

Neuromuscular Disease (NMD) 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/Domain	Recommendations
Neuromuscular Disease	<p>These instruments and data elements are recommended for use in all neuromuscular disease studies:</p> <p>Core instruments: Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale</p> <p>Core elements: See Start-Up Resource Listing document (Neuromuscular Disease Start-Up Resource Listing)</p>
Participant/Subject Characteristics/Demographics	<p>Core: Demographics, General Core</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Demographics</p> <p>Exploratory: None</p>
Participant/Subject Characteristics/Social Status	<p>Core: Social Status</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Social Status</p> <p>Exploratory: None</p>
Participant/Subject History and Family History/General Health History	<p>Core: Medical History, Family History</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Family History; Medical History; Prenatal and Perinatal History; Surgical History</p> <p>Exploratory: None</p>

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Disease/Injury Events/History of Disease/Injury Event	<p>Core: Medical History of NMD</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Medical History of NMD</p> <p>Exploratory: None</p>
Assessments and Examinations/Physical/Neurological Examination	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Physical Examination, Polysomnography</p> <p>Exploratory: None</p>
Assessments and Examinations/Vital Signs and Other Body Measures	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Vital Signs</p> <p>Exploratory: Vital Signs</p>
Assessments and Examinations / Laboratory Tests and Biospecimens/ Biomarkers	<p>Core: None</p> <p>Supplemental – Highly Recommended: Fat Aspirate Tissue Specimens, Muscle Biopsy and Autopsy Tissue, Mutation Analysis, Skin Biopsies for Qualification of Intraepidermal Nerve Fibers, Laboratory Tests and Tracking, Peripheral Nerves – Biopsies and Autopsies</p> <p>Supplemental: Biomarkers Sample Data Guidelines, Biospecimen Collection Guidelines for Use, Gene Table and Guidelines for Use, Laboratory Tests Recommendations Table and Guidelines for Use, Muscle Biopsy and Autopsy Tissue, Mutation Analysis, Skin Biopsies for Qualification of Intraepidermal Nerve Fibers, Fat Aspirate Tissue Specimens, Peripheral Nerves – Biopsies and Autopsies</p> <p>Exploratory: None</p>
Assessments and Examinations/Imaging Diagnostics	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Brain Magnetic Resonance Imaging (MRI), Cardiac Magnetic Resonance Imaging (MRI), Diffusion Tensor Imaging (DTI), Dual-Energy X-Ray Absorptiometry (DEXA), Echocardiogram, Magnetic Resonance Imaging (MRI), Magnetic Resonance Spectroscopy (MRS)</p> <p>Exploratory: None</p>

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Assessments and Examinations / Non-Imaging Diagnostics	<p>Core: None</p> <p>Supplemental – Highly Recommended: Electrocardiogram (ECG)</p> <p>Supplemental: Electrocardiogram (ECG), Electrophysiology, Holter Examination</p> <p>Exploratory: None</p>
Treatment / Intervention Data/Therapies	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Gastrointestinal Therapies, Respiratory Interventions</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Summary of All Outcome Recommendations	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: NMD Clinical Outcome Recommendations by Domain and Classification</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Assessing Comorbidities	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Child Health Questionnaire (CHQ)</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Emotional and Cognitive Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Cognitive Guidelines for Use, McGill Pain Questionnaire</p> <p>Exploratory: None</p>
Outcomes and Endpoints/End Points	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Death Report</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Functional Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: 10 Meter Timed Walk, 6 Minute Walk Test, Barthel Index, Purdue Pegboard, Stair Climb, Timed Up and Go (TUG),</p> <p>Exploratory: Jebsen-Taylor Hand Functioning Test</p>

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Outcomes and Endpoints/Muscle Strength Testing	<p>Core: Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Hand Held Dynamometry, Pinch Strength, Maximum Voluntary Isometric Contraction Testing (MVICT), Grip Strength</p> <p>Exploratory: Grip Strength</p>
Outcomes and Endpoints/Other Non-Motor	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Quantitative Sudomotor Axon Reflex Test (QSART)</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Pulmonary Function Testing/Respiratory Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: Pulmonary Function Testing</p> <p>Supplemental: Measures of Gas Exchange, Pulmonary Assessment Master Outline and Guidelines for Use, Pulmonary Function Testing</p> <p>Exploratory: None</p>
Outcomes and Endpoints/Pediatric	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Pediatric Cognitive Instrument Recommendations by Domain and Classification, Bayley Scales of Infant Development (Bayley III, BSID), Beery-Buktenica Developmental Test of Visual-Motor Integration (Beery VMI), California Verbal Learning Test - Children (CVLT-C), Child Behavior Checklist (CBCL), Comprehensive Test of Phonological Processing (CTOPP) Clinical Evaluation of Language Fundamentals (CELF-4), Mullen Scales of Early Learning (MSEL), Peabody Picture Vocabulary Test 4th Edition (PPVT-4), Pediatric Cognitive Instrument Table, Pediatric Evaluation of Disability Inventory (PEDI), Social Communication Questionnaire (SCQ), Social Skills Rating Scale (SSRS), Test for Executive Control (TEC), Wechsler Abbreviated Scale of Intelligence (WASI), Wechsler Intelligence Scale for Children-IV (WISC-IV), Wechsler Preschool and Primary Scale of Intelligence (WPPSI-III), Wide Range Assessment of Memory and Learning, Second Edition (WRAML-2), Woodcock-Johnson III Test of Cognitive Abilities</p> <p>Exploratory: None</p>



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Outcomes and Endpoints/Quality of Life	Core: None Supplemental – Highly Recommended: None Supplemental: Quality of Life in Neurological Disorders (Neuro-QOL), 36-Item Short Form Health Survey (SF-36) Exploratory: None