Primary Care and Emergency Department Management of the Patient With Duchenne Muscular Dystrophy

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abstract

Primary care providers (PCPs) are usually the first point of contact with the health care system for patients with Duchenne muscular dystrophy (DMD), and patients often present to emergency departments in which providers have little experience in dealing with this condition. With this article, we give primary care and emergency medicine providers a background in the common issues that affect people with DMD. By acquiring some specialized knowledge about the multisystem medical complications of DMD and by applying general principles of primary care, such as timely immunization, anticipatory safety counseling, behavioral screening, and routine nutritional and developmental assessments, the PCP can be a valued and effective medical provider to patients with DMD. The PCP can provide access to and effective coordination among the patient's specialty caregivers. Moreover, the PCP can become a trusted advisor to the patient and his family about important medical decisions, as well as issues in the psychosocial, behavioral, and educational domains. This article also contains a "pocket guide" used to assess and manage common urgent medical problems that cause patients with DMD to seek care in the emergency department. With the background information discussed in this article, both PCPs and emergency medicine physicians can skillfully care for patients with DMD in their respective settings, optimizing patient outcomes.

THE MEDICAL HOME

Primary care providers (PCPs) are usually the first point of contact with the health care system for patients with Duchenne muscular dystrophy (DMD). Depending on age, patient choice, and local circumstances, PCPs are those physicians or nurse practitioners in the fields of pediatrics, family medicine, or internal medicine who are engaged in providing a "medical home" to their patients. The primary care medical home (PCMH) is defined as a "model or philosophy of primary care that

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is patient-centered, comprehensive, team-based, coordinated, accessible, and focused on quality and safety."[1] A basic tenet of the medical home is that it benefits all patients, particularly those with special needs[2]; in fact, the processes of care required to provide a medical home were first developed for children with special needs.[3]

PCPs are encouraged to familiarize themselves with rare diseases like DMD. General guidance exists for the care of patients with neuromuscular diseases and can be used as a reference or "care map." Specifically, in this article, we expand on content found in the 2018 DMD Care Considerations, sponsored by the Centers for Disease Control and Prevention (CDC).[4–8] For the most part, people with DMD should receive the same primary care interventions as other patients. However, as outlined here, specific medical issues arise more commonly in patients with DMD, and it is critically important for the PCP to monitor, recognize, and act on those issues.

As the medical home provider, the PCP is responsible for the comprehensive and coordinated management of the patient. Traditional primary care tasks, such as immunizations, anticipatory guidance, and disease specific screening, are completed, as are health maintenance examinations and sick visits. In addition, the PCP partners with the patient's specialists to provide an environment of ongoing and seamless care.

KEY PRIMARY CARE MEDICAL ISSUES FOR PATIENTS WITH DMD

Psychosocial Management

Developing and maintaining the relationship between the patient, family, and PCP is a core component of the medical home. Although the PCMH is a team concept, each patient's care team is led by a personal physician who knows him well. The pediatrician is expert in the developmental stages from infancy through adulthood and can guide the family through the expected behavioral challenges and developmental tasks that face children with neuromuscular diseases. These include toilet training, bedtime routines, school preparation, and discipline. These may be more challenging in children with DMD, who are physically dependent on their families and cannot easily assert their growing independence.

The PCP can be instrumental in identifying families who are under stress and can link those families with community resources that enhance functionality and coping, including individual or group counseling, parent training, or association with disease-specific support groups.

The PCMH will have ample contacts with schools in the area and is well versed in the supports available to children in the United States with special educational needs, such as individualized educational programs and 504 plans.[9] Accommodations for both physical and educational needs can be covered in these plans. The PCP can help coordinate interactions between the family and school system to assure that patients with DMD have every opportunity to excel in school. For more details, see the "Psychosocial Management of the Patient With Duchenne Muscular Dystrophy" article that is part of this supplement.[10]

The PCMH is a natural place for discussions of transition to autonomy to begin. The American Academy of Pediatrics recommends that discussions of transition begin at age 12 years.[11] This is a patient-centered process that should include multiple domains, including continuation of medical management, educational and vocational planning, living arrangements, and advanced care planning. Patients with DMD should be encouraged to take on more responsibility for their own care as they progress through adolescence into adulthood. For more details, see the article "A Transition Toolkit for Duchenne Muscular Dystrophy," which is part of this supplement.[12]

For adults, the internist or family practitioner is expert in care coordination for patients with chronic illness and is key in promoting healthy behaviors, which can be valuable for adults with DMD. The PCP should ensure that adults with DMD have completed advanced directives and appointed a health care power of attorney.

Preventing Illness and Injury

With a few exceptions, patients with DMD should receive the same immunizations, on the same schedule, as all other patients. The most current recommendations for immunizations can always be found at the immunization schedules portion of the CDC Web site.[13] None of the nonlive vaccines are contraindicated for patients with DMD. Patients with DMD, or others who take daily corticosteroids (at doses >20 mg or 2 mg/kg per day of prednisone equivalent), should be considered immunosuppressed and should not receive live vaccines. Patients on intermittent dosing of corticosteroids are not considered immunosuppressed.[14] The common live vaccines include the measles-mumps-rubella and varicella vaccines. All patients with DMD (older than 6 months) should receive the inactivated influenza vaccine annually. Family members and other close contacts of patients with DMD should be encouraged to receive the annual influenza vaccine as well. This is known as "cocooning" the individual.

The measles-mumps-rubella and varicella vaccines are usually given at age 12 to 15 months and again at age 4 to 6 years. However, the

2 doses may be given as soon as 4 weeks apart if necessary. If possible, these live virus vaccines should be administered before initiating corticosteroid therapy. Other live vaccines not commonly used in the United States, such as oral polio vaccine and yellow fever vaccine, should be avoided in patients taking daily corticosteroids.

Because patients with DMD are prone to respiratory illness, they should receive pneumococcal vaccines (including the pneumococcal conjugate vaccine and pneumococcal polysaccharide vaccine). Guidelines on these vaccines and their timing are available from the CDC,[15] the Immunization Action Coalition,[16] and the Parent Project Muscular Dystrophy.

Good dental health is a cornerstone of good overall health and is often problematic to attain for people with disabilities.[17] The PCMH should ensure that every patient with DMD has access to dental care. In many areas, water is not fluoridated, so the PCP may prescribe a fluoride supplement. In addition, the PCMH may provide fluoride varnish in the office. Maintenance of good oral health is particularly important for children with DMD because of a high prevalence of osteoporotic fractures.[18–22] Such patients may be prescribed bisphosphonate treatment, and good dental care can decrease the likelihood of developing osteonecrosis of the jaw as a side effect of this treatment.[23]

Unintentional injuries due to accidents involving automobiles or bicycles, drowning, or firearms are the primary cause of death for patients aged 1 to 44 years.[24] Safety counseling is an important task of the PCMH. Particular considerations for patients with neuromuscular disease include proper vehicle restraint. For patients with good trunk control who can safely transfer into the car, the use of a car seat, booster seat, and/or seatbelt is mandatory. Patients with poor trunk control and patients who ride in vehicles in their wheelchair, require specialized passenger safety measures. The manufacturer's recommendations for seatbelt use should always be followed. Other recommendations for the safe transport of children with disabilities have been published.[25]

Screening for Illness

Formal, standardized developmental screening is a vital task for the PCP and recommended by the American Academy of Pediatrics.[26,27] Many children with neuromuscular diseases are first identified through this process during their routine PCMH visits. However, developmental screening does not end with the identification of a neuromuscular disorder, because a patient may have a concurrent developmental problem, either related to the underlying neuromuscular disease or distinctly unrelated to the disease. For example, children with DMD have a higher rate of intellectual disability, learning disability, language disorder, autism, and attention-deficit/ hyperactivity disorder than unaffected children.[28] For more details, see the article, "Psychosocial Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[10]

Identification of hearing and vision impairment is important in this population because these problems are often correctable and can improve school and social performance.[29,30] Patients who use steroids are at risk for the development of cataracts, and appropriate screening should occur.[28]

Screening for cardiovascular risk factors, such as hypertension and hypercholesterolemia, should possibly be conducted more frequently in patients with DMD.[31–33] For more details, see the specialty article, "Cardiac Management for the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[34]

Mood disorders are more prevalent in patients with disabilities than in the general population.[35] However, it is wrong to assume that all patients with disabilities are depressed. Standardized screening is recommended by the US Preventive Services Task Force,[36] and standard treatment may be offered. The prevalence of substance use disorders has not been reported among patients with neuromuscular disease, so a general screening strategy may be applied.[37]

Patients with DMD severe enough to limit weight-bearing exercise or who take chronic corticosteroids are at increased risk for bone fractures.[18–22] Dual-energy radiograph absorptiometry scanning, attention to nutrition, and encouragement of weight-bearing exercise have potential roles in a strategy to optimize bone health in patients with DMD. For more details, see the article, "Bone Health and Osteoporosis Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[38]

Nutrition and Growth

The PCMH can be an excellent resource for general nutrition advice and monitoring over time. For the patient with DMD, particular attention should be paid to ensuring adequate fluid intake, adequate calcium and vitamin D intake, and avoidance of food that is high in calories but low in nutritional content (junk food). Registered dieticians may be engaged by either the PCMH or neuromuscular specialist to provide counseling and follow-up.

Both weight and height can be difficult to measure in children with neuromuscular disabilities. The PCMH must make accommodations for patients who cannot stand on a

regular scale. If a patient is weighed in a wheelchair, the wheelchair must be reweighed at each visit with that weight subtracted to ensure accurate measurement. Patients who are unable to stand for measurement with a stadiometer may be measured supine on a standing board or by using arm span or ulnar estimates of height. However, the true height may be difficult to measure because of scoliosis and joint contractures. Thus, BMI calculations can be unreliable.

Because of dysphagia, many patients with advanced DMD will have feeding tubes that augment or replace oral feeding. Such patients do not need to achieve a weight that is considered healthy for unaffected patients of the same age and sex. In many cases, this would make the patient unnecessarily heavy, with detrimental effects on transfers, mobility, and respiratory status.

An important concept for monitoring nutrition in DMD is that an individual measurement is less important than the pattern of growth over time. Growth curves for patients with DMD do not exist, so children should be plotted on standard growth curves[39] to monitor change over time. Many children with neuromuscular disease who are experiencing good growth and nutrition will be "on their own curve" (ie, below the fifth percentile for age), paralleling the standard growth curves. Other objective measures that can be employed to monitor nutrition status include body fat percentage estimation by measuring skinfold thickness, electrical impedance, or dual-energy radiograph absorptiometry scan, but these are often outside the purview of the PCMH. For more details, see the article, "Nutritional and Gastrointestinal Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[40]

MANAGEMENT OF ACUTE PROBLEMS

Patients with DMD may present with any of the acute problems experienced by unaffected patients, but some problems are more common or of particular importance. When symptoms are mild to moderate, care may be sought in the primary care setting, whereas severe problems may require evaluation in the emergency department (ED).

1. Chest Pain. DMD is associated with cardiomyopathy, and a cardiac cause of chest pain and/or dyspnea must always be considered. However, noncardiac causes of chest pain are common and include esophagitis and muscle strain, particularly with coughing. These can often be handled in the primary care setting and can be identified through a careful history and physical examination. Chest radiography can be useful to rule out pneumonia, pneumothorax, or pleural disease. For more details, see the Cardiology specialty article that is part of this supplement.[34]

2. Abdominal Pain. Constipation is extremely common in patients with DMD, but patients may also be at risk for more serious pathology, such as peptic ulcer disease (as a result of corticosteroid use), as well as all of the usual serious intra-abdominal pathologies, including appendicitis. For more details, see the specialty article, "Nutritional and Gastrointestinal Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[40]

3. Back pain is a common complaint. Patients with DMD have a high risk for vertebral fractures,[41] as well as renal colic.[42] Back strain related to abnormal body mechanics and problems with seating are common as well. Unless symptoms are severe, these problems can often be evaluated in the primary care setting with appropriate physical examination, with or without radiographs.

4. Dyspnea needs to be carefully evaluated. It can be caused by several serious etiologies, such as pneumonia or atelectasis, untreated chronic or acute hypoventilation, pulmonary or fat embolism, or worsening cardiomyopathy with heart failure. The severity of the dyspnea may be out of proportion to the examination; the patient may not appear to be distressed because he cannot manifest the increased work of breathing. A chest radiograph and an oxygen saturation measurement are helpful. Even mild hypoxemia (ie, oxygen saturation <95% in room air) should be viewed as highly concerning. The provider needs to be keenly aware of the potential for hypercapnia in patients with neuromuscular disease. With hypoventilation, early administration of noninvasively assisted ventilation is necessary. Supplemental oxygen therapy without assisted ventilation must be used cautiously to prevent inhibition of the patient's respiratory drive. Although minor respiratory illnesses without dyspnea can be treated safely in the outpatient setting, more serious symptoms are more safely evaluated in the ED (see the "In the ED" section below) or by an appropriate specialist. Empirical antibiotics are often given in the ambulatory setting to patients with impaired cough, even if the underlying illness is thought to have a viral cause. More important than antibiotic coverage is the need to increase the intensity of the patient's assisted ventilation and mucus clearance therapies; for example, by having the patient use his cough-assist device more frequently than usual and by adding daytime ventilatory

assistance to the patient's usual schedule of nocturnal ventilation (most patients will be ventilated noninvasively, through a nasal or full-face mask; some may have a tracheostomy). For more details, see the article, "Respiratory Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[43]

5. Fatigue is an extremely common complaint and can have many causes. For patients with DMD, the following should be considered: inadequate sleep time, chronic nocturnal hypoventilation, advancing heart failure, and mood disorder. The approach to this problem should include a discussion of sleep habits, recognizing that a person's need for sleep varies over time and is different among individuals, and screening for depression. Laboratory investigations may include testing for hypothyroidism and anemia. An abnormally elevated red blood cell count or serum bicarbonate level may signal unrecognized hypoventilation, with nocturnal hypoxemia and hypercapnia. Nocturnal oximetry, oximetry and/or capnography, or polysomnography may be necessary and should be coordinated with the neuromuscular specialist. A cardiac evaluation also may be needed.

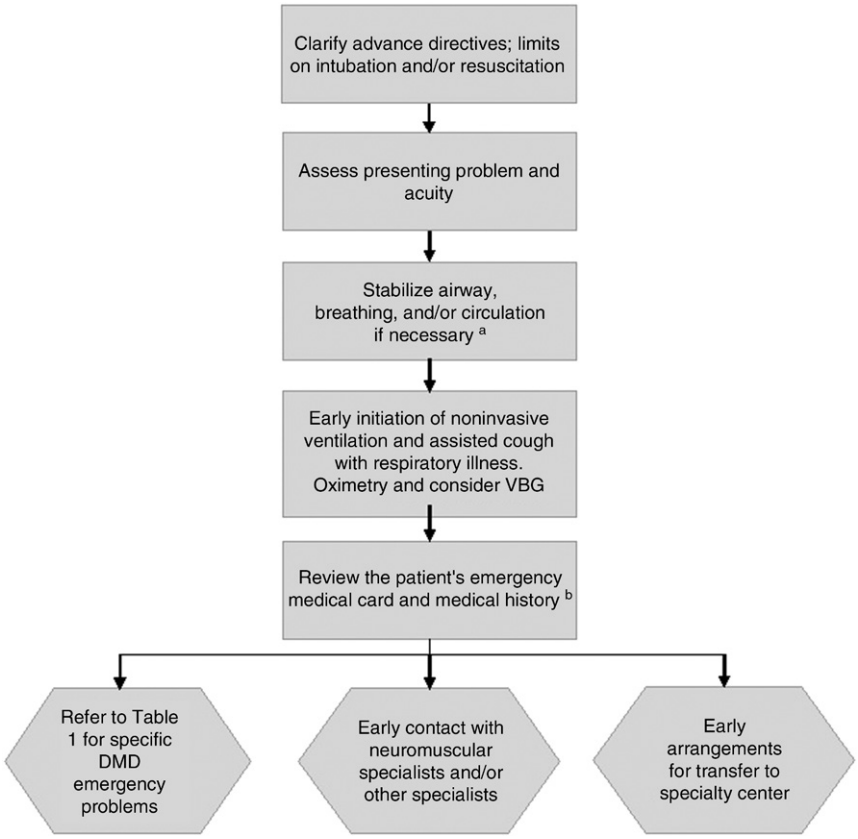

FIGURE 1

Initial approach to emergency management of patients with DMD. [a]Intubation is often difficult because of macroglossia and limited jaw mobility. If intubating, consider urgent anesthesia involvement. [b]Assess baseline cardiopulmonary function, medical device use, and chronic medications, especially glucocorticoids. Mild cardiomyopathy is common by age 18 years. Consider fat embolism syndrome with dyspnea, altered mental status, especially after fractures. VBG, venous blood gas.

IN THE ED

For acute medical problems, it is optimal for patients with DMD to receive care from providers who know them personally, are aware of their medical histories, and are familiar with the management of urgent medical situations commonly affecting people with DMD. This scenario cannot always occur, and it is not uncommon for patients with DMD to present to the local ED with an urgent medical problem. Although formal studies are lacking, the experience of patients with DMD and their families suggests that emergency medicine providers are often unfamiliar with DMD and its complications. See Fig 1 for an algorithm outlining a quick initial approach to emergency management of patients with DMD. This algorithm is linked to Table 1, which is intended to be used as a "Pocket Guide" for emergency medicine providers. For additional details, see the relevant 2018 DMD Care Considerations article.[7]

Certain key issues are of particular importance in regard to the care of patients with DMD in the ED. The patient should have an emergency card that has been provided by his neuromuscular specialist. The card should provide a brief medical summary, including the patient's baseline pulmonary and cardiac function and recommendations for initial management of any acute medical issues that are likely to occur. The emergency medicine provider should ask for this card at the time of initial assessment and also should ask about advanced directives and resuscitation status. The patient's neuromuscular specialist should be consulted early in the ED visit for assistance in management. Respiratory issues are probably the most common reason for patients with DMD to present to the ED. The patient's own perception of his symptoms must be taken seriously, because patients with DMD rarely appear

TABLE 1 Overview of Key Issues Related to DMD Emergency Care

Advance directives, history, and contacts
Determine whether there are restrictions on resuscitation
Ask for the patient's emergency card and baseline test results, including ECG results
Obtain a brief history with a focus on baseline respiratory and cardiac status, including the use of relevant devices and medications
Determine whether the patient is treated with chronic steroid therapy
Contact the patient's neuromuscular specialist
Breathing problems
Ask about respiratory symptoms and home equipment
Monitor SpO_2 levels via pulse oximetry; even mild hypoxemia (SpO_2 <95% in room air) is a concern; do a blood gas analysis if necessary
Treat with noninvasive ventilation and frequent application of a cough assistance device (or manual assisted coughing if device is unavailable); use the patient's home equipment when available
Obtain a portable chest radiograph
Obtain early consultation with a respiratory therapist and respiratory physician
Cardiac problems
Ask about cardiac symptoms
Monitor heart rate and rhythm
Obtain an ECG (this is typically abnormal and Q waves might be expected) and portable chest radiograph
Measure blood levels of B-type natriuretic peptide, troponin I, or both, as indicated
Consider worsening cardiomyopathy, congestive heart failure, and arrhythmias
Obtain an echocardiogram when necessary
Obtain early consultation with a cardiologist
Endocrine problems
Determine whether stress steroid dosing is necessary
For critical adrenal insufficiency, administer intravenous intramuscular hydrocortisone: 50 mg for children <2 y old; 100 mg for children ≥2 y and adults
In less critical situations, consult the PJ Nicholoff Steroid Protocol
Obtain early consultation with an endocrinologist
Orthopedic problems
Assess for long-bone or vertebral fractures as indicated
Review critical precautions related to sedation and anaesthesia if applicable (see text)
Consider fat embolism syndrome if individual has dyspnea or altered mental status
Obtain consultation with an orthopedic specialist early in the process
Disposition after discharge from emergency care
Be aware that most patients will need hospital admission (eg, to initiate or intensify respiratory or cardiac therapy, or to manage fractures)
Early in the process, initiate emergency transport to a center specializing in the care of patients with DMD, in cooperation with the individual's neuromuscular specialist

This table is adapted with permission from Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, an update, part 3: primary care and emergency medicine, psychosocial care, and transitions of care across the lifespan. *Lancet Neurol.* 2018;17(5):447. B-type, brain; SpO_2, blood oxygen saturation.

to be in respiratory distress because they cannot manifest the increased work of breathing. The importance of initiating noninvasive ventilatory support and assisted coughing early cannot be overstated. Intubation in patients with DMD is difficult[44] and usually not necessary if noninvasive ventilation and coughing are initiated in a timely way. A chest radiograph and, if necessary, a venous blood gas should be obtained, followed by consultation with the patient's neuromuscular specialist. The patient's noninvasive ventilation should not be interrupted for a trip to the radiography suite; instead, a portable film can be obtained. If the patient is on chronic steroid therapy, stress-dose steroids should be administered. A rare but life-threatening cause of respiratory distress is fat embolism syndrome. Patients with DMD are at risk for this condition because of increased fat in bone marrow. The typical presentation is new onset dyspnea and/or mental status changes, especially after an orthopedic injury (typically long-bone fracture) or surgery.[45]

SELECTED ISSUES IN LONG-TERM MONITORING

Because patients with DMD may access their PCMH more frequently than the neuromuscular clinic, for routine management and for urgent visits, the PCP should be aware of the potential for deterioration in pulmonary function. In early stages, signs of pulmonary impairment may be subtle. Hypoventilation can cause symptoms such as fatigue, mood changes, morning headaches, and deteriorating school and/or work performance. Respiratory muscle weakness often presents with more frequent or prolonged upper respiratory infections due to impaired cough. Decreasing exercise tolerance may be related to advancing muscle disease, but early respiratory insufficiency should be considered as well. Early investigation regarding hypoventilation and impaired cough strength can take place within the PCMH in coordination with the neuromuscular clinic. Relevant evaluations include serial pulmonary function testing, end-tidal or transcutaneous blood carbon dioxide testing, blood tests used to assess for hypercarbia, awake and nocturnal pulse oximetry, nocturnal oximetry and/or capnography, and/or polysomnography, if available. It is important to share the results of these tests with the relevant specialists for assessment and action. In contrast to patients with obstructive sleep apnea, inadequate ventilation during sleep must be treated with assisted ventilation (for example, with bilevel nasal ventilation rather than continuous positive airway pressure or supplemental oxygen, which do not treat the underlying ventilatory impairment and can be harmful in this clinical situation). For more details, see the article, "Respiratory Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[44]

In patients with DMD, the PCP also must be alert for early signs of cardiomyopathy and for cardiac conduction abnormalities. Affected patients may present with fatigue, dyspnea, edema, dizziness, or syncope. The physical examination in early cardiomyopathy is often subtle but may reveal jugular venous distension, hepatomegaly, or edema. As with suspected pulmonary insufficiency, the PCP may begin the investigations, but the relevant specialist (in this case, the cardiologist) should be involved early in the process. Standard testing can be considered, such as chest radiograph, electrocardiogram (ECG), and echocardiogram. However, interpretation can be complex and some "abnormalities" (such as Q waves on the ECG) are expected findings in patients with DMD. For more details, see the article, "Cardiac Management for the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[34] For patients treated with corticosteroids, the PCP must be vigilant for signs of adrenal insufficiency and ensure that the patient has an understanding of how and when to use stress-dose steroids. Increased steroid doses are commonly given as a precaution during febrile illnesses or in response to surgery or trauma. For more details, see the specialty article, "Obesity and Endocrine Management of the Patient With Duchenne Muscular Dystrophy," that is part of this supplement.[46]

CONCLUSIONS

Patients with DMD can benefit from the care and resources provided by a PCMH and by having a PCP as their point of first medical contact. By acquiring some specialized knowledge about the multisystem medical complications of DMD and by applying general principles of primary care, the PCP can become a valued member of the DMD care team. Additionally, the PCP can access the patient's specialty caregivers and coordinate their recommendations. Moreover, the PCP can become a trusted advisor to the patient and his family about important medical decisions and issues in the psychosocial, behavioral, and educational domains, helping the patient and family to optimize their quality of life. This article also contains an approach to assessing and managing urgent medical problems that cause patients with DMD to seek care in ED settings. By understanding the special issues that affect patients with DMD when they are acutely ill, emergency medicine providers can provide effective and efficient medical management, improving patient outcomes, preserving continuity of care, and avoiding preventable medical complications.

ABBREVIATIONS

CDC: Centers for Disease Control and Prevention
DMD: Duchenne muscular dystrophy
ECG: electrocardiogram
ED: emergency department
PCMH: primary care medical home
PCP: primary care provider

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