Address correspondence to Dr John T. Kissel, The Ohio State University Wexner Medical Center, Department of Neurology, 395 W. 12th Avenue, 7th Floor, Columbus, OH 43210, john.kissel@osumc.edu.

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Ms King and Dr Kissel discuss the unlabeled use of corticosteroids in the treatment of Duchenne muscular dystrophy.

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Multidisciplinary Approach to the Management of Myopathies

Wendy M. King, PT; John T. Kissel, MD, FAAN

ABSTRACT

Purpose of Review: Aside from some inflammatory myopathies and very few genetic disorders, there are no therapies that make most patients with myopathies stronger. Consequently, the management of these patients can be frustrating for patients and their families as well as the clinicians taking care of them. Treatment of these patients must involve a comprehensive approach focused on limiting the secondary effects of skeletal muscle weakness, managing comorbidities associated with specific diseases, and, most importantly, optimizing patients' functional abilities and quality of life in terms of their ability to accomplish activities of daily living. While the approach to each patient differs depending on their disease, certain common themes can be addressed in each patient. This review highlights an approach centered on four conceptual themes ("the Four S's"): *Strength* therapies, *Supportive* care, *Symptomatic* therapies, and *psychological* support.

Recent Findings: Although relatively few well-designed studies have been done that highlight conservative management of patients with various myopathies, an emerging literature helps guide the clinician in certain key areas, especially in relation to cardiac and pulmonary management of these patients.

Summary: While disease-altering therapies have proven elusive for many muscle diseases, a multimodal approach to the conservative and supportive care of these patients can markedly improve their quality of life. Pharmacologic treatment options for specific myopathies will not be addressed in this article but are covered elsewhere in this issue of **CONTINUUM**.

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INTRODUCTION

Despite advances in the understanding of the genetics and molecular pathogenesis underlying most muscle diseases, specific therapies for most of these disorders have remained elusive. Aside from immunosuppressive therapy (eg, prednisone, IV immunoglobulin) for some inflammatory myopathies, corticosteroid therapy for Duchenne dystrophy, and enzyme replacement therapy for Pompe disease, disease-modifying therapies that

make patients stronger are lacking for most myopathies. Consequently, the management of myopathy patients must focus on conservative care to limit the effects of weakness on the joints, bones, and other systems; manage comorbidities associated with the diseases; and, most importantly, optimize patients' functional abilities and quality of life. All too often, patients are told "there is nothing to be done," when judicious bracing or a referral to an appropriate therapist would have a

profound impact on the patient's well-being and function.

Effective management of patients with myopathies requires a multimodal approach that includes a team of health care professionals. Conceptually, it is helpful to consider four categories of interventions: (1) *Strength* therapies aimed at improving strength (eg, prednisone for Duchenne muscular dystrophy [DMD]); (2) *Supportive* care aimed at problems resulting directly from muscle weakness (eg, respiratory compromise in some limb-girdle muscular dystrophies [LGMD]); (3) *Symptomatic* care aimed at treating problems that are not directly related to muscle disease but are part of the condition nonetheless (eg, cardiac involvement in myotonic dystrophy [DM]); and (4) *Psychological* support aimed at improving patients' mental outlook and providing state of the art information to patients about their diseases. Although somewhat artificial, this "Four S" approach provides a useful conceptual framework for the clinician to consider when managing patients with otherwise "untreatable" muscle disorders.

STRENGTH THERAPIES

Since weakness is the predominant manifestation of most muscle disorders, patients naturally are most interested in therapies to improve strength. This type of therapy is best exemplified in the various immunotherapies effective in dermatomyositis (DMY), polymyositis (PM), and immune-mediated necrotizing myopathy. High-dose daily corticosteroids improve strength and pulmonary and cardiac function in DMD and significantly prolong ambulation.^{1,2} Enzyme replacement therapy is life-saving in infantile Pompe disease and improves pulmonary function and 6-minute walk times in the late-onset disease.³

Side effects accompanying most pharmacologic interventions can counteract the strength benefit and must be aggressively managed. Creatine monohydrate is an over-the-counter supplement that has been evaluated in several neuromuscular disorders. A Cochrane review concluded that creatine monohydrate (3 g/d to 20 g/d) slightly increased strength and function in dystrophinopathies and DM2 and inconsistently in DM1; it also produced slight functional improvement in PM and DMY, with no benefit in facioscapulohumeral dystrophy (FSHD). Creatine was well tolerated, but no study exceeded 6 months.⁴ Although many patients are eager to try creatine because it is "natural," available over the counter, and popular among athletes, most do not stay on this supplement long because its effects are negligible.

Exercise

Patients with muscle disorders invariably ask whether exercise will improve their strength, and it is not uncommon for patients to begin intense exercise programs soon after diagnosis. While the concept that exercise can improve even damaged muscle is appealing, there is a theoretical risk that exercise may increase muscle damage, especially in the inflammatory myopathies and genetic disorders affecting structural proteins (eg, dystrophin in DMD). However, exercising has significant benefits beyond effects on strength, such as maximizing cardiopulmonary function, limiting osteoporosis, and improving patients' well-being. Unfortunately, there is a paucity of controlled trials evaluating the role of aerobic or resistive exercise in most myopathies. In the inflammatory myopathies, aerobic exercise in PM and DMY was well tolerated and led to significant improvement in maximal oxygen uptake, isometric

KEY POINT

- Effective management of patients with myopathies requires a multimodal approach that includes a team of health care professionals.

KEY POINTS

- In the studies done to date, low-intensity aerobic exercise does not seem to result in increased muscle damage as assessed by clinical, biochemical, or pathologic criteria.
- Patients beginning a strength training program should obtain careful baseline strength measurements via computerized muscle assessments and be closely followed periodically afterward.
- Aquatic therapy can be an efficient exercise, as it allows many muscles to be worked at the same time, but respiratory-compromised patients must use caution in chest-deep (or higher) water.

force, and functional scores in both a 6-week, small, randomized controlled trial and a longer 6-month trial.⁵ Resistive exercise is well tolerated and improves peak torque in adults with chronic myositis. Results in inclusion body myositis (IBM) have been mixed for resistive exercise, and no controlled trials have been performed. One 12-week open trial showed improvement in aerobic exercise and strength in four muscle groups but no improvement of the knee extensors or finger flexors.⁶ For dystrophic disorders, several short-term (less than 12 weeks) open studies found that low-intensity aerobic exercise (working at a heart rate that is 60% to 65% of maximum heart rate) is beneficial in FSHD, LGMD type 2I, and DM1, while a controlled trial concluded the same in Becker muscular dystrophy (BMD). The long-term effects of high-intensity aerobic exercise (working at 75% to 85% of maximum heart rate) in these populations, however, is less clear and needs further investigation.⁷ Resistance training over 6 months in patients with LGMD2 and BMD increased strength in some of the muscles tested, but results were varied.⁸ Importantly, no clinical, biochemical, or pathologic evidence indicated that the exercise led to increased muscle damage in any of the strength studies. A recent Cochrane report concluded that, "In myotonic dystrophy and facioscapulohumeral muscular dystrophy, moderate strength training appears not to do harm, but there is insufficient evidence to conclude that it offers benefit."⁹

Resistive exercise programs are designed to build muscles and improve strength through repetitive contractions against a resistive force, such as free weights, dumbbells, exercise machines, and resistive bands. Whether a patient with a myopathy should begin a

resistive exercise program depends on the extent and distribution of weakness and the patient's baseline activity. A basically sedentary person may benefit from a careful, low-intensity strengthening program while a person already active at work or home may not require resistive exercises in his or her daily routine. A patient starting a strengthening program should obtain baseline strength measurements before beginning the program with periodic (eg, every 6 months) follow-up evaluations scheduled to gauge the success (or failure) of the program. A physical therapist with experience in muscle disease and strength testing is the ideal person to perform the strength evaluations. It is also helpful for the patient to maintain a daily log of activities, exercises performed, and symptoms experienced on the following day. By documenting physical tasks along with physical well-being (including pain and fatigue), a patient with a myopathy will be able to determine whether an exercise program is beneficial.

Although no controlled trials of aquatic therapy in muscle disease have been done, therapists often encourage aquatic therapy because the buoyancy of the water assists mobility. Aquatic therapy can be an efficient exercise, as it allows many muscles to be worked at the same time, with the caveat that patients with reduced forced vital capacity (FVC) may experience dyspnea in water at chest level or higher. Adults may avoid aquatic therapy because of accessibility, safety, or social issues.

In all cases of myopathic patients for whom an exercise program is being considered, the clinician should:

1. Know the specific type of myopathy present, and whether there is cardiac involvement that would

require cardiologic consultation and clearance before an exercise program is initiated.

2. Assuming the cardiac status is satisfactory, reassure the patient that low-intensity aerobic exercise is probably beneficial, make sure the patient is aware of the symptoms of “overdoing it,” and suggest initiating the program under the supervision of a therapist.
3. Explain that there are few data to support a resistive/strengthening program and that, if one is chosen, no more than a low-intensity program should be started, and a baseline strength evaluation with periodic rechecks is indicated.
4. Advise the patient that data are insufficient to currently recommend high-intensity aerobic or resistive exercise for any type of myopathy.

SUPPORTIVE THERAPY

Several supportive and assistive measures are useful in limiting limb contractures, respiratory compromise, swallowing and gastrointestinal issues, and other complications that develop secondary to the weakness associated with most myopathies. The judicious implementation and monitoring of these measures is a crucial component to the management of myopathic patients.

Stretching and Positioning

Chronic or progressive muscle weakness often leads to soft-tissue contractures. These are caused by a complex set of factors including muscle strength imbalance, prolonged immobility, the effects of gravity, and other factors inherent in the disease pathogenesis. For example, hip, knee, and ankle joint contractures are common in nonambulatory patients and are especially difficult to prevent if the hip flexors, knee flexors, and ankle

plantar flexors remain stronger than the corresponding muscle antagonists. Contractures may sometimes also develop as a compensatory mechanism to preserve function. This appears to be the case in DMD, for example, when Achilles tendon contractures develop to help the boy continue standing by keeping his center of gravity behind the hip and in front of the knee (**Figure 7-1**).¹⁰ Fixed joint contractures are inevitable in some disorders (eg, DMD, BMD, some LGMD, congenital muscular dystrophies [CMD], Emery-Dreifuss muscular dystrophy [EDMD], Ullrich and Bethlem myopathies, hereditary IBM type 3, and severe X-linked centronuclear myopathy).

Although most clinicians and therapists recommend regular stretching for patients with chronic myopathies, no firm evidence has shown that soft-tissue joint contractures can be prevented even by aggressive positioning, range-of-motion exercises, or the use of night splints. A 2011 review of passive stretch (defined as self-, therapist-, or device-administered and including positioning, splinting, and serial casting) concluded that, “there is little or no effect of stretch on pain, spasticity, or active limitation in people with neurological conditions.”¹¹ The effectiveness of stretch has been best studied in DMD, in which a 2010 Cochrane review concluded that, “there is limited evidence supporting any intervention for improving ankle flexibility in patients with Duchenne muscular dystrophy.”¹² Recommendations for passive stretch or positional splinting should therefore be made on an individual basis, taking into account that specific patient and his or her family’s attitude toward therapy; available time; and motivation, expectations, and interests in such a program. Patients and families that elect to begin a stretching program should be

KEY POINT

- No convincing evidence has shown that passive range-of-motion exercise or stretching prevents contractures in patients with muscle disease.

FIGURE 7-1 A 14-year-old boy with Duchenne muscular dystrophy (on daily corticosteroids for 6 years) showing his posterior (A) and lateral (B) standing posture. He is nearing end-stage walking but ambulates short distances by maintaining his center of gravity behind his hips. He also must remain on his toes to stabilize his weak quadriceps. He regained ambulation following a left tibial fracture, but the required immobilization led to further Achilles tendon shortening and therefore a need for a built-up heel in the left shoe.

taught by a therapist how to perform the technique safely and correctly.

It is possible to temporarily improve joint range of motion in DMD by serial casting.¹³ This technique requires weekly cast changes and interferes with bathing or aquatic play but has a role in certain acute circumstances (a contracture following an injury and temporary immobility, for example). The use of botulinum toxin and other similar methods must be considered experimental at this time.¹⁴

Orthotics

Unlike with passive stretching, evidence suggests that function can be improved with appropriately pre-

scribed orthoses. Patients with FSHD, DM1, distal myopathies, and other disorders that cause significant ankle dorsiflexion weakness can benefit from ankle-foot orthoses (AFOs) to provide dorsiflexion assist and ankle stabilization. These may be custom made or off the shelf. Some off-the-shelf carbon fiber orthoses can provide plantar flexion dynamic assist in addition to maintaining neutral dorsiflexion (Figure 7-2). Stance-control knee-ankle-foot orthoses (KAFOs) (Figure 7-3) permit ambulation with the knee unlocked during swing phase and should be considered for appropriate patients with disorders associated with prominent quadriceps weakness, such as IBM

or some cases of muscular dystrophy (Case 7-1). Cervical orthoses provide support for weak neck flexors or extensors. Although several lightweight rigid cervical braces are available, patients often do not tolerate them because they make walking awkward and chewing practically impossible. Such orthotics may, however, decrease pain and muscle fatigue during activities such as watching television. Scapular retraction orthoses and lumbosacral corsets can also provide support in selected patients with axial weakness and back pain (eg, FSHD). Body habitus may predict how tolerable the device will be, as obesity may preclude comfort. Also, many patients with muscle disease will have difficulties putting the supports on without assistance. Thoracic lumbosacral orthoses (TLSOs) have been used in children with CMD to reduce the rate of scoliosis and allow the child to develop increased trunk length before planned scoliosis surgery (Figure 7-4).¹⁵ Hand or wrist splints are prescribed in an attempt to control joint contractures or to improve function. They typically work well initially, but if contractures or weakness progress, they must be regularly adjusted or refitted.

Surgical Intervention

The most important factor causing scoliosis in myopathy patients is lengthening and weakened trunk muscles attempting to remain vertical against gravity. The degree of scoliosis is related to the severity of truncal weakness and the age of the individual patient: curves progress more rapidly during growth spurts. Consequently, any congenital myopathy or dystrophy can result in severe scoliosis in the first decade of life. As the child grows, the curve may rapidly progress as the weak trunk muscles must work harder against gravity and the elongated

FIGURE 7-2 A patient in a “toe-off” style ankle-foot orthosis (AFO). This type of brace is often helpful for the patient with weak dorsiflexors, plantar flexors, or quadriceps weakness.

body. Boys with DMD not treated with corticosteroids and children with sarcoglycan-related LGMDs, Ullrich congenital muscular dystrophy, and CMD with rigid-spine syndrome also develop scoliosis by the teenage years. Essentially all pediatric myopathy patients should therefore be clinically monitored for scoliosis at every visit and scoliosis x-rays initiated when a curve is suspected.

The only proven nonsurgical treatment for the prevention for scoliosis is corticosteroid treatment in DMD. Boys treated with daily prednisone or deflazacort did not develop significant (over 10 degrees) scoliosis for at least 8 years.¹⁶ A prompt referral to an orthopedic specialist with experience in the management of scoliosis is

KEY POINT

- Pediatric myopathy patients should be clinically monitored for scoliosis, and x-rays should be initiated if a curve is suspected. During peak growth years (generally age 9 to mid-teenage years), yearly spinal x-rays should be obtained even without signs of a curvature.

FIGURE 7-3 Stance-control knee-ankle-foot orthosis (KAFO). This type of brace is appropriate if the patient's hip strength allows upright posture but quadriceps weakness promotes the knee to buckle. The patient may use a walker if hip strength declines.

indicated whenever a curve is progressing; spinal orthoses or growing rods may be indicated for younger children.¹⁵ Although the management of scoliosis by spinal fusion has improved in terms of decreased hospital days and recumbent time, no randomized controlled trials have been done to evaluate the benefits versus risks of scoliosis surgery.¹⁷ Specific spinal fusion methodology has evolved from the Harrington rod, Luque procedure, and pedicle screws to combinations of all three. Complications from any surgical method depend to a large degree on the severity of the curve and the respiratory and cardiac status of the patient.

Scapular winging can occur with any of the myopathies that present

with proximal weakness but is often severe in FSHD and some LGMDs. Scapulothoracic fixation in carefully selected patients (as illustrated by **Case 7-2**) can improve function by anchoring the scapula to the spine and providing stability that the weak serratus anterior, middle and lower trapezius muscles, and rhomboids cannot.¹⁸ Each patient needs to be evaluated thoroughly before considering surgery and each needs to weigh the benefits, required rehabilitation, and possible complications carefully before making a decision. Evidence suggests that such procedures may result in significant benefits (**Figure 7-5**) which must be considered in light of the need for postoperative immobilization, rehabilitation time, and complications such as stretching of the slings, loosening of the wires and screws, and, in some cases, nerve damage.¹⁹

Pes planus, pes cavus, pes equinovarus, and congenital hip dislocation all occur in congenital myopathies and in many dystrophies. Surgical treatment is dependent on the child's ambulation potential or level of pain. In boys with DMD who are no longer walking, reports are mixed regarding the advisability of surgical intervention to reduce or prevent foot deformities.

Cervical hyperextension as a fixed joint contracture is an uncommon but painful and debilitating complication that can develop in children with severe weakness. This contracture is seen most frequently in the congenital myopathies, in which the child may lack sufficient head control to keep the eyes focused horizontally. The children, almost invariably not walking, must routinely hyperextend their necks, and thus the cervical extensor muscles become restricted in length. Along with cervical weakness, truncal weakness may mean the children

Case 7-1

A 45-year-old woman with facioscapulohumeral dystrophy had slowly progressive weakness and increased difficulty with ambulation over a 15-year period. Initially, her major problem was ankle dorsiflexor weakness (3+ grade Medical Research Council [MRC] scale) and she walked with a steppage quality due to her foot drop. She did well with a prescribed pair of custom-molded plastic ankle-foot orthoses (AFOs) that provided dorsiflexion assist and helped stabilize her ankles. She walked much more naturally with these, but as her disease progressed she developed increasing quadriceps weakness, her knees tended to buckle, and she fell easily. Her AFOs had been set at 90 degrees and during heel strike actually increased the flexor moment at her knee joints and contributed to her instability. She began using a wheelchair and became essentially nonambulatory. After much consultation and discussion with a physical therapist and an orthotist experienced in muscular dystrophy, she was prescribed a set of "stance control" knee-ankle-foot orthoses (KAFOs) with hinged knee joints that locked during stance phase and released with plantar flexion during the push-off phase of her gait. Using these braces, she resumed walking, and her falls ceased (**Supplemental Digital Content 7-1**, links.lww.com/CONT/A115).

Comment. This case illustrates well the challenges and rewards of effective bracing for patients with chronic myopathies. Too often, bracing is considered from a "one size fits all" viewpoint so that if the initially prescribed device is not effective, either the patient or clinician (or both) give up on further efforts. Limitations in insurance also can affect the number and type of orthotic devices that can be tried. In this case, careful consideration of the biomechanics of the patient's gait led to a solution that had a profound effect on the patient's quality of life and ability to function.

assume a degree of trunk forward flexion, which also results in cervical hyperextension as the only means to view the world normally. A few reports have been published of successful surgical treatment for this problem.

Osteoporosis

Since the primary factor that determines bone density is muscle strength, virtually all patients with muscle weakness develop osteopenia or osteoporosis. This is particularly true in nonambulatory patients and those treated with corticosteroids. The problem is confounded in children because chronic steroid use will delay puberty, resulting in smaller (and potentially more likely to fracture) bones (**Figure 7-6**). If severe, osteoporosis

can lead to higher fracture risk, hypercalciuria, and kidney stones. Dual-emission x-ray absorptiometry (DEXA) is useful for diagnosing osteoporosis, with referral to an experienced internist or endocrinologist usually indicated. No regimen has been agreed upon for preventing osteoporosis in patients on corticosteroids, although calcium and vitamin D supplementation are recommended.¹ Bisphosphonates have also been used, but evidence remains insufficient to recommend bisphosphonates prophylactically to reduce fractures.²⁰ One retrospective study in DMD found that treatment with steroids and bisphosphonates was associated with significantly improved survival over steroid treatment alone.²¹

KEY POINT

■ Diagnosing and managing osteoporosis is critical in patients with chronic myopathies. Referral to an experienced pediatrician, internist, or endocrinologist is usually appropriate for severe cases.

FIGURE 7-4 Congenital muscular dystrophy in a 6-year-old girl. A, Thoracic lumbosacral orthosis (TLSO) does not prevent scoliosis but may decrease the rate of progression in order to “buy time.” B, C-shaped scoliosis without orthosis.

Case 7-2

A 28-year-old woman with facioscapulohumeral dystrophy was seen for concerns about midback and bilateral scapular pain. She worked full-time in a hospital, and her job responsibilities included reaching above her head. Her scapular stabilizer Medical Research Council (MRC) scores were not antigravity. She and her husband were shown active assistive range-of-motion exercises. She tried but did not tolerate a scapular retraction orthosis and was then referred to an orthopedic surgeon with experience in scapular fixation surgery. She underwent scapular fixation surgery using a bone graft from the pelvis on the right scapula, completed her rehabilitation, and returned to work with much improved function of the arm and less pain. Six months later she repeated the procedure on her left scapula. Two years after surgery, she reported much less pain on the McGill Pain Questionnaire, was still able to raise her arms above her head, and was a willing volunteer to discuss the surgery and rehab with other patients considering the procedure.

Comment. This case illustrates the importance of recognizing when surgical intervention may be the best solution for a particular patient. Conservative options were initially tried, and when they did not provide pain relief, the patient was judged to be an excellent surgical candidate. She was highly motivated and had an available caretaker during her convalescence as well as support from her employer. Her arm function and quality of life remained improved since her surgery.

Mobility Aids and Adaptive Equipment

A variety of wheelchairs (manual, powered, or power-assisted [Figure 7-7]), motorized scooters, walkers, bath and toilet aids, chair lifts, ceiling lifts, and stair lifts are available. Unfortunately, economic considerations and insurance coverage rather than true medical need are often the primary determinants for what equipment a patient can obtain. Occupational and physical therapists and physiatrists experienced in neuromuscular diseases and their rate of progression are essential as adaptive equipment consultants. Understanding the patient's insurance benefits is an unfortunate but necessary step in prescribing such equipment. Sometimes prescribing a wheeled walker, for example, can preclude a patient from receiving motorized mobility until a set period of time

FIGURE 7-5 A patient with facioscapulohumeral dystrophy before (*left*) and after (*right*) scapular fixation surgery.

has expired. In this regard, patient support groups and organizations (eg, the Muscular Dystrophy Association) often have access to loan closets and

FIGURE 7-6 Orthopedic effect of chronic corticosteroid therapy in Duchenne muscular dystrophy (DMD). A, X-ray of a 16-year-old boy without DMD, and B, a 16-year-old boy with DMD on chronic corticosteroid therapy. Delayed puberty secondary to chronic corticosteroid therapy results in smaller, narrower bones.

KEY POINTS

- Motorized mobility can be considered whenever the patient or his or her family find themselves altering plans or depriving themselves of activities because ambulation is too fatiguing or labored or becoming unsafe. A history of falling, no matter how infrequent, is a red flag.
- Both dangerous weight gain and weight loss are frequent complications of chronic myopathies.

equipment banks that can help patients obtain the necessary equipment even with limited resources.

On a practical basis, a motorized wheelchair or scooter can be considered whenever the patient or family find themselves altering plans or depriving themselves of activities because ambulation is too fatiguing or labored or becoming unsafe. Other factors for the patient to consider, however, are accessibility to enter and exit the home, accessibility within the home, and ability to transport a heavy (at least several hundred pounds) wheelchair or scooter.

Dietary Management: Complications of Obesity and Weight Loss

In any disorder causing chronic weakness, affected people become less

active, burn fewer calories, and all too often gain unacceptable amounts of weight unless their diet is modified. Most attention in this regard has focused on DMD, in which patients have reduced muscle but a higher fat mass proportion compared to the rest of the population. The use of corticosteroids in DMD has further exacerbated this issue. DMD patients on the recommended daily prednisone dose of 0.75 mg/kg demonstrate excessive weight gain compared to control groups.²² Deflazacort, another corticosteroid (not available in the United States), has a lesser effect on weight but still results in weight gain. A low-sodium, low-simple-sugar, and reduced caloric diet is indicated for any myopathy patient on chronic corticosteroid therapy. As in any diet, however, compliance is a frequent problem in helping patients maintain an ideal weight.

Myopathic patients with dysphagia are at risk for developing cachexia if their intake is inadequate to cover expended energy; sometimes even the work of breathing contributes to weight loss. Ventilatory assistance to reduce energy expenditure may be necessary, as well as supplemental caloric intake with nutritional supplements. This may entail insertion of a nasogastric tube or percutaneous endoscopic gastrostomy (PEG) for indefinite use, as the work of self-feeding or mastication becomes profoundly exhausting. Referral to a licensed dietitian is always helpful in managing these issues.

Swallowing Abnormalities and Treatment

Dysphagia is a prominent feature of many muscle disorders, including oculopharyngeal muscular dystrophy (OPMD), mitochondrial disorders, and IBM, among others. Swallowing problems can even be the presenting

FIGURE 7-7 Power-assist manual wheelchair. This device is lightweight and portable, yet batteries on each wheel allow some powered mobility.

manifestations in some patients. Impaired pharyngeal muscle function may result in pooling of secretions in the hypopharynx, choking, and aspiration. In other disorders such as FSHD, severe facial weakness may impair the ability to effectively manipulate food in the mouth or to chew efficiently. Tongue weakness can also contribute to dysphagia in some disorders. In other patients (eg, those with DM1), weakness of striated muscle in the upper third of the esophagus results in food “sticking” in the suprasternal region. Occasionally, swallowing problems are subclinical and not recognized by patients, who may present with unexplained cough or shortness of breath that occurs during or after meals; unexplained weight loss; or the need to take smaller bites. Patients with DM1 frequently deny swallowing problems even when they are present. Dysphagia has been associated with nutritional insufficiencies, aspiration pneumonia, atelectasis, decreased quality of life, and poor prognosis.

Despite the fact that it is a relatively common problem, no randomized controlled trials have evaluated the treatment of dysphagia in chronic muscle disease. Swallowing difficulties require multidisciplinary intervention including speech, occupational, and physical therapists; nutritionists; dentists; otolaryngologists; and gastroenterologists. Treatment options include dietary adjustments, exercises and the teaching of safe swallowing techniques (supervised by a speech/swallow therapist), surgical intervention, and enteral feeding. Published series are anecdotal or retrospective. A Cochrane report concluded that both cricopharyngeal myotomy and upper esophageal dilatation offer relief to people with dysphagia due to OPMD, but data are lacking in other disorders. Cricopharyngeal myotomy, dilatation, and botulinum

injection all lose effectiveness over time as symptoms progress.²³ Severe weight loss or recurrent aspiration may necessitate a pharyngostomy or operative or percutaneous gastrostomy to provide adequate nutrition via long-term enteral feeding.

Other Gastrointestinal Complications

Smooth muscle may be involved in several myopathies, including DM1, DM2, DMD, CMD, mitochondrial myopathies, and DMY with resultant gastrointestinal complications. Gall bladder dysfunction and a variety of esophageal and intestinal disorders are frequent in DM1 and DM2.^{24,25} Patients with DMD have impaired gastric motility associated with dystrophin deficiency of smooth muscle and may present with gastric dilatation and intestinal pseudo-obstruction that requires nasal-gastric tube suction, promotility agents, and IV fluids. Any patient with muscle disease and decreased mobility, low fluid intake, and weak abdominal muscles may develop constipation; those who also have hypomotility of the gastrointestinal tract require a regular bowel program.^{1,15,24}

Respiratory Management

Many muscle disorders lead to respiratory dysfunction as a result of diaphragmatic or intercostal muscle dysfunction; impaired glottis function with ineffective cough; upper airway dysfunction during sleep; kyphoscoliosis; chest wall stiffness; and sometimes (eg, in DM1) impaired CNS ventilator drive (Table 7-1²⁶). Generalized respiratory muscle weakness results in restrictive lung disease with reduced FVC, a decrease in total lung volume, and a decrease in lung compliance. In most cases, the earliest detectable abnormality is a reduction in maximum expiratory pressure (MEP) and maximum

KEY POINTS

- Dysphagia may present with unexplained cough or shortness of breath in addition to weight loss.
- Myopathy patients with decreased mobility, low fluid intake, and weak abdominal muscles may develop constipation.

KEY POINTS

- Often the earliest sign of respiratory muscle weakness is a reduction in maximum expiratory pressure and maximum inspiratory pressure.
- Patients with severe or rapidly progressive weakness must be monitored carefully for signs of respiratory compromise, even when asymptomatic.
- Dyspnea is an uncommon presenting symptom of chronic respiratory failure in the myopathies.

inspiratory pressure (MIP) (or negative inspiratory pressure). If the diaphragm is selectively involved before other muscles become weak (eg, late-onset Pompe disease), FVC is reduced to a much greater extent when the patient is supine, causing orthopnea.

With few exceptions, patients with a chronic myopathy should have an annual assessment of lung function beginning at the time of diagnosis. Patients with disorders at high risk of developing respiratory compromise or who have low initial FVC or rapidly progressive disease should be monitored more frequently. MEP and MIP are useful to detect early respiratory muscle involvement, and peak cough flow is valuable to monitor expiratory function. Forced expiratory volume in 1 second (FEV1), peak cough flow, or flow volume loop may be obtained depending on the specific information needed.²⁷ In patients with suspected respiratory failure, the vital capacity should be measured with the patient supine as well as seated. Oxygen saturation should be monitored in patients with respiratory signs or symptoms or if the vital capacity is less than 1.5 L.²⁶ In selected patients with symptoms or signs, arterial blood gas level determination may provide useful information, although this is not needed in the majority of patients.^{28,29} Noninvasive overnight monitoring of oxygenation with ear lobe or digit oximetry is often necessary. Evaluation of pulmonary function can also include (1) chest x-ray to assess heart size, diaphragmatic position, and evidence of atelectasis; (2) electrocardiography to search for signs of cor pulmonale (abnormal P waves, right axis deviation, right ventricular hypertrophy); (3) hematocrit to detect an elevation that may indicate chronic hypoxia; and (4) electrolyte determination to look for an elevation in bicarbonate which would

indicate metabolic compensation for chronic hypercapnia.

In myopathy patients, dyspnea is an uncommon presenting symptom of chronic respiratory failure. Rather, patients usually present insidiously with symptoms of nocturnal oxygen desaturation such as frequent nighttime arousals, morning headache, excessive daytime sleepiness, anxiety, depression, and lethargy. Generalized fatigue, unexplained weight loss, orthopnea, or cough are also suggestive of respiratory failure. Chronic hypoxia increases pulmonary vascular resistance, which may in turn lead to symptoms of cor pulmonale: ankle edema, nocturia, and orthopnea. Signs of respiratory compromise include tachypnea, tachycardia, and use of accessory respiratory muscles (the external intercostals, parasternal intercartilaginous muscles, sternalocleidomastoids, and scalene muscles) (Table 7-2).²⁹ Many patients, particularly those with DMD, develop glossopharyngeal breathing (“frog breathing”), in which rapid cycling of the tongue muscles forces air into the lungs. If the diaphragm is selectively weakened, a seemingly paradoxical breathing pattern occurs during which the patient’s abdomen flattens or moves inward during inspiration. Examination of the patient with respiratory insufficiency usually shows diminished breath sounds and atelectatic (pan-inspiratory) rales at lung bases, diminished diaphragmatic excursion as detected by percussion of the chest, and decreased cough effectiveness.

Prophylactic measures. The US Centers for Disease Control and Prevention recommends that essentially all patients with significant myopathic illness should receive one or more doses of the pneumococcal vaccine and annual vaccines for the pertinent influenza virus.³⁰ Respiratory muscle exercises may

TABLE 7-1 Respiratory Involvement in Myopathies^a

Frequency of Involvement	Myopathy
Always	Duchenne muscular dystrophy
Common	Acid maltase deficiency Limb-girdle muscular dystrophy (LGMD) 2C, 2D, 2F, 2I Nemaline myopathy Ullrich congenital muscular dystrophy Minicore myopathy
Occasional	LGMD 1B Becker muscular dystrophy Bethlem myopathy
Rare	Facioscapulohumeral dystrophy Mitochondrial myopathies Central core disease Oculopharyngeal dystrophy Various distal myopathies

^a Reprinted from Shahrizaila T, Kinnear W. *Neuromuscul Disord*.²⁶ © 2007, with permission from Elsevier. www.sciencedirect.com/science/article/pii/S096089660600544X.

be considered, although such exercises are not recommended in DMD since they may result in contraction-induced injury. Inspiratory exercises with an incentive spirometer should emphasize improving chest wall elasticity by gentle range-of-motion exercises. Patients should be given a prescription for antibiotics to keep at home and advised to begin taking these when they develop a respiratory infection. Adequate hydration must also be maintained to assure easy mobilization of secretions. Weight control limits the effect of obesity on breathing. In myopathies with bulbar involvement, inability to clear airway secretions can impair breathing. Anticholinergic medication, botulinum toxin injections, and surgical interventions are potential treatments for sialorrhea.

Sleep-related breathing complications. Sleep increases the susceptibility to respiratory difficulties.^{28,31} If a patient with a muscle disease has symptoms of respiratory impairment

during the day, it is likely that he or she is also having difficulties at night. Frequent nocturnal arousals, daytime sleepiness, weight loss, morning headaches, and snoring are common symptoms of sleep-disordered breathing. The etiology of sleep-disordered breathing is multifactorial and depends on the specific myopathy but is common in patients with diaphragmatic weakness, obesity, or scoliosis. In DM1, sleep apnea can be both central and obstructive. Further investigation with nighttime oximetry or a polysomnogram is recommended when the patient has symptoms of nocturnal hypoventilation; the FVC is less than 1 L to 1.5 L or FEV1 less than 40%; unexplained gas exchange abnormalities (ie, with FEV1 greater than 40% predicted or FVC greater than 50% predicted) are present; or the patient demonstrates daytime desaturation or carbon dioxide retention.³¹

Acute respiratory failure. When FVC falls below 50% of normal in

TABLE 7-2 Symptoms and Signs of Respiratory Impairment in Muscle Disease^a**► Symptoms**

Constitutional

Generalized fatigue

Weakness

Cardiopulmonary

Dyspnea

Leg edema

Orthopnea

Secretion retention

CNS

Early morning headaches

Daytime hypersomnolence

Irritability and mood disturbances

Hallucinations and other psychiatric disorders

Sleep

Restless sleep

Nightmares

Enuresis

Frequent arousals

► Signs

Vital signs

Tachycardia and tachypnea

Respiratory

Accessory chest wall muscle and abdominal muscle use

Paradoxical breathing pattern

Diminished diaphragm excursion

Cardiac

Distended neck veins

Edema

Cyanosis

^a Reprinted with permission from Perrin C, et al, *Muscle Nerve*.²⁹ © 2003 Wiley Periodicals, Inc. onlinelibrary.wiley.com/doi/10.1002/mus.10487/abstract.

and the management of both acute and chronic respiratory failure. These discussions should always take place in concert with a pulmonologist or respiratory therapist expert in managing patients with neuromuscular disorders, and the opinions and wishes of patients and their families should be the prime consideration. If the topics of a living will and durable power of attorney for health care have not yet been addressed, they should be discussed.^{32,33}

Acute respiratory failure in myopathy patients is usually precipitated by an acute infection or other disease (most often atelectasis) and is seldom due to muscle weakness alone. For dyspneic or obtunded patients, emergency evaluation for endotracheal intubation may be necessary. If hypoxia alone is present and the patient is alert, oxygen at low flow rates (less than 1 L/min) may correct hypoxia, although such therapy must be administered with caution since patients with neuromuscular disease are often hypercapnic, and oxygen often depresses ventilatory drive and exacerbates hypoventilation.²⁸ Noninvasive positive pressure ventilation (NPPV) is usually tried initially if control of secretions and cough assistance is successful.³⁴ Whether intubation or NPPV is utilized, patients with some myopathies can be weaned after the precipitating respiratory crisis; others may require ventilation assistance indefinitely. Pulmonary consultation is always indicated in these patients.

Chronic respiratory failure. A gradual deterioration in respiratory function with symptoms or signs of hypoxia should prompt consideration of elective respiratory support. Optimally, patients and their families will have been made aware of the risk of respiratory failure and options for support in advance of an acute deterioration.^{32,33} Expert

patients with myopathic diseases, discussions should be initiated about the possible need for ventilator assistance

consensus documents recommend NPPV over tracheostomy for chronic ventilatory failure due to myopathies and other restrictive respiratory diseases.^{1,27} The institution of NPPV should be managed only by health care providers with experience in this modality. Different interfaces—including nasal mask, nasal pillows, full-face masks, or mouthpieces—should be used to maximize patient comfort. NPPV should be started at low pressures to maximize the patient's tolerance to the device. Generally, the inspiratory pressure (IPAP) is set at 6 cm to 8 cm water and the expiratory pressure (EPAP) at 3 cm to 5 cm water. The IPAP can then be titrated in 1 cm to 2 cm increments based on patient tolerance and clinical status. The device should be placed on a spontaneous timed mode with a backup rate set at the patient's nocturnal respiratory rate (generally 8 breaths/min to 10 breaths/min). Frequent follow-up by a trained respiratory therapist (at least weekly) is crucial to deal with any problems and improve patient tolerance. A heated humidifier can be prescribed if the patient develops symptoms of nasal dryness or congestion.

Although NPPV is usually effective in managing hypoventilation, patients with difficulty clearing secretions or a weak cough (ie, when peak cough expiratory flow [PCEF] is less than 270 L/min) may require assistance with a portable mechanical insufflator-exsufflator or cough assist device.^{33,35} These devices allow delivery of pressurized air (30 cm to 50 cm water) followed by an immediate forced exsufflation of negative pressure (30 cm to 50 cm water) for several seconds via a mouthpiece or naso-oral mask. The use of mechanical insufflation-exsufflation safely allows patients with respiratory tract infections and profuse mucus production to clear their secretions and permit the ongoing use of NPPV. The insufflation and exsufflation pressures

should be set to the maximum comfortable level tolerated by each patient in order to eliminate airway secretions. In general, inspiratory pressures are set at 20 cm to 40 cm water and expiratory pressures at 5 cm to 20 cm water. Another device useful for helping patients clear secretions is the “chest vest,” which rapidly inflates and deflates in a vibratory manner, resulting in up to 25 chest wall compressions/s. This helps dislodge mucus from the bronchial walls for removal via cough or a cough assist device.

NPPV is contraindicated in patients with copious secretions, cognitive impairment, or lack of caregiver support.^{29,36} In these instances, ventilation may be more easily managed with tracheostomy and positive-pressure ventilation. Initiation of chronic respiratory support poses major practical, financial, medical, and ethical responsibilities for patients and their families, and it is often helpful for patients to discuss the procedure with others receiving such support. The input of experienced pulmonary consultants and palliative medicine services can be invaluable in these instances.

SYMPTOMATIC THERAPY

Many myopathies are associated with symptoms and signs that are not related to skeletal muscle involvement but require management. Most notable are the myotonic dystrophies and the mitochondrial encephalomyopathies, which are multisystem disorders affecting a number of organ systems. A detailed review of the complications in these disorders is beyond the scope of this review, and many of these topics are covered elsewhere in this issue of **CONTINUUM**. Table 7-3 lists the variety of symptoms that mitochondrial patients may present to the practitioner.^{37,38} Table 7-4 shows the comparative symptoms of DM1 and DM2

KEY POINTS

- At-risk family members of patients with genetic myopathies that may involve the heart need to be screened for cardiac involvement.
- Symptoms of congestive heart failure in myopathy patients may be quite nonspecific and include insomnia, cough, poor appetite, abdominal pain, nausea, or ankle edema.

and highlights the multiple system involvement of these disorders.²⁵

A few symptoms unrelated to weakness merit mention in this review, however. Daytime sleepiness in DM1 is quite common and affects all aspects of the patient's life. Although modafinil is sometimes prescribed for these patients (100 or 200 mg/d), a Cochrane review found insufficient evidence to outweigh potential side effects.³⁹ Glucose intolerance⁴⁰ and higher rates of some types of cancer have also been reported in DM1 and DM2,⁴¹ but by far the most important nonskeletal muscle manifestation is cardiac involvement that is prominent in numerous myopathies.

Cardiac Management

Cardiac involvement occurs in many myopathies (Table 7-5).⁴² Although it is often asymptomatic, detailed studies (eg, ECG, echocardiogram, Holter monitoring, and cardiac magnetic resonance [CMR]) frequently reveal subclinical cardiac involvement in these disorders.⁴³ Dilated cardiomyopathy, conduction defects, and arrhythmias are the most common abnormalities found, but a few disorders (eg, desmin myopathy, nemaline myopathy) may display hypertrophic cardiomyopathy. Not infrequently, patients demonstrate combinations of more than one type of cardiomyopathy, and certain myopathies may present with cardiac manifestations. Because of the complexity in management of cardiac issues, all patients with these disorders should be referred at the time of diagnosis for cardiac evaluation and follow-up. This is particularly crucial since adult myopathy patients are also susceptible to coronary artery disease, hypertension, and other cardiovascular disorders found in the general population. Another important aspect of cardiac management is that family members at high risk of developing the muscle

disorder (eg, offspring of DM1 patients) need to be screened for cardiac problems even if the muscle disease has not yet been diagnosed.

Common symptoms of cardiomyopathy include insomnia, cough, poor appetite, abdominal pain, nausea, and ankle swelling. Fatigue, orthopnea, and dyspnea seldom occur in early heart failure, and exercise intolerance is rarely a concern because myopathic patients are often relatively inactive. Patients with arrhythmias may report dizziness or syncope. Congestive heart failure develops insidiously; early signs are cough, pedal edema, and late inspiratory rales. Non-cardiac-dependent edema is frequent in patients who use wheelchairs, but its presence should still prompt consideration of heart failure, especially in the young. More florid signs of congestive heart failure include tachycardia, right or left ventricular enlargement, gallop rhythm, alternating pulse, hepatomegaly, and neck vein distention.

Laboratory evaluation. The ECG is indispensable for detecting early cardiac abnormalities in at-risk patients. The presence of first-degree heart block or other significant change should prompt referral to a cardiologist. Echocardiography evaluates cardiac contractility and can suggest a specific myopathy.⁴⁴ Because technical problems can interfere with the reproducibility and sensitivity of echocardiograms, especially in obese and scoliotic patients, CMR imaging is increasingly being used, especially in dystrophy patients. The cardiac isoform troponin I (cTnI) is the most informative biomarker to detect cardiac ischemia in IBM.⁴⁵

Current guidelines recommend ECGs and echocardiograms at time of diagnosis or by 6 years of age, repeated every 2 years until the age of 10 and then performed annually for patients

TABLE 7-3 Nonmuscle Involvement in Mitochondrial Disease Requiring Symptomatic Care^a

- ▶ **Constitutional**
 - Failure to thrive
 - Short stature
 - Fatigue
- ▶ **Neurologic**
 - Headaches
 - Dementia
 - Hypotonia
 - Seizures
 - Myoclonus and other movement disorders
 - Sensorineural hearing loss
 - Axonal neuropathy
 - Cerebellar dysfunction
 - Strokelike episodes
- ▶ **Cardiovascular**
 - Cardiac conduction block and arrhythmias
 - Cardiomyopathy
- ▶ **Ophthalmologic**
 - Retinal pigmentary changes
 - Optic nerve hypoplasia
 - Ophthalmoplegia/ophthalmoparesis
- ▶ **Gastrointestinal**
 - Hepatic dysfunction
 - Dysmotility with constipation
- ▶ **Pancreatitis**
 - Diarrhea
- ▶ **Dermatologic**
 - Lipomatosis
 - Cushingoid appearance
- ▶ **Endocrine**
 - Hypothyroidism
 - Hypoparathyroidism
 - Growth hormone deficiency
- ▶ **Renal**
 - Nephrotic syndrome
 - Renal tubular acidosis

^a Reprinted from Haas RH, et al, *Pediatrics*.³⁸
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TABLE 7-4 Nonmuscle Involvement in Myotonic Dystrophies Requiring Symptomatic Care

- ▶ **Ocular/Ophthalmic**
 - Ptosis
 - Cataracts
 - Retinal degeneration
- ▶ **Endocrine**
 - Insulin resistance/glucose intolerance
 - Hypogonadism/testicular atrophy/oligospermia
 - Reduced growth hormone
 - Reduced insulinlike growth factor-1
- ▶ **Cardiac**
 - Conduction defects and arrhythmias
 - Cardiomyopathy (usually in advanced cases)
- ▶ **Gastrointestinal**
 - Dysphagia (pharyngeal or esophageal)
 - Constipation/pseudo-obstruction
 - Cholelithiasis
- ▶ **Skeletal**
 - Frontal bossing
 - High arched palate
 - Temporomandibular joint dislocation
- ▶ **Dermatologic**
 - Epithelioma
 - Pilomatrixomas
- ▶ **Respiratory**
 - Chronic hypoventilation with secondary hypercapnia
 - Reduced response to hypoxia
- ▶ **Nocturnal/Sleep Disorders**
 - Obstructive sleep apnea; sometimes with central component
 - Periodic breathing

KEY POINT

■ All patients with a myopathy that is associated with cardiomyopathy should be followed by a cardiologist or internist experienced in neuromuscular disease.

TABLE 7-5 Myopathies Associated With Cardiac Involvement

► **Presenting Primarily as a Cardiomyopathy**

Dystrophies

Duchenne and Becker muscular dystrophies
 Emery-Dreifuss syndrome (late)
 Some limb-girdle dystrophies (especially types 1B, 2C-F, and 2I)
 Myotonic dystrophies (usually late)

Metabolic disorders

Infantile acid maltase deficiency
 Disorders of lipid metabolism
 Debranching enzyme deficiency
 Amyloidosis
 Alcoholic cardiomyopathy
 Endocrine myopathies

Other disorders

Nemaline myopathies
 Mitochondrial cytopathies
 Myofibrillar myopathies

► **Presenting Primarily With Conduction System Disease/Arrhythmias**

Myotonic dystrophies
 Emery-Dreifuss syndrome
 Mitochondrial cytopathies
 Myofibrillar myopathies
 Periodic paralyses (especially Andersen-Tawil syndrome)

with DMD and at-risk LGMD and CMD.⁴³ Carriers of X-linked muscle disorders (DMD, BMD, EDMD) are also at risk for cardiomyopathy. Current recommendations for these patients call for echocardiograms at least every 5 years beginning by 16 years of age.⁴⁴ Patients with myopathies

associated with conduction defects (eg, DM1 and DM2) or arrhythmias (EDMD, some LGMDs) should have annual ECGs and Holter monitoring with referral for more detailed studies, including intracardiac electrophysiology studies. Appropriate patients should be considered for a pacemaker or implanted cardioverter-defibrillator.⁴⁶ Dilated cardiomyopathy and conduction defects can also occur in DM2 but are less common.⁴⁷ For the inflammatory myopathies, an ECG is recommended upon the diagnosis of DM or PM but is not necessary in IBM patients without cardiac symptomatology, with cardiology referral and additional investigations performed for any significant abnormalities.⁴⁵

Treatment of all cardiac abnormalities in patients with myopathy is best directed by a cardiologist or internist familiar with neuromuscular disorders. Angiotensin-converting enzyme (ACE) inhibitors are recommended in DMD and BMD at the first indication of cardiac abnormality, with the role of beta-blockers under investigation.⁴⁸ Patients treated with corticosteroids have added complications that require management, including weight gain, increased blood pressure, and fluid retention. Several studies have shown substantial beneficial effects of steroids on the heart in DMD.^{49,50} Heart transplants are being performed for patients with end-stage myopathy; one retrospective report concluded that clinical outcomes were similar to those observed in patients without muscular dystrophy.⁵¹

Edema

Dependent foot, ankle, and leg edema are frequent in myopathy patients who use a wheelchair or who lack good venous return due to inadequate muscle-pumping function of leg musculature; congestive heart failure can

also contribute to edema. Edema exacerbates weakness due to the resulting extra weight as well as the mechanical limitations it imposes on leg joints. It also predisposes an affected person to skin infection and ulceration. Edema can be limited by elevating the legs periodically (with elevating leg rests on a wheelchair or by a tilt-in-space option on a motorized chair) and by reducing dietary sodium. Pressure stockings can help reduce edema if they are properly fitted and tolerated by the patient.

Pain

Multiple factors predispose patients with myopathy to developing pain, including prolonged immobility, unequal force generation across joints, a tendency to fall, and sometimes the pathologic processes themselves (Figure 7-8). Approximately 80% of patients with neuromuscular disorders experience pain, and 40% describe their pain as severe.⁵² Not surprisingly, pain and fatigue are associated with psychosocial factors and quality of life.^{52–54} Clinicians should inquire at every visit whether their patient has pain; including a short-form McGill Pain Questionnaire or other standardized pain assessment encourages an open dialogue on this issue. Electronic medical records allow easier compilation of longitudinal and cross-sectional pain responses that can guide the practitioner toward potential treatment. Although good evidence on the treatment of pain in myopathy patients is sparse, modalities used in other pain conditions such as moist heat, ice, massage, mobility intervention (eg, gentle range-of-motion exercise, aquatic therapy, yoga, tai chi) and medicinal interventions (eg, non-steroidal anti-inflammatory drugs, gabapentin, duloxetine, pregabalin, amitriptyline) can be utilized. A large

randomized controlled study did demonstrate that mexiletine reduced pain (and myotonia) in patients with nondystrophic myotonia.⁵⁵

PSYCHOLOGICAL SUPPORT

Disease severity, illness perception, and mood are all linked to the reported quality of life of people with muscle disease.⁵⁶ Every clinic visit should therefore include questions concerning the patient's mood and general mental outlook, as well as those of his or her caregivers and family. Providing psychological support must include an assessment of social and practical limitations caused by the patient's muscle disease and their effects on his or her quality of life. Inevitably, these patients encounter economic, employment, and personal issues as a result of their myopathy. Prompt institution of treatment with pharmacologic agents for depression and anxiety disorders and referral to a mental health professional is indicated in these patients and can often have a profound impact on the patient's quality of life.

Another crucial aspect to the psychological management of patients with myopathy—especially for those with chronic, progressive, and untreatable disorders—relates to patients' need to be aware of research endeavors for "their" disease. Because of Internet access and lay support groups, patients today are informed consumers who know more about their disease than ever before. They want to know about current advances and ongoing therapeutic trials in their disease, the role of alternative therapies, and a host of other issues and questions. The clinician who is uninformed or, worse, uninterested in the genetics and pathogenesis of the disease and current advances cannot hope to maintain a viable therapeutic

KEY POINT

- A majority of patients with myopathy experience pain, and many describe their pain as severe. Clinicians should inquire at every visit whether their patient has pain.

FIGURE 7-8 Dystrophies causing chronic pain. *A*, A patient with limb-girdle muscular dystrophy 2D with muscle weakness leading to severe postural abnormality and causing chronic pain. *B*, A patient with facioscapulohumeral dystrophy with muscle weakness leading to severe postural abnormality and causing chronic pain.

relationship. The clinician must therefore always be ready to provide solid information about a patient's condition by keeping up with current genetic discoveries and research advances related to a given patient's disorder. Supporting and facilitating networking opportunities for patients is also helpful in this regard. This may involve something as simple as providing patients with information on lay support groups or informative websites or putting individual patients in contact with each other (with each patient's permission). Patients should be encouraged to participate in clinical trials if feasible and practical. If no studies are available for their disease, it is important to help patients and their families understand that advances in other diseases (eg, gene therapy trials in DMD) may one day be applicable to their disorder.

On a practical basis, perhaps the most important component to providing psychological support to patients with an incurable muscle disorder involves letting them know that they have a team of dedicated health care professionals encompassing physical, respiratory, occupational, and speech therapists as well as orthotists, nutritionists, psychologists, genetic counselors, specialists, and social workers all committed to fighting their disease, optimizing their function, and improving their quality of life.

VIDEO LEGEND

Supplemental Digital Content 7-1

Reambulation using stance control knee-ankle-foot orthoses. Video shows a 45-year-old woman with facioscapulohumeral dystrophy with slowly progressive weakness and increased difficulty with ambulation and falls over a 15-year period. Initially, the patient ambulated well

with custom-molded plastic ankle-foot orthoses, but as her disease progressed, she began using a wheelchair and became essentially nonambulatory. After consultation and discussion with a physical therapist and an orthotist experienced in muscular dystrophy, she was prescribed a set of stance control knee-ankle-foot orthoses with hinged knee joints that locked during stance phase and released with plantar flexion during the push-off phase of her gait. Using these braces, she resumed walking, and her falls ceased.

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