

Mitochondrial Disease Version 2.0 NINDS CDE Project Gastrointestinal (GI)/Hepatology and Nutrition Subgroup Summary

The NINDS Mitochondrial Disease v2.0 Common Data Element (CDE) Gastrointestinal (GI)/Hepatology and Nutrition Subgroup reviewed and updated CDEs based on advancements in neuroscientific clinical research. The subgroup focused on the examination of gastrointestinal, liver, pancreatic, colonic, nutritional, and feeding and swallowing aspects of mitochondrial diseases. Gastrointestinal involvement in mitochondrial diseases can differ based on genotype and phenotype including gastrointestinal motor and sensory functions. GI/Hepatology and Nutrition assessments are used in mitochondrial disease research, natural history studies, observational studies, clinical trials, and prevention studies.

The subgroup reassessed the CRFs and validated instrument from version 1.0. The subgroup expanded their review to include additional content related to the liver and pancreas, nutrition and feeding.

Instruments that can be used across the life span were considered, since mitochondrial disease impacts both pediatric and adult populations. There are a broad range of clinical phenotypes as well as heterogeneity within the phenotypes.

Overlap addressed with other subgroups includes anthropometric measurements. This subgroup reviewed anthropometric measurements along with the Endocrinology and General Medical/Neurological Assessments Subgroups. Assessment tools were evaluated, and include question/answer, pencil/paper, and/or computerized methods.

Summary of Recommendations

Subdomain	Instrument/CRF Name	Classification
General Health History	Dietary Supplements	Supplemental
Physical Examinations	Automated Self-Administered 24-hour Dietary Recall (ASA24)	Supplemental – Highly Recommended
	Mitochondrial and Gastrointestinal Diseases Assessment	Supplemental
Quality of Life/ Patient Reported Outcomes	Pediatric Quality of Life Inventory (PedsQL)	Supplemental – Highly Recommended