

NINDS CDE Project

Working group: Myotonic Dystrophy (DM) –Strength and Function measures

The NINDS Myotonic Dystrophy (DM) Common Data Elements (CDE) project was comprised of a Working Group (WG) of researchers from the DM community. The group completed a literature search of ALL articles related to measures of muscle strength and function used in Natural History studies and Therapeutic Trials in PubMed on DM from 1980-2013. It reviewed the findings and recommendations. The research is sparse and conflicting, since there are no specific measures with documented superiority based on reliability or sensitivity as reported in the literature. Hence recommendations were developed using the literature, expert opinion and consensus among members of the WG.

There are two types of DM, DM-1 and DM-2, and the measures the group has recommended are applicable to both. In addition, there is an ongoing effort by an expert group of clinicians to arrive at CDEs for use in clinical care and research, called the OMMYD, or the Outcome Measures in Myotonic Dystrophy. This group has had two meetings and a third meeting is planned for June 8, 2015. It will be a satellite workshop prior to the International Myotonic Dystrophy Consortium meeting in Paris, France. Consensus has not yet been reached by the OMMYD on ALL data elements.

As noted earlier, there is a paucity of large scale longitudinal studies in DM. As a result the sensitivity, reproducibility, and validity at different severities of muscle weakness and function of many of the recommended measures have not been well established. The WG is aware that a 3 year longitudinal study has been completed in a population of ambulatory (able to walk for 6 minutes) DM1 patients and the results of this study will be available in the near future. Based on the findings from that study, the WG may be able to provide more evidence-based recommendations for ambulatory DM1 patients.

Summary recommendations

READ ME: This is a recommendations summary document of the instruments/measures/case report forms, sorted alphabetically. Details of the recommendations follow this spreadsheet in the form of information documents (e.g., Notices of Copyright) or case report forms and CDE details.

Table 1 Summary of Recommended Instruments for Myotonic Dystrophy

Instrument / Scale Name / CRF template Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs	Domain	Sub-domain	Classification [e.g., Core, Supplemental (also those highly-recommended), or Exploratory]
10 meter walk test	Outcomes and End Points	Functional Status	Supplemental
2 minute walk test	Outcomes and End Points	Functional Status	Supplemental

Instrument / Scale Name / CRF template Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs	Domain	Sub-domain	Classification [e.g., Core, Supplemental (also those highly-recommended), or Exploratory]
6 min walk test	Outcomes and End Points	Functional Status	Supplemental
9 - Hole Peg Test	Outcomes and End Points	Functional Status	Supplemental
Barthel Index	Outcomes and End Points	Functional Status	Supplemental
Bedside Myotonia	Assessments/Examinations	Physical/Neurological Examination	Supplemental - Highly Recommended for QOL, functional status, therapy
Epworth sleepiness scale	Assessments/Examinations	Physical/Neurological Examination	Supplemental - Highly Recommended for QOL, functional status, therapy
Evoked Myotonia through Electrical Stimulation	Assessments/Examinations	Physical/Neurological Examination	Exploratory
Gross Motor Function Measure (GMFM)	Outcomes and End Points	Functional Status	Supplemental
Individualized Neuromuscular Quality of Life Questionnaire (INQoL)	Assessments/Examinations	Quality of Life	Supplemental
Jebsen Taylor Hand Function Test	Outcomes and End Points	Functional Status	Supplemental

Instrument / Scale Name / CRF template Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs	Domain	Sub-domain	Classification [e.g., Core, Supplemental (also those highly-recommended), or Exploratory]
Manual Muscle Testing	Outcomes and End Points	Muscle Strength Testing	Supplemental - Highly recommended
Maximum Isometric Voluntary Contraction Testing (MVICT)	Outcomes and End Points	Muscle Strength Testing	Supplemental
McGill Pain Questionnaire (MPQ) short form	Outcomes and End Points	Pain	Supplemental
Myotonic Dystrophy Health Index (MDHI)	Assessments/Examinations and Outcomes and End Points	Multifactorial Patient Reported Burden of Disease	Supplemental - Highly recommended
Neuro-QOL	Outcomes and End Points	Quality of Life	Exploratory
Pediatric Quality of Life (PedsQL)	Outcomes and End Points	Pediatric	Exploratory
PedsQL - Neuromuscular	Outcomes and End Points	Pediatric	Exploratory
PROMIS	Outcomes and End Points	Quality of Life	Exploratory
Purdue Pegboard	Outcomes and End Points	Functional Status	Supplemental
Short Form Health Survey-36 (SF-36)	Outcomes and End Points	Quality of Life	Supplemental
<u>Sickness Impact Profile (SIP)</u>	Outcomes and End Points	Quality of life	Exploratory

Instrument / Scale Name / CRF template Name and acronym of the instrument/measure that is recommended for inclusion in the CDEs	Domain	Sub-domain	Classification [e.g., Core, Supplemental (also those highly-recommended), or Exploratory]
Stair climb (Time to climb 4 stairs)	Outcomes and End Points	Functional Status	Supplemental
Timed up and go	Outcomes and End Points	Functional Status	Supplemental
Wechsler Abbreviated Scale of Intelligence (WASI)	Outcomes and End Points	Performance Measures	Supplemental

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Table 2 Summary of Case Report Forms (CRF) for Myotonic Dystrophy

CRF Name	Domain	Subdomain	Classification (e.g., Core, Supplemental (also those highly-recommended), Exploratory)
Magnetic Resonance Imaging (MRI)	Assessments and Examinations	Imaging Diagnostics	Supplemental
Cardiac Magnetic Resonance Imaging	Assessments and Examinations	Imaging Diagnostics	Supplemental
Echocardiogram	Assessments and Examinations	Non-Imaging Diagnostics	Supplemental

CRF Name	Domain	Subdomain	Classification (e.g., Core, Supplemental (also those highly-recommended), Exploratory)
Electrocardiogram (ECG)	Assessments and Examinations	Non-Imaging Diagnostics	Supplemental; contains Supplemental - Highly Recommended CDEs
Electrophysiology	Assessments and Examinations	Non-Imaging Diagnostics	Supplemental
Hand Held Dynamometry	Outcomes and Endpoints	Muscle Strength Testing	Supplemental
Laboratory Tests and Tracking	Assessments and Examinations	Laboratory Tests and Biospecimens/Biomarkers	Supplemental
Measures of Gas Exchange	Outcomes and Endpoints	Pulmonary Function/Respiratory Status	Supplemental
Muscle Biopsy and Autopsy Tissue	Assessments and Examinations	Laboratory Tests and Biospecimens/Biomarkers	Supplemental
Muscle Strength Testing	Outcomes and Endpoints	Muscle Strength Testing	Supplemental
Mutation Analysis	Assessments and Examinations	Laboratory Tests and Biospecimens/Biomarkers	Supplemental, but contains some Core CDEs

CRF Name	Domain	Subdomain	Classification (e.g., Core, Supplemental (also those highly- recommended), Exploratory)
Nine Hole Peg Test	Outcomes and Endpoints	Functional Status	Supplemental
Pulmonary Function Testing	Outcomes and Endpoints	Pulmonary Function/Respiratory Status	Supplemental - Highly Recommended
Respiratory Interventions	Treatment/Intervention Data	Therapies	Supplemental