



Summary of Core/Supplemental-Highly Recommended Mitochondrial Disease CDEs

Start-up Resource – NINDS Mitochondrial 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 Mitochondrial Disease 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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<p>National Institute of Health (NIH) Resources: <i>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.</i></p>	<ul style="list-style-type: none"> • NIH Toolbox • Quality of Life in Neurological Disorders (Neuro-QOL) • Patient-Reported Outcomes Measurement Information System (PROMIS)
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Core CDEs for all NINDS Studies¹:

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 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

Supplemental-Highly Recommended CDEs for Mitochondrial Disease:

Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	2-hour glucose (OGTT)	C19725

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

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Anti-pancreatic autoantibodies	C19723
Assessments and Examinations; Vital Signs and Other Body Measures	Blood pressure measurement	C19565
Assessments and Examinations; Vital Signs and Other Body Measures	Blood pressure measurement position type	C01553
Assessments and Examinations; Vital Signs and Other Body Measures	Body mass index value	C11131
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Clinical Category	C19602
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Diagnostic Lab Name	C19600
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Fasting glucose	C19724
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Forced expiratory volume second result	C10173
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Forced vital capacity result	C10172
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Gene	C12690

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test karyotype tissue type	C19595
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test microarray tissue type	C19594
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test mitochondrial DNA content analysis tissue type	C19593
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test mitochondrial DNA genome deletion duplication analysis tissue type	C19592
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test mitochondrial DNA panel tissue type	C19590
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test methylation analysis tissue type	C19596
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test single gene deletion analysis tissue type	C19587
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test single gene sequence analysis tissue type	C19586
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test Trinucleotide repeat analysis tissue	C19597

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test whole exome sequence analysis tissue type	C19588
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test whole genome sequence analysis tissue type	C19589
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Genetic test whole mitochondrial DNA genome sequence analysis tissue type	C19591
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Glycosylated Hemoglobin (Hemoglobin A1c)	C19722
Assessments and Examinations; Vital Signs and Other Body Measures	Heart rate	C01521
Assessments and Examinations; Vital Signs and Other Body Measures	Height measurement	C01522
Assessments and Examinations; Vital Signs and Other Body Measures	Height unit of measure	C19573
Assessments and Examinations; Laboratory Tests and Biospecimens	Other	C19598
Assessments and Examinations; Laboratory Tests and Biospecimens	PR interval	C04509
Outcomes and End Points; Pulmonary Function Testing/Respiratory Status	Pulmonary function forced vital capacity (FVC) flow volume crossed x-axis indicator	C11109
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	ECG QRS duration	C04510

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	ECG QT interval	C04511
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Region Tested: Coverage	C19609
Assessments and Examinations; Vital Signs and Other Body Measures	Respiratory rate	C01535
Assessments and Examinations; Vital Signs and Other Body Measures	Length measurement infant	C19575
Assessments and Examinations; Vital Signs and Other Body Measures	Length unit of measure infants	C19577
Assessments and Examinations; Vital Signs and Other Body Measures	Length percentile value	C19608
Assessments and Examinations; Vital Signs and Other Body Measures	Length Z-Score	C19613
Assessments and Examinations; Vital Signs and Other Body Measures	Length Standard Used Infant	C19616
Assessments and Examinations; Vital Signs and Other Body Measures	Height measurement	C19572
Assessments and Examinations; Vital Signs and Other Body Measures	Height unit of measure pediatric	C19633
Assessments and Examinations; Vital Signs and Other Body Measures	Height percentile value	C15901
Assessments and Examinations; Vital Signs and Other Body Measures	Height Z-Score	C19617
Assessments and Examinations; Vital Signs and Other Body Measures	Height Standard Used Infant	C19618

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Domain; Sub-Domain	Data Element	CDE ID
Assessments and Examinations; Vital Signs and Other Body Measures	Temperature measurement	C01539
Assessments and Examinations; Vital Signs and Other Body Measures	Temperature measurement anatomic site	C19570
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Test Methodology	C19585
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Test Name	C19615
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	Tissue	C19601
Assessments and Examinations; Vital Signs and Other Body Measures	Vital signs date and time	C01519
Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers	What year was genetic testing performed?	C19584
Assessments and Examinations; Vital Signs and Other Body Measures	Weight measurement	C01541
Assessments and Examinations; Vital Signs and Other Body Measures	Weight measurement pediatric	C19631
Assessments and Examinations; Vital Signs and Other Body Measures	Weight pediatric unit of measure	C19632
Assessments and Examinations; Vital Signs and Other Body Measures	Weight percentile value	C15900
Assessments and Examinations; Vital Signs and Other Body Measures	Weight unit of measure	C01581



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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](#)).



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Supplemental Highly-Recommended Instruments for Mitochondrial Disease:

1. Apathy Evaluation Scale
2. Apathy Scale
3. Automated Self-Administered 24-hour Dietary Recall (ASA 24)
4. Barry Albright Dystonia Scale (BADs)
5. Behavior Rating Inventory of Executive Function (BRIEF)
6. Behavior Rating Inventory of Executive Function-Preschool Version (BRIEF-P)
7. Conners Continuous Performance Test, 3rd Edition
8. Modified Hammersmith Functional Motor Scale (MHFMS-SMA, MHFMS)
9. Newcastle Pediatric Mitochondrial Disease Scale (NPDMS)
10. Peabody Development Motor Scale II
11. Pediatric Quality of Life Inventory (PEDSQL)
12. Scale for the Assessment and Rating of Ataxia (SARA)
13. Test of Variable Attention (TOVA)
14. The Borg Scale of Perceived Exertion
15. Vineland Adaptive Behavior Scales, 2nd Ed.
16. World Health Organization Quality of Life Assessment (WHOQOL)

For the complete list of NINDS CDE recommendations for Mito, please see the [NINDS CDE website](#).