1*} Intra-rater: H ^{1*}	Criterion: TUG – M (0.816) ³ EK2 – H (0.87) ²	No	No known studies specifically looking at Reliability & Validity in SMA population.	¹ Barr et al. 1991 ² Steffensen et al. 2008 ³ Dunaway et al. 2014 NIH NINDS CDE for SMA *Duchennes or Beckers Muscular Dystrophy
Functional Motor Assessment	Canadian Occupational Performance Measure (COPM)	All Ages	Unknown	Multi-stage Scoring & Interviews	No Information Available	No Information Available	Yes	Website available with additional information.	COPM
Functional Motor Assessment	CHOP-ATEND	~18+	Unknown	Revised Edition of CHOP-INTEND	No Information Available	No Information Available	Unknown	Newly available outcome with very limited published data available.	
	Spinal Muscular Atrophy Health Index	Unknown	Unknown	Quantitative Index	No Information Available	No Information Available	Unknown	Very little published data available.	

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(SMA-HI)									
Functional Motor Assessment	Revised Upper Limb Module for SMA	2+	<10 minutes	Ordinal Scoring of 20 items	Inter-rater: H ¹	Content: H ^{1,2} Criterion: HFMS – M (0.75) ³	No	Revised in 2017, original ULM no longer available on website.	¹ Mazzone et al. 2017 ² Pera et al. 2017 ³ Mazzone et al. 2011 Montes et al. 2009
Functional Motor Assessment	North Star Ambulatory Assessment (NSAA)	2+ years	10-15 minutes	Ordinal Scoring of 17 items.	Inter-rater: H ¹ Intra-rater: H ¹ Internal: M-H ²	Not Validated in SMA	No		TREAT-NMD ¹ Mazzone et al. 2009 ² Cano et al. 2013
Functional Motor Assessment	Motor Unit Number Estimation (MUNE)	2+	Unknown	Electrophysiological Signal and computation to determine a Motor Unit Number Estimation	Test-retest: H ¹	Criterion: MHFMS – L (0.63) ¹ MHFMSE – M (0.73) ¹	Unknown	Requires specific equipment. Training is essential to reliable results. ¹ Manual not found.	¹ Lewelt et al. 2010
Functional Motor Assessment	Brooke and Vignos Scales	5+ years	<10 minutes	Scale of 1-6 and 1-10. Qualitative	Presently unavailable for SMA	Presently unavailable for SMA	No		Lue et al. 2009

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Functional Motor Assessment	Timed Stair Climb	5+ years	10 minutes	Timed Score	Presently unavailable for SMA	Presently unavailable for SMA	No		NIH NINDS CDE for SMA
Functional Motor Assessment	10m Walk Test	5+ years	<5 minutes	Time Recorded	Presently unavailable for SMA	Criterion: TUG – L-M (0.691) ¹	No		¹ Dunaway et al. 2014 NIH NINDS CDE for SMA
Functional Motor Assessment	Timed up and Go	5+ years	<15 minutes	Time Recorded	Inter-rater: H ^{1*} Intra-rater: H ^{1*} Internal: Test-Retest: H ¹	Criterion: 10MWT – L-M (0.691) ¹ 6MWT – L (-0.514) ¹ HFMSE – M (-0.717) ¹ MMT – M (0.816) ¹ HHD – L (-0.408/0.658) ¹	No		¹ Dunaway et al. 2014 NIH NINDS CDE for SMA *Not tested in SMA
Functional Motor Assessment	6-minute Walk Test	2+ years	<15 minutes	Distance Covered	Test-Retest: H ^{1,4}	Criterion: TUG – L (0.514) ³ Peak O ₂ Uptake – L (0.558) ¹ HFMSE – M (0.755) ¹ MMT – L (0.691) ¹	No	Able to detect fatigue, whereas other measures such as 10mwt, MVIC are unable to do this. ²	¹ Young et al. 2016 ² Montes et al. 2010 ³ Dunaway et al. 2014 ⁴ Takeuchi et al. 2008

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						HHD – L (0.377/0.622) ¹ 10MWT – H (-0.937) ¹			NIH NINDS CDE for SMA
Functional Motor Assessment	2 Minute Walk Test	2+ years	2 minutes per trial	Distance Covered		Criterion: 6MWT – H (0.99) ^{1*} TUG – H (No		¹ Andersen et al. 2016 *Neuromuscular Diseases, not SMA Specific
Functional Motor Assessment	Jebsen Taylor Hand Function Test	6+ years	15-45 minutes	Timed Sub-scores	Inter-rater: H ^{1*} Intra-rater: M-H ^{1*} Test-Retest: M-H ^{1*}	Content: L ^{1*} Criterion: Michigan Hand Function Test – L-M. ^{1*}	Yes	No Reliability or Validity information available for SMA population.	NIH NINDS CDE for SMA ¹ Shirley Ryan Ability Lab *Population measured was not SMA
Functional Motor Assessment	Box and Blocks Test	6+ years	<10 minutes	Count of Blocks transported in a minute	Presently unavailable for SMA	Presently unavailable for SMA	Yes	Used broadly in other populations. No Reliability or Validity information	SCIRE Project

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								available for SMA population.	
Functional Motor Assessment	Purdue Pegboard	6+ years	<15 minutes	Quantitative Data	Presently unavailable for SMA	Presently unavailable for SMA	No	Evaluation of Fine Coordination & Dexterity. Equipment may need to be purchased.	NIH NINDS CDE for SMA Shirley Ryan Ability Lab
Functional Motor Assessment	Nine Hole Peg Test (9HPT)	4+	Minutes	Time taken	Presently unavailable for SMA	Presently unavailable for SMA	Unknown	Widely studied in other populations.	TREAT-NMD
Functional Motor Assessment	SMA Functional Rating Scale	Adults	30 minutes	Total of 14 items - Quantitative Data	Inter-rater: M-H ¹ Intra-rater: H ¹ Internal: M ¹	Criterion: Limb Norris Score – H (0.853) ¹ ALSFRS Upper Limb – M (0.714) ¹ ALSFRS Trunk – M (0.734) ALSFRS Lower Limb – L (0.693) ¹	No	Evaluator administered.	NIH NINDS CDE for SMA ¹ Hashizume et al. 2015
Functional Motor Assessment	Handheld Dynamometry/Maximum Voluntary Isolation	5+ years	30 Minutes	Data recorded in lbs, N, or Kg	Inter-rater: M-H ^{1,3} Intra-rater: H ¹ Test-Retest: H ¹	Varies depending on joint/muscles measured.		Equipment necessary	¹ Merlini et al. 2002 ² Bertini et al.

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	Contraction Testing							Foot Dorsiflexion has poor reliability ¹ Elbow Flexor is most reliable muscle group. ²	2005 ³ Iannaccone et al. 2003 NIH NINDS CDE for SMA Kissel et al. 2014
Pulmonary Function Test	Forced Vital Capacity	5+ years	~10 minutes	Scored in Litres	Presently unavailable for SMA	Criterion: HFMSE - M-H (0.78-0.92) ¹ EK2 – M (0.77) ²	No		¹ Glanzman et al. 2011 ² Steffensen et al. 2008 Bertini et al. 2005
Pulmonary Function Test	Arterial Blood Gas Testing (ABG)	Any	<20 minutes	% Saturation	Presently unavailable for SMA	Presently unavailable for SMA	No	Reliability & Validity Data unavailable in SMA.	McKim et al. 2011
Pulmonary Function Test	Peak Cough Flow	5+ years	Unknown	Measured in Litres/Minute or Litres/Second	Presently unavailable for SMA	Presently unavailable for SMA	No	Reliability & Validity Data unavailable in SMA.	Bertini et al. 2005 McKim et al. 2011 Suárez et al. 2002
Pulmonary Function Test	Maximum Expiratory Pressure (MEP)	5+ years	As part of PFT	Measured in cmH ₂ O	Presently unavailable for SMA	Presently unavailable for SMA	No	Reliability & Validity Data unavailable in SMA.	McKim et al. 2011

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Pulmonary Function Test	Maximum Inspiratory Pressure (MIP)	5+ years	As part of PFT	Measured in cmH ₂ O	Presently unavailable for SMA	Presently unavailable for SMA	No	Reliability & Validity Data unavailable in SMA.	Bertini et al. 2005 McKim et al. 2011
Pulmonary Function Test	Pressure at 100ms (P0.1)	5+ years	As part of PFT	Measured in cmH ₂ O	Presently unavailable for SMA	Presently unavailable for SMA	No	Reliability & Validity Data unavailable in SMA.	Bertini et al. 2005
Quality of Life	PedsQL / PedsQL Neuromuscular Module	Children 5-18 years Parent/Caregiver 2-18 years	~5 minutes	25 item Questionnaire	Child Inter-rater: H ² Internal: M Test-Retest: H ^{1,2} Parent Inter-rater: H ¹ Test-Retest: H ^{1,2}	Presently unavailable for SMA	Yes		¹ Iannaccone et al. 2009 ² Dunaway et al. 2010
Quality of Life Questionnaire	Short Form 36 (SF-36)	~15 years +	5-20 minutes	Scores of 0-100 assessing varying aspects of QoL.	Inter-rater: M-H ^{1*} Internal: M ^{1*} Intra-rater: M-H ^{1*}	Criterion: IADL – L (0.262) ^{1*}	No	Reliability & Validity Data unavailable in SMA.	¹ SCIRE NIH NINDS CDE for SMA *Not measured in SMA
Quality of Life Questionnaire	Sickness Impact Profile 68	Unknown	30 minutes	Dichotomous Survey	Internal: H ^{1*}	Criterion: Barthel Index – M (0.74) ^{1*} LiSAT – L (-0.52) ^{1*}	Yes	Reliability & Validity Data unavailable in SMA.	¹ SCIRE NIH NINDS CDE *Not measured in SMA

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