

## Mitochondrial Disease Version 2.0 NINDS CDE Project Biomarkers Subgroup Summary

Biomarkers are accessible, measurable variables found in blood, other body fluids (e.g., urine and CSF), and tissues with potential utility to confirm or evaluate mitochondrial diseases through testing in a biochemical or histology laboratory or by magnetic resonance spectroscopy. Clinical assessment variables are also biomarkers (e.g., height, weight, growth, head circumference, indirect calorimetry, O<sub>2</sub>, 6 minute walk test, etc.). Biomarkers serve more than one purpose which include to:

1. Substantiate or provide additional evidence of a mitochondrial disease diagnosis
2. Indicate severity of disease
3. Determine standard of care
4. Monitor medication/therapy

### Summary of Recommendations

Subdomain	CRF/Guidance Document Name	Classification
Laboratory Tests and Biospecimens/Biomarkers	Biomarkers in Mitochondrial Disease	Supplemental
Laboratory Tests and Biospecimens/Biomarkers	Biomarkers Guidance Mitochondrial Disease	N/A