

## Spinal Muscular Atrophy (SMA) CDE Highlight Summary

### NIH Resources

The NINDS also strongly encourages researchers to use these NIH developed materials for NINDS-sponsored research, when appropriate. Utilization of these resources will enable greater consistency for NINDS-sponsored research studies. These tools are free of charge.

- National Institutes of Health (NIH) Toolbox
- Quality of Life in Neurological Disorders (Neuro-QOL)
- Patient-Reported Outcomes Measurement Information System (PROMIS)

### Suicidal Ideation

Investigators should review the FDA's ["Guidance for Industry: Suicidal Ideation and Behavior: Prospective Assessment of Occurrence in Clinical Trials"](#) for the most up-to-date information about suicidal ideation and behavior. One scale that FDA suggests is the Columbia Suicide Severity Rating Scale (C-SSRS) (available at [Columbia Suicide Severity Rating Scale Website](#)).

| Disease and Subdomains                                                   | Recommendations and List of Forms                                                                                                 |
|--------------------------------------------------------------------------|-----------------------------------------------------------------------------------------------------------------------------------|
| Spinal Muscular Atrophy (SMA)                                            | Core CDEs:<br>See Start-Up Resource Listing document <a href="#">SMA Start-Up Resource Listing</a>                                |
| Participant / Subject Characteristics; Demographics                      | Core: Demographics<br>Supplemental: None                                                                                          |
| Participant / Subject Characteristics; Social Status                     | Core: Social Status<br>Supplemental: None                                                                                         |
| Participant / Subject History and Family History; General Health History | Core: Family History; Medical History; Prenatal and Perinatal History; Surgical and Hospitalization History<br>Supplemental: None |
| Disease / Injury Related Events; History of Disease / Injury Event       | Core: None<br>Supplemental: Medical History of SMA; Motor Milestones and Current Level of Function                                |
| Assessments and Examinations; Physical / Neurological Examination        | Core: None<br>Supplemental: Physical Examination; Polysomnography                                                                 |
| Assessments and Examinations; Vital Signs and Other Body Measures        | Core: None<br>Supplemental: Vital Signs                                                                                           |

## Spinal Muscular Atrophy (SMA) CDE Highlight Summary

| Disease and Subdomains                                                     | Recommendations and List of Forms                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
|----------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Biomarkers and Sample Data Guidelines; Biospecimen Collection Guidelines for Use; Fat Aspirate Tissue Specimens; Gene Table and Guidelines for Use; Laboratory Tests and Tracking; Laboratory Tests Recommendations Table and Guidelines for Use; Muscle Biopsy and Autopsy Tissue; Mutation Analysis; Nerve Biopsy; NINDS Repository Spinal Muscular Atrophy Data Elements; Skin Biopsies for Qualification of Intraepidermal Nerve Fibers</p> |
| Assessments and Examinations; Imaging Diagnostics                          | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Brain Magnetic Resonance Imaging (MRI); Cardiac Magnetic Resonance Imaging (MRI); Diffusion Tensor Imaging (DTI); Dual-Energy X-Ray Absorptiometry (DEXA); Magnetic Resonance Imaging (MRI); Magnetic Resonance Spectroscopy (MRS)</p>                                                                                                                                                                                                          |
| Assessments and Examinations; Non-Imaging Diagnostics                      | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Echocardiogram; Electrocardiogram (ECG); Electrophysiology; Holter Examination</p>                                                                                                                                                                                                                                                                                                                                                              |
| Treatment/Intervention Data; Devices                                       | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> External Devices</p>                                                                                                                                                                                                                                                                                                                                                                                                                            |
| Treatment/Intervention Data; Therapies                                     | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Gastrointestinal Therapies; Respiratory Interventions</p>                                                                                                                                                                                                                                                                                                                                                                                       |
| Outcomes and Endpoints; Summary of All Outcome Recommendations             | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> SMA Clinical Outcomes Recommendations by Domain and Classification</p>                                                                                                                                                                                                                                                                                                                                                                          |
| Outcomes and Endpoints; Emotional and Cognitive Status                     | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Cognitive Guidelines for Use</p>                                                                                                                                                                                                                                                                                                                                                                                                                |
| Outcomes and Endpoints/End Points                                          | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Death Report</p>                                                                                                                                                                                                                                                                                                                                                                                                                                |
| Outcomes and Endpoints; Functional Status                                  | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> 10 Meter Timed Walk; 6 Minute Walk Test; Grip Strength; North Star Ambulatory Assessment (NSAA); Pinch Strength; Purdue Pegboard; Timed Up and Go (TUG)</p>                                                                                                                                                                                                                                                                                     |

## Spinal Muscular Atrophy (SMA) CDE Highlight Summary

| Disease and Subdomains                                                | Recommendations and List of Forms                                                                                                                                                                                                                                                                          |
|-----------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Outcomes and Endpoints; Muscle Strength Testing                       | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale; Maximum Voluntary Isometric Contraction Scale (MVIC)</p>                                                                                                                   |
| Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Measures of Gas Exchange; Pulmonary Assessment Master Outline and Guidelines for Use; Pulmonary Function Testing</p>                                                                                                                                      |
| Outcomes and Endpoints; Pediatric                                     | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Pediatric Cognitive Instrument Recommendations by Domains and Classification; Pediatric Cognitive Instrument Table; SMA Pediatric Clinical Outcomes Recommendations by Domain and Classification; Pediatric Evaluation of Disability Inventory (PEDI)</p> |
| Outcomes and Endpoints; Quality of Life                               | <p><b>Core:</b> None</p> <p><b>Supplemental:</b> Canadian Occupational Performance Measure (COPM); Nine Hole Peg Test; Pediatric Quality of Life Inventory (PEDIQL); Short Form 36-Item Health Survey (SF-36)</p>                                                                                          |