

## Overview

### **Mitochondrial Disease Working Group: Patient Reported Outcome/Quality of Life**

The development of the Mitochondrial Disease (Mito) Patient Reported Outcome/Quality of Life (PRO/QOL) Common Data Elements (CDEs) was a multi-tiered process which required compilation, review, and re-review of pre-existing validated instruments by means of monthly meetings.

The National Institute of Neurological Disease and Stroke (NINDS) PRO/QOL Working Group (WG) members along with the NINDS CDE Team compiled a list of mitochondrial disease PRO/QOL scales to be reviewed for content and usability for recommendations from the WG. Individual committee members were assigned instruments for review (minimum of 2 reviewers per instrument). Each assigned reviewer independently reviewed the instrument and completed a questionnaire detailing the following: short description of scale, population (age and criteria), diagnoses/diseases where scale has been applied and validated, validation criteria, how the scale is scored, notable studies which have used the scale, limitations for use in mitochondrial disease patients, advantages for use in mitochondrial disease patients, time required to complete the scale, fee associated or copyright limitations, and general recommendations. Each committee member submitted their independent review to the NINDS CDE Team for distribution and collation of findings. Conference calls were then conducted where primary and secondary reviewers for each scale presented their findings and recommendations. Following this presentation, the group decided if the instrument should (1) definitely be included, (2) definitely not be included, or (3) retained for further review. Once the final list of scales was determined, each committee member was asked to submit a priority list. A conference call was held to review and finalize the priority list and final additions and eliminations were made to the instrument recommendations list. The WG then classified the instruments as Supplemental-Highly Recommended, Supplemental, or Exploratory. The WG found no QOL CDEs to be essential and thus no instruments were classified as Core.

The instruments recommended by the WG had no differential applications to types of mitochondrial disease. However, the scales are often dependent on age for administration, and some are better suited for subjects with higher cognitive skills. (To see a summary of the WG's recommendations, please go to their Instrument Recommendations Documents, which are segregated by Adult and Pediatric instruments.)

Validated QOL CDEs are lacking. These CDEs are often dependent on age and on the overall health/cognitive status of a given subject. Due to these limitations, it was difficult to define universal CDEs for quality of life within mitochondrial diseases.

## Summary Recommendations

READ ME: This is a recommendations summary document of the instruments/measures for ADULT recommendations. Details of the recommendations follow this spreadsheet in the form of information documents (e.g., Notices of Copyright).

**Table 1 Patient Reported Outcome Quality of Life Working Group Summary**

Instrument / Scale Name <i>Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs</i>	Domain	CDE Subdomain (e.g. Mitochondrial Diseases Working Group)	Classification (e.g., Core, Supplemental–Highly Recommended, Supplemental, Exploratory)
World Health Organization Quality of Life Assessment (WHOQOL)	End Points and Outcomes	Patient Reported Outcome/ Quality of Life	Supplemental–Highly Recommended
Craig Handicap and Assessment Reporting Technique (CHART)	End Points and Outcomes	Patient Reported Outcome/ Quality of Life	Supplemental
EuroQoL-5 Dimension Questionnaire (EQ-5D)	End Points and Outcomes	Patient Reported Outcome/ Quality of Life	Exploratory
Pediatric Quality of Life Inventory (PEDS QL)	Outcomes and End Points	Patient Reported Outcome/ Quality of Life	Supplemental–Highly Recommended for Pediatric QOL
Newcastle Pediatric Mitochondrial Disease Scale (NPMDS)	Outcomes and End Points	Patient Reported Outcome/ Quality of Life	Supplemental
EuroQoL-5 Dimension Questionnaire (EQ-5D)	Outcomes and End Points	Patient Reported Outcome/ Quality of Life	Exploratory

Family Strain Questionnaire Assessment of Preschooled Children's Participation (APCP)	Outcomes and End Points	Patient Reported Outcome/ Quality of Life	Exploratory
Pediatric Quality of Life Multidimensional Fatigue Scale	Outcomes and End Points	Patient Reported Outcome/ Quality of Life	Exploratory