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	**These instruments and elements are recommended for use in CMD studies:** **Core elements:** See Start-Up Resources Listing document (CMD Start-Up Resource Listing) **Supplemental – Highly Recommended instruments:** 6 Minute Walk, Bayley Scales of Infant Development[i], Egen Klassifikation Scale Version 2 (EK 2)[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], Quality of Life in Neurological Disorders (Neuro-QOL), Patient-Reported Outcomes Measurement Information System (PROMIS)
Participant / Subject Characteristics; Demographics	**Core:** Demographics[vii], General Core **Supplemental – Highly Recommended:** None **Supplemental:** Demographics **Exploratory:** None
Participant / Subject Characteristics; Social Status	**Core:** Social Status[viii] **Supplemental – Highly Recommended:** None **Supplemental:** Social Status **Exploratory:** None

NINDS Common Data Elements
Harmonizing information. Streamlining research.

Myotonic Dystrophy CDE Highlight Summary Document

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

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Disease/Domain	Recommendations
Treatment / Intervention Data; Therapies	**Core:** None **Supplemental – Highly Recommended:** None **Supplemental**: Respiratory Interventions **Exploratory:** None
Outcomes and Endpoints; Functional Status	**Core:** None **Supplemental – Highly Recommended:** 6 Minute Walk Test, Bayley Scales of Infant Development[xi], Egen Klassifikation Scale Version 2 (EK 2), Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend), North Star Ambulatory Assessment (NSAA) **Supplemental:**, Alberta Infant Motor Scale (AIMS), Brooke Upper Extremity Scale, Goniometry, Pediatric Evaluation of Disability Inventory (PEDI[xii]), Purdue Pegboard, Stair Climb, **30 Foot Go**, Barthel Index, Stair Climb, Vignos Lower Extremity Scale **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)
Outcomes and Endpoints; Muscle Strength Testing	**Core:** None **Supplemental – Highly Recommended:** Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale **Supplemental:** Grip Strength Fatigue, Hand Held Dynamometry, Maximum Voluntary Isometric Contraction Testing (MVICT), Pinch Strength **Exploratory:** None
Outcomes and Endpoints; Neuropsychological Testing	**Core:** None **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) **Supplemental:** Peabody Picture Vocabulary Test 4th Edition (PPVT-4), Purdue Pegboard, Wechsler Individual Achievement Test-III (WIAT-III) **Exploratory:** None
Outcomes and Endpoints; Performance Measures	**Core:** None **Supplemental – Highly Recommended:** None **Supplemental:** Motor Function Measure (MFM) **Exploratory:** None

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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] Highly recommended for studies analyzing motor function -age limit 2+
[iv] for studies with ambulatory CMD patients
[v] 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] 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] Contains some Core CDEs
[ix] Contains some Core CDEs
[x] Contains some Core CDEs

[xii] Particularly appropriate in assessing functional capabilities in CMD children in terms of both current status and change over time