

Congenital Muscular Dystrophy CDE Highlight Summary Document

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 <http://www.cssrs.columbia.edu>)

Disease/Domain	Recommendations
Congenital Muscular Dystrophy	<p>These instruments and elements are recommended for use in CMD studies:</p> <p>Core elements:</p> <p>See Start-Up Resources Listing document (CMD Start-Up Resource Listing)</p> <p>Supplemental – Highly Recommended instruments: 6 Minute Walk, Bayley Scales of Infant Developmentⁱ, Egen Klassifikation Scale Version 2 (EK2)ⁱⁱ, Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend)ⁱⁱⁱ, North Star Ambulatory Assessment (NSAA)^{iv}, Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale, Wechsler Abbreviated Scale of Intelligence (WASI)^v, Wechsler Intelligence Scale for Children-IV (WISC-IV)^{vi}, Wechsler Preschool and Primary Scale of Intelligence (WPPSI-III), Quality of Life in Neurological Disorders (Neuro-QOL), Patient-Reported Outcomes Measurement Information System (PROMIS)</p>
Participant / Subject Characteristics; Demographics	<p>Core: Demographics^{vii}, 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^{vii}</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Social Status</p> <p>Exploratory: None</p>



Congenital Muscular Dystrophy CDE Highlight Summary Document

Disease/Domain	Recommendations
Participant/Subject History and Family History; General Health History	<p>Core: Surgical History^{vii}</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Family History, Intake Medical History, Interval Medical History, Prenatal and Perinatal History, Surgical History</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^{viii}, Dual-Energy X-Ray Absorptiometry (DEXA), Muscle Imaging</p> <p>Exploratory: Cardiac Magnetic Resonance Imaging, Diffusion Tensor Imaging</p>
Assessments and Examinations; Laboratory Tests and Biospecimens / Biomarkers	<p>Core: Muscle Biopsies and Autopsy Tissue^{vii}</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Muscle Biopsies and Autopsy Tissue, Peripheral Nerves – Biopsies and Autopsies, Skin Biopsies for Qualification of Intraepidermal Nerve Fibers</p> <p>Exploratory: None</p>
Assessments and Examinations; Non-Imaging Diagnostics	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Electrophysiology, Short Scalp Electroencephalography (EEG)</p> <p>Exploratory: Echocardiogram</p>
Assessments and Examinations; Physical/Neurological Examination	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: None</p> <p>Exploratory: Electrical Impedance Myography (EIM)</p>
Treatment / Intervention Data; Drugs	<p>Core: Prior and Concomitant Medications^{vii}</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Prior and Concomitant Medications</p> <p>Exploratory: None</p>

Congenital Muscular Dystrophy CDE Highlight Summary Document

Disease/Domain	Recommendations
Treatment / Intervention Data; Therapies	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Respiratory Interventions</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Functional Status	<p>Core: None</p> <p>Supplemental – Highly Recommended: 6 Minute Walk Test, Bayley Scales of Infant Development^{ix}, Egen Klassifikation Scale Version 2 (EK2), Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend), North Star Ambulatory Assessment (NSAA)</p> <p>Supplemental: Alberta Infant Motor Scale (AIMS), Brooke Upper Extremity Scale, Goniometry, Pediatric Evaluation of Disability Inventory (PEDI^x), Stair Climb, Vignos Lower Extremity Scale</p> <p>Exploratory: 10 Meter Timed Walk, 2 Minute Walk Test, Barthel Index, Gross Motor Function Measure (GMFM-88, GMFM-66), Jebsen Taylor Hand Function Test, Nine Hole Peg Test, Timed Up and Go (TUG)</p>
Outcomes and Endpoints; Muscle Strength Testing	<p>Core: None</p> <p>Supplemental – Highly Recommended: Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale</p> <p>Supplemental: Grip Strength Fatigue, Hand Held Dynamometry, Maximum Voluntary Isometric Contraction Testing (MVICT), Pinch Strength</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Neuropsychological Testing	<p>Core: None</p> <p>Supplemental – Highly Recommended: Wechsler Abbreviated Scale of Intelligence (WASI), Wechsler Intelligence Scale for Children-IV (WISC-IV), Wechsler Preschool and Primary Scale of Intelligence (WPPSI-III)</p> <p>Supplemental: Peabody Picture Vocabulary Test 4th Edition (PPVT-4), Purdue Pegboard, Wechsler Individual Achievement Test-III (WIAT-III)</p> <p>Exploratory: None</p>
Outcomes and Endpoints; Performance Measures	<p>Core: None</p> <p>Supplemental – Highly Recommended: None</p> <p>Supplemental: Motor Function Measure (MFM)</p> <p>Exploratory: None</p>



Congenital Muscular Dystrophy CDE Highlight Summary Document

Disease/Domain	Recommendations
Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status	Core: None Supplemental – Highly Recommended: Pulmonary Function Testing Supplemental: Measures of Gas Exchange Exploratory: None
Outcomes and Endpoints; Quality of Life	Core: None Supplemental – Highly Recommended: Quality of Life in Neurological Disorders (Neuro-QOL), Patient-Reported Outcomes Measurement Information System (PROMIS) Supplemental: None Exploratory: None

ⁱ Supplemental – highly recommended for developmental, psychological, and neuropsychological studies of infants and toddlers up to 42 months old. Highly recommended as a means of characterizing study participants

ⁱⁱ Specifically for studies involving adolescents and adults with CMD

ⁱⁱⁱ Supplemental - highly recommended for studies analyzing motor function -age limit 2+

^{iv} For studies with ambulatory CMD patients

^v Supplemental - highly recommended for psychological and neuropsychological CMD studies for ages 6 years and up; Recommended for other types of CMD studies as a way to characterize the study population

^{vi} Supplemental – highly recommended for psychological and neuropsychological studies for ages 6 to 16 years; Recommended for other types of studies as a way to characterize the study population

^{vii} Contains some Core CDEs

^{viii} The elements on this CRF are Supplemental - highly recommended for dystroglycanopathies; Supplemental for MDC1A; and Exploratory for all other congenital muscular dystrophies

^{ix} Supplemental - highly recommended for developmental, psychological, and neuropsychological studies of infants and toddlers up to 42 months old. Supplemental - highly recommended as a means of characterizing study participants.

^x Particularly appropriate in assessing functional capabilities in CMD children in terms of both current status and change over time