Summary of Core and Supplemental – Highly Recommended Recommendations: Congenital Muscular Dystrophy CDEs

Start-up Resource – NINDS Congenital Muscular Dystrophy Disease CDE Recommendations

The National Institute of Neurological Disorders and Stroke (NINDS) and other Federal agencies and international organizations have the common mission of developing data standards for clinical research. Through the efforts of subject-specific working groups, topic-driven data elements have been created. The first set of Common Data Elements (CDEs) for Congenital Muscular Dystrophy was developed in 2014. The Core data elements to be used by an investigator when beginning a research study in this disease/disorder are listed in this resource document. All other recommendations are listed on the website and should be considered based on study type.

Each CDE or instrument could be classified according to the definitions below:

General Core: A data element that is required for all NINDS funded studies.

Disease Core: A data element that collects essential information applicable to any disease-specific study, including all therapeutic areas. The NINDS and its appointed working groups assign the disease "Core" classification based on the current clinical research best practices. In each case, the disease Core CDEs are a small subset of the available CDEs, where it is anticipated that investigators will need to collect the disease Core CDEs on any type of study. These are required for all disease-specific studies.

Disease Supplemental - Highly Recommended: A data element which is essential based on certain conditions or study types in clinical research studies. In most cases, these have been used and validated in the disease area. These data elements are strongly recommended for the specified disease condition, study type or design.

Disease Supplemental: A data element which is commonly collected in clinical research studies. Use depends upon the study design, protocol or type of research involved. These are recommended, but not required, for studies.

Disease Exploratory: A data element that requires further validation but may fill current gaps in the CDEs and/or substitute for an existing CDE once validation is complete. Such data elements show great promise but require further validation before they are ready for prime-time use in clinical research studies. They are reasonable to use with the understanding that it has limited validation in the target group.

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

Core CDEs for all NINDS Studies[1]:

Domain/Sub-Domain	CDE Name	CDE ID	Study Type
Participant Characteristics; Demographics	Birth date	C00007	All studies
Participant Characteristics; Demographics	Ethnicity USA category	C00020	All studies
Participant Characteristics; Demographics	Race USA category	C00030	All studies
Participant/Subject Characteristics; Demographics	Birth sex assigned type	C58676	All studies
Participant Characteristics; Demographics	Gender identity type	C58677	All studies
Participant History and Family History; General Health History	Medical history condition text	C00322	All studies
Participant History and Family History; General Health History	Medical history condition SNOMED CT code	C00313	All studies

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[1] Note: Education year count C00015 is no longer a general Core CDE

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General Core for all Studies:

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).

Core CDEs for Congenital Muscular Dystrophy:

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Surgery lifetime total count	C12671
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Specimen source type	C12226
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen size measurement	C12229
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen collection date and time	C12230
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Preservation technique used type	C12233
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Histochemical stains used type	C12235

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Histochemical stains used other text	C18828
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy histochemical stains diagnostic abnormalities present type	C12249
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy fiber abnormality status	C19939
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy type 1 predominance fiber percentage value	C12526
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy fiber abnormality type	C12525
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biopsy and autopsy type 2 predominance fiber percentage value	C12527
Treatment/Intervention Data; Drugs	Medication prior or concomitant use indicator	C02002

Supplemental-Highly Recommended CDEs for Congenital Muscular Dystrophy:

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Biospecimen fragment collect count	C12277
Assessments and Examinations; Imaging Diagnostics	Imaging magnetic resonance imaging performed indicator	C17931
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function complete exhalation indicator	C12310
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity early termination indicator	C12317
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity large value range indicator	C12316
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity leak indicator	C12315
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity peak flow indicator	C12318
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function best trial measurement	C12312

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function index of lung function type	C12311
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function maximal pressure trial difference indicator	C12313
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test date and time	C11098
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment manufacturer name	C12303
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment model name	C12304
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test equipment software program name	C12305
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test mouth apparatus type	C11101
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done reason	C12298

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test not done other text	C18833
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test position type	C11100
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function test seat position type	C12301
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Specimen histochemical stain diagnostic abnormality type	C12278
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen collection anatomic site	C12285
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen collection anatomic site other text	C18830
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen section count	C12287
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue specimen section thickness measurement	C12286
Outcomes and End Points; Muscle Strength Testing	Trial number	C10171

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Domain; Sub-Domain	Data Element	CDE ID
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Ulna length measure tool name	C12307
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Ulna length measure tool other text	C18834

Supplemental – Highly Recommended Instruments for Congenital Muscular Dystrophy:

1. 6 Minute Walk

2. Bayley Scales of Infant Development*

3. Egen Klassifikation Scale Version 2 (EK2)*

4. Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend)*

5. North Star Ambulatory Assessment (NSAA)*

6. Manual Muscle Testing-Using the Medical Research Council Muscle Grading Scale

7. Wechsler Abbreviated Scale of Intelligence (WASI)*

8. Wechsler Intelligence Scale for Children-IV (WISC-IV)*

9. Wechsler Preschool and Primary Scale of Intelligence (WPPSI-III)

10. Quality of Life in Neurological Disorders (Neuro-QoL)*

11. Patient-Reported Outcomes Measurement Information System (PROMIS)

* Special circumstances apply

For the complete list of NINDS CDE recommendations for Congenital Muscular Dystrophy, please see the NINDS CDE website.