Quality improvement in neurology: Muscular dystrophy quality measures

SPECIAL ARTICLE

Quality improvement in neurology

Muscular dystrophy quality measures

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The muscular dystrophies (MDs) are a heterogeneous group of genetically determined myopathies. Identification of underlying genetic defects has demonstrated that MDs exhibit significant phenotypic and genetic heterogeneity. One genetic mutation can lead to a variety of phenotypes while different genetic mutations can manifest similar phenotypes; therefore MDs are challenging to diagnose.

A major goal of health care reform in the United States is to replace the traditional fee-for-service model with a value-based system, which incentivizes high-quality care. Quality measurement is an integral and necessary part of this process.[1,2] While standardizing care of MDs can be challenging because of their heterogeneity, common themes of management, such as the maintenance of nutrition, sustaining mobility, and management of complications, are applicable to many MDs. We report a quality measurement set for the management of MDs.

BACKGROUND

Prevalence of MDs. MDs are rare disorders. The most common form, Duchenne MD (DMD), affects 1/3,500–6,000 male births yearly in the United States, representing approximately 50% of all cases.[3–6] Myotonic dystrophy (DM), the most common adult-onset MD, has an estimated prevalence of 11/100,000.[7] Facioscapulohumeral dystrophy (FSHD) is the third most common form, with a prevalence of 4–6/100,000.[8,9]

Challenges in the diagnosis and management of MDs. MDs often present with nonspecific symptoms such as muscle weakness, which are features of many other diseases. Accurate diagnosis is a prerequisite for appropriate management and cost-effective use of medical resources. Knowledge of the specific type of MD is necessary to define long-term prognosis, and promotes efficient care (e.g., timely monitoring for MDs associated with cardiopulmonary complications and, conversely, avoiding unnecessary testing for MDs infrequently associated with these complications). Disease severity, rate of progression, medical complications, and life expectancy vary significantly with the type of MD.[10–12]

Impact of MD on health-related quality of life. Quality of life (QoL) studies in MD are sparse. MD may negatively influence health-related QoL (HRQoL) in physical and psychosocial domains.[13,14] HRQoL in DMD was not affected by the need for noninvasive assisted ventilation in one study, suggesting that patients' perceptions of QoL are important to consider when therapeutic decisions are made.[15]

Disparities in MD care and costs of care of MD. MD occurs worldwide and affects all races. There are scant data regarding disparities in the care of patients with MD. In one study,[16] age-adjusted mortality rate was higher for white patients, median age at death was lower for black patients, and cardiac complications were more common among MD-associated deaths in black patients. An increase was noted over time in age at death for white male patients, suggesting possible inequities in access to health care.[16]

Costs of MD include direct health system expenditure, nonmedical costs (home modifications, transportation, professional care, travel), and indirect expenditure (loss of productivity, absenteeism, informal caregiving). These have been estimated to be approximately $126,000/person/year.[17] In a recent

GLOSSARY

AAN = American Academy of Neurology; **CMD** = congenital muscular dystrophy; **DM** = myotonic dystrophy; **DMD** = Duchenne muscular dystrophy; **FSHD** = facioscapulohumeral dystrophy; **HRQoL** = health-related quality of life; **MD** = muscular dystrophy; **QoL** = quality of life; **WG** = Muscular Dystrophy Measure Development Work Group.

Supplemental data at Neurology.org

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US study,[18] national costs per annum were $787 million and $448 million for DMD and DM.[18]

METHODS The American Academy of Neurology (AAN) formed an interdisciplinary Muscular Dystrophy Measure Development Work Group (WG) to define quality measures to support the delivery of high-quality care and improve outcomes for patients with MD. The WG focused on gaps in care, identified areas of improvement, and reviewed available clinical evidence. Process, outcome, individual practitioner, and system-level measures were considered, following the AAN Quality and Safety Subcommittee process for measure development.[19] The WG evaluated the relevance of each measure to the 6 aims of health care improvement recommended by the Institute of Medicine (National Academy of Medicine).[20] Appendix e-1 on the *Neurology*® Web site at Neurology.org details the measurement set, as well as topic importance, desired outcomes, and evidence and literature search.

RESULTS This measurement set focuses on DMD, congenital MD (CMD), FSHD, myotonic MD, and limb-girdle MD, for which evidence supporting a gap in care was present. Our literature search identified 259 relevant recommendations from 19 clinical practice guidelines. Peer-reviewed publications were used where guidelines did not exist.

The WG evaluated 91 recommendations based on the strength of evidence, validity, clinical relevance, gaps in care, and feasibility, to serve as the basis for 14 draft measures. At an in-person meeting on September 16, 2013, the WG reviewed and eliminated 5 draft measures, including 2 outcome measures (patient-reported QoL and satisfaction with care) due to lack of high-level evidence.

The 9 selected draft measures were posted on the AAN Web site for a 30-day public comment period. Sixty-three comments were received, with resultant revisions. The final set of 9 measures was approved by the WG, the AAN Quality and Safety Subcommittee, the AAN Practice Committee, and the AANI Board of Directors. The table summarizes the measures. The complete measure set is available online (appendix e-1).

DISCUSSION A brief rationale for each measure is provided.

Corticosteroid treatment in DMD. Despite the evidence for beneficial effects of corticosteroids on muscle strength, preserving ambulation, improving pulmonary function, delaying onset of cardiomyopathy, and reducing the need for scoliosis surgery in DMD,[21] they remain underused. In a population-based cohort study, only 50.9% of individuals with DMD had received corticosteroids, and use varied widely across clinics (8.4%–80.2%).[22] In a recent survey, approximately 16% of neuromuscular specialists reported not using corticosteroids for DMD.[23]

Multidisciplinary care plan. The management of MD is complex, requires input from several specialists, and is dependent on the subtype and stage of MD. Care coordination is crucial to ensure that patients have access to relevant specialists because of the need for multisystem management. This may be performed by a wide range of health care professionals, who should be able to identify potential complications proactively and appropriately refer for management. A multidisciplinary care model with a network of providers is also endorsed by the Muscular Dystrophy Association.[10,12,24,25]

Pulmonary evaluation. Many MDs are associated with pulmonary complications.[10,12] Impending respiratory failure may not be preceded by symptoms, and may only be identified by pulmonary function testing. Respiratory failure is a major source of morbidity, interfering with cognitive function and negatively affecting QoL.[10,12] Noninvasive ventilation and treatment of sleep-disordered breathing improve QoL and prolong survival.[26]

Cardiac evaluation. Cardiac abnormalities (dysrhythmias, conduction disturbances, and cardiomyopathy) are prominent features of several MDs, and common causes of death.[10,27] Patients often do not have symptoms that precede cardiac dysfunction or sudden cardiac death, and cardiac complications may only be identified through testing. Detection and appropriate management of cardiac dysfunction are essential to reduce morbidity and mortality.[10,27] Patients with MD have improved QoL following appropriate medical treatment, device placement, or surgery for cardiac complications.[25]

Scoliosis evaluation. Spine deformities can occur in several MDs, resulting in pain, functional impairment, gait problems, and compromised pulmonary function. Their management is essential to reduce discomfort, preserve mobility or ability to sit in a wheelchair, and reduce pulmonary complications.[10,12,25] Management of scoliosis involves multiple modalities; this measure addresses only the detection of scoliosis.

Physical, occupational, or speech/swallowing therapy. Maintaining mobility and functional independence are imperative to maximize QoL. This includes prevention and management of comorbidities, both expected (joint contractures, scoliosis, osteoporosis, pain, dysphagia, restrictive lung disease) and acquired (obesity, stress fractures).[10,11]

Monitoring nutritional status. Dysphagia and limb weakness may reduce oral intake, resulting in nutritional compromise and failure to thrive.[10,12] Maintaining adequate nutrition and body weight is essential for optimizing strength, function, and QoL. Patients with CMD often have a growth curve below that expected for age, which may be from

Table	American Academy of Neurology muscular dystrophy quality measures
Duchenne muscular dystrophy pharmaceutical treatment	
Patients with Duchenne muscular dystrophy prescribed appropriate disease-modifying pharmaceutical therapy	
Muscular dystrophy management	
Muscular dystrophy multidisciplinary care plan developed or updated	
Evaluation of pulmonary status ordered	
Evaluation of cardiac status ordered	
Scoliosis evaluation performed	
Patient referred for physical, occupational, or speech/swallowing therapy	
Nutrition status or growth trajectories monitored	
Patient queried about pain and pain interference with function	
MD planning and patient engagement	
Patient counseled about advanced health care decision-making, palliative care, or end-of-life issues	

treatable causes (recurrent infections, cardiopulmonary complications).[28] When oral intake is inadequate, other means of maintaining nutritional intake such as percutaneous endoscopic gastrostomy may be required.[10,12] Conversely, patients with MDs are also prone to obesity.[25,28] Corticosteroid treatment can exacerbate weight gain in DMD.[25]

Evaluation for pain. Between 68% and 82% of patients with MDs experience pain, which may be underrecognized.[29,30] Untreated pain interferes with physical functioning and is associated with depression.[31,32] Nonpharmacologic and pharmacologic interventions are available to treat pain and improve attendant comorbidities.

Advanced health care decision-making, palliative care, and end-of-life issues. Patient-centered, proactive, informed, and collaborative decision-making is important in the care of MD. This includes sensitively preparing patients and families for the long-term consequences of MD and engaging in discussions regarding end-of-life care. This helps them to explore options, come to terms with their condition, and prepare for the complications of their form of MD, avoiding hasty decisions made during a medical crisis.[10,28,33] The plan should be reviewed at regular intervals as decisions may change. However, end-of-life care discussions may not be appropriate in patients with early or mild MD, especially in childhood. Palliative care minimizes suffering and improves QoL in patients with MD.[34]

These quality measures, based on evidence-based practice guidelines, represent the minimum standards of care for patients with MDs. They are intended to be applicable across all levels of medical care, from primary care providers to tertiary multidisciplinary clinics. This is essential, because distance and impaired mobility may make travel to tertiary centers difficult or impossible for patients with severe or end-stage disease. Managing the complications of MDs reduces morbidity, prolongs survival, and improves QoL. Implementation of these measures aims to standardize and improve the quality of care of patients with MD.

AUTHOR CONTRIBUTIONS

Dr. Narayanaswami contributed to study concept and design, acquisition of data, analysis or interpretation of data, drafting/revising the manuscript, critical revision of the manuscript for important intellectual content, and study supervision. Dr. Dubinsky contributed to study concept and design, acquisition of data, analysis or interpretation of data, drafting/revising the manuscript, and study supervision. Dr. Wang contributed to study concept and design, acquisition of data, analysis or interpretation of data, drafting/revising the manuscript, and study supervision. G. Gjorvad contributed to study concept and design, acquisition of data, analysis or interpretation of data, drafting/revising the manuscript, critical revision of the manuscript for important intellectual content, and study supervision. Dr. David contributed to acquisition of data and drafting/revising the manuscript. Dr. Finder contributed to acquisition of data and drafting/revising the manuscript. Dr. Smith contributed to acquisition of data and drafting/revising the manuscript. Dr. Cheng contributed to acquisition of data and drafting/revising the manuscript. Dr. Shapiro contributed to acquisition of data and drafting/revising the manuscript. Dr. Mellion contributed to acquisition of data and drafting/revising the manuscript. Dr. Spurney contributed to acquisition of data and drafting/revising the manuscript. Dr. Wolff contributed to acquisition of data and drafting/revising the manuscript. Dr. England contributed to study concept and design, acquisition of data, analysis or interpretation of data, drafting/revising the manuscript, critical revision of the manuscript for important intellectual content, and study supervision.

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P. Narayanaswami serves on the Blue Cross Blue Shield of Massachusetts Pharmacy and Therapeutics Committee; serves as a consultant for both Advanced Medical and Boston Clinical Research Institute; and has

received funding from MERZ Pharmaceuticals, NIH, and AHRQ for clinical research studies. R. Dubinsky serves on an advisory board for Allergan Pharmaceuticals on the treatment of cervical dystonia and chronic daily headache and has received honoraria and travel from them for speaking engagements; and has received funding from the National Institute of Neurological Disorders and Stroke, NIH, and HHS for 3 clinical research studies. D. Wang serves on the speaker's bureau of BMS and Boehringer-Ingelheim. G. Gjorvad reports no disclosures relevant to the manuscript. W. David has been involved in the production of a video CME course on EMG and neuromuscular medicine. J. Finder has delivered lectures and received travel paid by the Muscular Dystrophy Association of New Zealand, Parent Project Muscular Dystrophy, and Muscular Dystrophy Association of Western Australia; served as a peer reviewer for several medical journals and as an unpaid board member of *RT* magazine; and has served as a defense consultant on a non-neuromuscular disease case (no affidavit or testimony was provided). B. Smith has received compensation and travel for Grand Rounds at the University of Vermont for lectures on Autonomic Physiology and Clinical Neurophysiology. J. Cheng has served in an editorial capacity at the following publications: *Pain Practice*, *World Journal of Anesthesiology*, and *Journal of Perioperative Science*; is the inventor of 3 devices for pain management, 2 of which have been submitted for patent; is an editor of the textbook *Fundamentals of Pain Medicine*; has received compensation from the Taiwan Pain Society for lectures he delivered; and has received funding from the NIH on 3 clinical trials involving pain management. F. Shapiro receives royalties from Elsevier Publishers for a book published in 2002. M. Mellion received funding for 3 studies on peripheral neuropathy. C. Spurney serves on a scientific advisory board for Nicox. J. Wolff reports no disclosures relevant to the manuscript. J. England serves as the Editor-in-Chief of the *Journal of the Neurological Sciences*. Go to Neurology.org for full disclosures.

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