

MITOCHONDRIAL DISORDERS (DISEASES) COMMON DATA ELEMENTS SUMMARY

Developing standard common data elements (CDEs) for Primary Mitochondrial disorders is challenging because these disorders are systemic, protean and are a class of disorders, rather than a single disease. Diagnosis is often based on laboratory tests, including genetic tests, and most disorders do not have predictable genotype-phenotype correlations. Perhaps for these reasons, developing CDEs for research, including clinical trials, is especially valuable. Unlike many of the other CDEs developed for this project, the Mitochondrial Disorder CDEs are not limited to the neurological or neuromuscular realms of clinical medicine, but span almost all organ systems.

To develop the [Mitochondrial Disease CDEs](#), the [Mitochondrial Disease CDE Working Groups](#) are divided into subgroups to focus on identifying and defining data elements in the domains of: biomarkers; cognitive/behavioral/psychological outcomes; endocrinology/diabetes/GI/nutrition; exercise physiology; genetics; imaging; neurological assessments; patient reported outcomes/quality of life; and vision.

Core elements for the mitochondrial disorders include those core elements for all disorders: see [General Core](#). There are no additional core elements specifically for Mitochondrial disorders. However, an extensive set of assessments exist in the categories Supplemental-Highly Recommended, Supplemental and Exploratory. [Supplemental-Highly Recommended](#) CDEs for Mitochondrial disorders categories include:

- Participant/Subject History and Family History; General health history: the Diabetes-Related Medical History.
- Assessments and Other Examinations; Vital Signs and Other Body Measures: Anthropometrics – Vital Signs
- Assessments and Examinations; Physical Examinations: the Automated Self-Administered 24-hour Dietary Recall (ASA 24), Maximal Exercise Test, Sub-Maximal Exercise Test
- Assessments and Examinations; Laboratory Tests and Biospecimens/Biomarkers: Laboratory Tests and Non-Imaging Diagnostics (Diabetes).
- Assessments and Examinations; Imaging Diagnostics: Echocardiogram
- Assessments and Examinations; Non-Imaging Diagnostics: Electrocardiogram
- Outcomes and Endpoints; Adaptive: Vineland Adaptive Behavior Scales, 2nd Ed
- Outcomes and Endpoints; Attention: Conners III, Test of Variable of Attention (TOVA)
- Outcomes and Endpoints; Ataxia and Performance Measures: Scale for the Assessment and Rating of Ataxia
- Outcomes and Endpoints; Emotional/Behavioral: Apathy Evaluation Scale

- Outcomes and Endpoints; Executive Functioning: Behavior Rating Inventory of Executive Function (BRIEF), Behavior Rating Inventory of Executive Function –Preschool Version (BRIEF-P)
- Outcomes and Endpoints; Motor/Physical Function: Borg Rating of Perceived Exertion (RPE) Scale, Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend), Newcastle Pediatric Mitochondrial Disease Scale (NPDMS), Peabody Developmental Motor Scale II (PDMS-2), Barry Albright Dystonia Scale (BADs)
- Outcomes and Endpoints; Pulmonary Function Testing/Respiratory Status: Pulmonary Function
- Outcomes and Endpoints; Quality of Life: Pediatric Quality of Life (PEDS QL) for pediatric QOL, World Health Organization Quality of Life Assessment (WHOQOL-BREF)

All of the above assessment tools can be downloaded via the [Mitochondrial Disease NINDS Common Data Elements Website](#): see “Download Mitochondrial Disease CDE Recommendations” or scroll down for the full list of elements.