

## **Mitochondrial Disease Version 2.0 NINDS CDE Project General Medical/Neurological Assessments Subgroup Summary**

The NINDS Mitochondrial Disease v2.0 Common Data Element (CDE) General Medical/Neurological Assessments Subgroup reviewed and updated CDEs based on advancements in neuroscientific clinical research. The General Medical/Neurological Assessments Subgroup focused on data collection points related to general medical and neurological assessments, including study endpoints and outcomes within those areas.

The subgroup reassessed the following recommended case report form (CRF) and instruments:

- Vital Signs CRF
- Barry Albright Dystonia Scale (BADs)
- Bayley Scales of Infant Development (Bayley III, BSID)
- Burke-Fahn-Marsden Movement Scale (BFMMS)
- Gross Motor Function Measure (GMFM-88, GMFM-66)
- International Cooperative Ataxia Rating Scale (ICARS)
- International Pediatric Mitochondrial Disease Score (IPMDS)
- Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend)
- Motor Function Measure (MFM)
- Newcastle Mitochondrial Disease Adult Scale (NMDAS)
- Newcastle Paediatric Mitochondrial Disease Scale (NPMDS)
- Peabody Developmental Motor Scale Second Edition (PDMS-2)
- Scale for the Assessment and Rating of Ataxia (SARA)
- Unified Dystonia Rating Scale (UDRS)

The subgroup expanded their review to include the following CRFs and instruments:

- Family History CRF
- Medical History CRF
- Physical Exam CRF
- 12 Minute Walking Test
- Mitochondrial Myopathy Composite Assessment Tool (MM-COAST)
- Primary Mitochondrial Myopathy Symptom Assessment (PMMSA)

Overlap addressed with other subgroups includes the following CRF and instruments:

- Mitochondrial Myopathy Composite Assessment Tool (MM-COAST)
- Newcastle Mitochondrial Disease Adult Scale (NMDAS)
- Physical Exam: Anthropometric measurements and Tanner Stage

## Summary of Recommendations

Subdomain	Instrument/CRF Name	Classification
Ataxia and Performance Measures	International Cooperative Ataxia Rating Scale (ICARS)	Supplemental
	Scale for the Assessment and Rating of Ataxia (SARA)	Supplemental – Highly Recommended
General Health History	Family History	Core; Supplemental
	Medical History	Core; Supplemental
Motor Function	12 Minute Walking Test	Supplemental
	Barry Albright Dystonia Scale (BADs)	Supplemental – Highly Recommended
	Burke-Fahn-Marsden Movement Scale (BFMMS)	Supplemental
	Gross Motor Function Measure (GMFM-88, GMFM-66)	Supplemental
	International Pediatric Mitochondrial Disease Score (IPMDS)	Supplemental
	Mitochondrial Myopathy Composite Assessment Tool (MM-COAST)	Supplemental – Highly Recommended
	Modified Hammersmith Functional Motor Scale for Children with Spinal Muscular Atrophy (MHFMS-SMA/MHFMS-Extend)	Supplemental - Highly Recommended
	Motor Function Measure (MFM)	Supplemental – Highly Recommended
	Newcastle Mitochondrial Disease Adult Scale (NMDAS)	Supplemental – Highly Recommended
	Newcastle Paediatric Mitochondrial Disease Scale (NPMDS)	Supplemental – Highly Recommended
	Peabody Developmental Motor Scale Second Edition (PDMS-2)	Supplemental
	Primary Mitochondrial Myopathy Symptom Assessment (PMMSA)	Supplemental
	Unified Dystonia Rating Scale (UDRS)	Supplemental
Neuropsychological Testing	Bayley Scales of Infant Development (Bayley III, BSID)	Supplemental
Physical Examinations	Physical Exam	Supplemental – Highly Recommended
Vital Signs and Other Body Measures	Vital Signs	Supplemental