Neuromuscular diseases include disorders of the motor neuron (anterior horn cells and peripheral nerves), neuromuscular junction, and muscle. Muscular dystrophy (MD) and spinal muscular atrophy (SMA) are two prevalent, progressive neuromuscular diseases that require physical therapy. Progressive weakness, muscle atrophy, contracture, deformity, and progressive disability characterize both diseases. No cure is available for either disease. “Incurable,” however, is not synonymous with “untreatable,” and the physical therapist can be influential in prevention of complications, preservation of function, and issues concerning quality of life.

The objective of this chapter is to present an overview of the childhood forms of MD and SMA, including the role of the physical therapist as a member of the management team. The clinical presentation of the diseases is reviewed, and examination procedures are presented to assist the clinician in identifying impairments, functional limitations, and disabilities associated with MD and SMA. Guidelines for physical therapy management are also outlined based on my clinical experience and review of related literature.
Examples of status change include the period before the loss of walking, before the need for architectural modifications to accommodate adaptive equipment for mobility, during transition from the educational to the vocational/avocational environment, or during the terminal stages of the disease when the decision to use mechanical ventilation will be a major issue for the family.

Providing information to family members, persons with MD or SMA, and other members of the team regarding physical limitations and expected participation restrictions is an important role for the physical therapist. Many resource materials are available online through the national Muscular Dystrophy Association (MDA) or through state chapter MDA offices.

**PHYSICAL THERAPY EXAMINATION AND EVALUATION**

Although the progression of MD and SMA is relatively well known, the clinician must carefully observe the child for changes that require intervention modifications. As stated by Thomas McCrae (1870–1935), “More is missed by not looking than by not knowing” (Siegel, 1986). Ongoing dialogue with families is invaluable in identifying family-centered goals and the need for program modification.

The physical therapy examination is the initial step in management of the child with MD or SMA and should include those components identified in the *Guide to Physical Therapist Practice* (American Physical Therapy Association [APTA], 2001). Specifically, the following must be carefully examined:

1. History with family concerns
2. Aerobic capacity and endurance
3. Assistive and adaptive devices
4. Community and work (job/school/play) integration
5. Environmental, home, and job/school/play barriers
6. Gait, locomotion, and balance
7. Integumentary status (when using orthoses, adaptive equipment, or wheelchair)
8. Muscle performance
9. Neuromotor development
10. Orthotic, protective, and supportive devices
11. Posture
12. Range of motion
13. Self-care and home management
14. Ventilation/respiration

Systematic documentation of disease progression is essential in timing of interventions during transitions from one functional status to another or during times of increased family need.

**MUSCULAR DYSTROPHY**

The etiology of MD is genetic inheritance. The pathophysiology underlying the disease is progressive loss of muscle contractility caused by the destruction of myofibrils. The specific cellular mechanism behind the destruction in Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) has been partially identified and is discussed later in the chapter. The rate of progression of myofibril destruction is variable among the various forms of MD, giving evidence for the possibility of more than one cellular mechanism in the destructive process.

The diagnosis of MD is confirmed by clinical examination and laboratory procedures, including electromyography, muscle biopsy, DNA analysis, and selected enzyme levels assayed from blood samples (Jones & North, 1997; Siegel, 1986). The criteria for classification of the various forms of MD include the mode of inheritance, age at onset, rate of progression, localization of involvement, muscle morphologic changes, and presence of a genetic marker if available. The MDA recognizes nine primary classifications of MD (Muscular Dystrophy Association, 2001). Table 15-1 lists the six most prevalent types that exhibit initial clinical signs in infancy, childhood, or adolescence. Emery-Dreifuss MD (humeroperoneal) is very rare and is discussed only briefly. Limb-girdle MD may exhibit signs in the teenage years, but the onset of symptoms is more typically in early adulthood, and therefore, along with the adult-onset forms of MD, it is not discussed in this chapter. The reader should refer to texts on neuromuscular diseases by Brooke (1986), Siegel (1986), and Harper (1989) for further information on clinical presentation and general management of MD.

The primary impairment in MD is insidious weakness secondary to progressive loss of myofibrils. In the case of the congenital forms of MD the weakness is pronounced at birth and easily recognizable. In DMD, the weakness becomes evident by age 3 to 5 years. In congenital and congenital myotonic MD, contractures present at birth also cause primary impairment. The incidence of mental retardation is highest in congenital myotonic MD, but it is less frequently reported in DMD or the other childhood forms.

Secondary impairments in all forms of MD include the development of contractures and postural malalignment. Postural malalignment is seen in antigravity positions of sitting and standing and often includes development of scoliosis. Other secondary impairments include decreased...
respiratory capacity, easy fatigability, and occasionally obesity. Although significant intellectual impairment is not usual, IQ commonly averages 85, and consequently, 30% of boys with DMD have an IQ below 70 (Anderson et al., 2002). This finding has been related to a loss of dystrophin in the brain, and more specifically to a disruption in GABA receptors in the central nervous system (Anderson et al., 2002).

With the progression of muscle weakness, increasing caregiver assistance is required for persons with MD to carry out activities of daily living (ADL). Progressive disability is a hallmark of MD and requires multidisciplinary team management to maximize participation through the use of adaptive equipment and environmental adaptations.

Physical management in the treatment of MD is a key intervention because no drug or other therapy has been found to be curative (Brooke, 1986; Merlini et al., 2003). Physical therapy has been used to prolong the child’s independence, slow the progression of complications, and improve the quality of life.

**Dystrophin-Associated Proteins and Muscular Dystrophy**

Within the past 15 years significant advances have been made in the molecular genetics and biology of the muscular dystrophies. These advances have followed the identification of the genetic defect behind DMD and the missing protein dystrophin. Many other proteins that are associated with dystrophin have been found to be defective or missing in other forms of MD. The proteins are termed *dystrophin-associated proteins* (DAPs) (Blake et al., 2002). The DAPs form a complex of extracellular, transmembrane, and intracellular proteins, which are represented in Figure 15-1.

Dystrophin acts as an anchor in the intracellular lattice to enhance tensile strength. The other proteins are thought to act as a physical pathway for transmembrane signaling. Absence of any transmembrane protein, however, would result in faulty mechanics of the cell membrane. For example, sarcoglycan defects are present in adult forms of MD (Emery, 2002).

**Duchenne Muscular Dystrophy**

DMD is the most common X-linked disorder known, with an incidence of about 1 in 3500 live male births (Muscular Dystrophy Association, 2001). The prevalence of DMD in the general population is reported at about 3 cases per 100,000 (Emery, 1993; Muscular Dystrophy Association, 2001). Longevity is variable, from the late teens to early twenties up to the end of the third decade, depending on the rate of disease progression, presence of complications, and aggressiveness of respiratory care, including the use of assisted ventilation (Curran & Colbert, 1989).

Kunkel and associates (1985) identified the gene on the X chromosome (Xp21) that, when missing or defective, causes DMD and BMD, and Hoffman and associates (1987) then identified the protein (dystrophin) of the chromosome locus. Cloning of the dystrophin gene was the next major accomplishment, which provided a mechanism for prenatal or postnatal diagnosis and development of gene therapy (Koenig et al., 1987).

The etiology of muscle cell destruction in DMD and BMD is due to abnormal or missing dystrophin and its effect at the muscle cell membrane. Mechanical weakening
of the sarcolemma, inappropriate calcium influx, aberrant signaling, increased oxidative stress, and recurrent muscle ischemia are all hypothesized as mechanisms associated with myofibril damage (Petrof, 2002).

The focus of research for the treatment of DMD involves myoblast transfer therapy, gene and cell-based replacement therapy, attempts to increase production of other dystrophin-related sarcolemmal proteins (utropin), and use of drugs such as steroids. All therapies are in the experimental stage; myoblast transfer therapy is currently being evaluated in humans, the first human gene therapy trials for DMD using dystrophin are just beginning (Romero et al., 2002), but no trials have begun in humans on the use of utropin (Perkins & Davies, 2002). Evidence for the use of steroids and creatine is growing.

In myoblast transfer therapy the embryologic precursor cells of skeletal muscle (i.e., myoblasts) are obtained from a histocompatible donor. The cells are then injected into the muscle of the individual with DMD with the hope that the normal myoblasts will grow, mutate with the surrounding cells that are lacking dystrophin, and result in development of dystrophin. The results of the research in animals have shown that donor myoblast cells fuse with the dystrophic cells to form a hybrid multinucleated cell that produces dystrophin (Partridge et al., 1978). Myoblast transfer therapy is currently controversial, with the research by Law and colleagues (1997) supporting its efficacy, but others have reported disappointing results (Gussoni et al., 1997; Karpati et al., 1992; Partridge, 2002) and lack of substantiation for the findings of Law and associates.

Gene therapy research involves the introduction of the dystrophin gene that is packaged in a modified adenovirus or retrovirus called a vector. Lee and colleagues (1991) produced an entire dystrophin gene, and more recently Amalfitano and associates (1998) developed the vector for delivering the gene. Minigenes, found to be effective in animal models of MD, have also been developed (Biggar et al., 2002). Human trials using gene therapy in limb-girdle MD (Stedman et al., 2000) and DMD/Becker MD are in progress (Romero et al., 2002).

Utropin is a muscle protein that has molecular similarity to dystrophin. Utropin levels in the muscle are high in the fetus and newborn but gradually diminish, until utropin is primarily found at the neuromuscular or musculotendinous junction in adults Courdier-Fruh and colleagues (2002) have demonstrated in vitro that utropin levels can be increased through upregulation. It is hypothesized that utropin might act as a substitute for abnormal or missing dystrophin.

Medical management of DMD has also included clinical trials of various drugs. Long-term steroid use (prednisone, deflazacort, and oxandrolone) has been shown to improve outcomes, including prolonged independent and assisted walking by up to 3 years, improved isometric muscle strength by 60% in the arms and 85% in the legs when compared to untreated control subjects, as
well as improved pulmonary function (Biggar et al., 2001; Merlini et al., 2003; Wong & Christopher, 2002). Reported side effects, however, include weight gain, particularly with prednisone, growth suppression, and osteoporosis. Strict dietary controls to offset the side effects are recommended (Wong & Christopher, 2002). The use of creatine in a randomized double-blind study of 15 boys with DMD has also demonstrated improved muscle strength and endurance and a reduction in joint stiffness (Louis et al., 2003). Louis and colleagues also reported improved bone mineral density in boys who were wheelchair users, suggesting that negative effect of steroids on bone mineral density might be offset with the use of creatine.

Although it is commonly agreed that the prevention of contractures and the preservation of independent mobility are primary goals of a physical management program (Vignos, 1983), the prolongation of ambulation through surgery or orthotics remains controversial. Some authors promote the use of surgery and lightweight bracing (Bach & McKeon, 1991; Bakker et al., 2000; Heckmatt et al., 1985; Hsu, 1995; Miller & Dunn, 1982; Taktak & Bowker, 1995); others express skepticism about prolonging the inevitable in a progressive disease when the financial and emotional costs to the family may be very high (Gardner-Medwin, 1979). A decreasing trend in the use of knee-ankle-foot orthoses (KAFOs) from 69% of the MDA clinics surveyed in 1989 to 27% in 2000 has been reported (Bach & Chaudhry, 2000) and would suggest a trend of less aggressive orthotic management in this population.

Surgical management has focused on the control of lower extremity contractures, use of orthoses in conjunction with surgery to prolong ambulation, and spinal stabilization for control of scoliosis. Achilles tendon lengthenings and fasciotomies of the tensor fasciae latae and iliotibial bands are two procedures commonly reported to be used in conjunction with orthotics and physical therapy to prolong ambulation (Bach & McKeon, 1991; Hsu, 1995). Posterior tibialis transfer into the third cuneiform to reverse equinovarus deformity has also been reported (Hsu, 1995). Surgical management of scoliosis typically includes the use of spinal instrumentation with Luque rods (Marchesi et al., 1997). Conservative management of scoliosis using orthoses remains prevalent (Bach, 2000), however, 85% of boys with DMD develop severe scoliosis (Rideau et al., 1984) and orthotic management has not been shown to stop the progression of the curve (Heller et al., 1997).

**Impairments, Activity Limitations, and Participation Restrictions**

Examination of the 4- to 5-year-old child demonstrates the onset of classical clinical features of DMD and the primary impairment of muscle weakness. The posterior calf is usually enlarged as a result of fatty and connective tissue infiltration, which corresponds to the term pseudohypertrophic MD that is used for the eponym DMD. The pseudohypertrophy can occasionally be seen to affect the deltoid, quadriceps, or forearm extensor muscle groups. Initial weakness of the neck flexor, abdominal, interascapular, and hip extensor musculature can be noted with a more generalized distribution with progression of the disease. Figure 15-2 demonstrates the trends of muscle strength decline up to age 16 years from a study by Brooke (1986). These data were obtained in a multiclinic study of 150 children with DMD over a follow-up period of 3 to 4 years. The data represent approximately 15 data points per boy as recorded during follow-up visits. Similar findings on anthropometric data, range of motion, spinal deformity, pulmonary function, and functional skills were reported by McDonald and colleagues (1995) in a cohort of 162 boys with DMD followed over a 3-year period.

Muscle strength can be documented using manual muscle testing (MMT), which has been reported to have acceptable intrarater reliability (Florence et al., 1992), although it is not as accurate as using specialized devices such as a dynamometer. Instruments such as a handheld dynamometer (Stuberg & Metcalf, 1988) or strain gauge devices (Brussock et al., 1992) can be used to obtain objective strength recordings in the older child to assist in prediction of disability, such as the loss of independent ambulation.

No limitations in range of motion (ROM) are typically noted before 5 years of age in DMD. Mild tightness of the gastrocnemius-soleus and tensor fasciae latae muscles usually occurs first. The normal lordotic standing posture is increased, and mild winging of the scapulae is then seen as a compensation to keep the center of mass behind the hip joint to promote standing stability (Fig. 15-3). Scoliosis typically develops just before or during adolescence.

**Infancy to Preschool-Age Period**

No significant impairments, activity limitations, or participation restrictions are typically seen in the infant or toddler with DMD, although Gardner-Medwin (1980) reported that half of the children fail to walk until 18 months of age. Delay in walking, however, rarely leads to the diagnosis of DMD. Symptoms are seldom noted before age 3 to 5 years, unless there is a positive family history and caregivers are looking for early signs. The mean age at diagnosis is usually reported to be around 5 years (Miller & Dunn, 1982).

Although there is no significant disability in early childhood, many disability-related issues must be addressed. The family will have questions regarding peer interaction,
**Figure 15-2** Muscle strength 50th percentile lines plotted against age of 150 children with Duchenne muscular dystrophy. (Redrawn from Brooke, MH. A Clinician’s View of Neuromuscular Diseases, 2nd ed. Baltimore: Williams & Wilkins, 1986.)

**Figure 15-3** Typical standing posture of a 7-year-old boy with Duchenne muscular dystrophy. **A**, Posterior view; note the winging of the scapula, equinus contracture of the left ankle, and calf pseudohypertrophy. **B**, Lateral view; note increased lumbar lordosis.
routine activity level for the child, and the prognosis. The therapist must be aware of each family’s coping response, goals, and needed supports to provide family-centered care. This is the appropriate time to discuss with the family the social aspects of the disability and to answer questions without portraying a future without hope.

**Early School-Age Period**

The initial disability in DMD typically occurs by age 5 years and includes clumsiness, falling, and inability to keep up with peers while playing. The young child’s gait pattern is only slightly atypical, with an increased lateral trunk sway (waddling). Attempts at running, however, accentuate the waddling progression, and neither running nor jumping is attained. The Gowers sign (using the arms to push on the thighs to attain standing) is usually present after one or repeated trials of assuming standing position from sitting on the floor.

Stair climbing and arising to standing from the floor become progressively more difficult and signal the first significant functional limitation by age 6 to 8 years. Progressive changes in the gait pattern include the deviations of an increased base of support, pronounced lateral trunk sway (compensated Trendelenburg), toe-walking, and retraction of the shoulders with lack of reciprocal arm swing. Toe-walking initially may be a compensation for weakness of the abdominal and hip extensor muscles, resulting in lordosis and forward shift of the body’s center of mass with later evidence of contracture of the posterior calf musculature.

Toe-walking caused by contracture of the posterior calf musculature, in-toeing with substitution of the tensor fasciae latae to compensate for weakness of the iliopsoas muscles, falls resulting from progressive weakness, and complaints of fatigue while walking become increasingly frequent from age 8 to 10 years. A restrictive pattern of pulmonary impairment and progressive decline in maximal vital capacity also becomes increasingly evident (Galasko et al., 1995).

**Examination Considerations**

An examination to document functional impairment and disability progression is essential. Various formats have been reported (Brooke, 1986; Steffensen & Hyde, 2001; Vignos et al., 1996) that are variations of the guidelines initially published by Swinyard and associates (1957). A classification system published by Vignos and colleagues (1963) is outlined in Box 15-1. A more detailed examination format for DMD has been published by Brooke and associates (1981), which includes pulmonary function and timed performance of activities (see Appendix I). Normative data for DMD has been published using the clinical protocol by Brooke and associates (1989). Other functional assessment tools, such as the Pediatric Evaluation of Disability Inventory (PEDI) (Haley et al., 1992), School Function Assessment (SFA) (Coster et al., 1998), EK Scale (Steffensen & Hyde, 2001), or Barthel Index (Nair et al., 2001), should also be considered for use to give more specific information on the child’s functional skills. The EK Scale was recently validated for DMD and SMA, and includes ordinal scoring of 10 categories including items on mobility, transfers, ability to cough/speak, and physical well-being. The PEDI, the SFA, or the Vignos (Brooke) functional testing format can be used for diagnosis of other types of MD or of SMA.

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**Box 15-1 Vignos Functional Rating Scale for Duchenne Muscular Dystrophy**

1. Walks and climbs stairs without assistance
2. Walks and climbs stairs with aid of railing
3. Walks and climbs stairs slowly with aid of railing (over 25 seconds for eight standard steps)
4. Walks, but cannot climb stairs
5. Walks assisted, but cannot climb stairs or get out of chair
6. Walks only with assistance or with braces
7. In wheelchair: sits erect and can roll chair and perform bed and wheelchair ADL
8. In wheelchair: sits erect and is unable to perform bed and wheelchair ADL without assistance
9. In wheelchair: sits erect only with support and is able to do only minimal ADL
10. In bed: can do no ADL without assistance

Muscle weakness is apparent in the school-age child by age 6 to 8 years and should be objectively documented using a handheld dynamometer (Stuberg & Metcalf, 1988), electrodynamometer (Saranti et al., 1980), isokinetic dynamometer (Molnar & Alexander, 1973; Scott et al., 1982), or other device. Use of a dynamometer in conjunction with manual muscle testing has been shown to provide reliable information on the progression of weakness in key muscle groups (Brussock et al., 1992; Fowler & Gardner, 1967; Stuberg & Metcalf, 1988). Contracture development should be documented using goniometry and a standardized protocol. Intra-rater reliability of the measurements has been shown to be acceptable to provide objective information for program planning when a standardized measurement protocol is used (Pandya et al., 1985).

A clinical estimate of respiratory function can be obtained through measurement of respiratory rate and chest wall excursion (using a tape measure) and by noting the child’s ability to cough and clear secretions. A portable spirometer is recommended to obtain a more direct and objective reading of expiratory capacity before the need for formal pulmonary function testing.

Physical therapy management typically begins when the child is initially diagnosed at age 3 to 5 years. Goals of the program are to provide family support and education, obtain baseline data on muscle strength and ROM, and monitor for the progression of muscle weakness that will lead to disability. Initial therapeutic input should not be burdensome to the child or family because the child is usually independent in all ADL before age 5 years. Information should be provided to the family pertaining to the therapist’s role as a member of the management team. An appropriate activity level to avoid fatigue should be discussed with the family and school staff. Information on services through the local MDA office should be provided, including identification of support groups or contact families.

Intervention Considerations
The role of exercise in the treatment of MD is controversial (Ansved, 2001; Brooke, 1986; Fowler, 1982; Vignos, 1983). It is widely accepted that both overexertion (Johnson & Braddom, 1971; Vignos et al., 1963) and immobilization (Vignos et al., 1963) are detrimental. The use of graded resistive exercise has been reported to have a range of results from good (Vignos & Watkins, 1966) to limited (de Lateur & Giaconi, 1979) to adverse (Ansved, 2001). Resistive exercise would theoretically be indicated with the disproportionate loss of type II (fast-twitch) muscle fibers in DMD (Edwards, 1980). However, the use of resistive exercise in the young school-age child should not be universally prescribed. Prescribing a submaximal exercise program early has been shown to have beneficial effects, but it should be offered only to families who have a specific desire to include it in the child’s program. Consideration should be given to the fact that significant muscle weakness is not seen in the early stage of the disease and the use of an exercise program may be burdensome to the child and family.

If exercise is initiated early, the key muscle groups to be included are the abdominal, hip extensor and abductor, and knee extensor groups. Abdominal exercises should include trunk curls as opposed to sit-ups, which will primarily strengthen the hip flexors. Assistance may be required for neck flexor weakness, because the head typically cannot be flexed from a supine position. Cycling and swimming are excellent activities for overall conditioning and are often preferred over formal exercise programs (Gardner-Medwin, 1980; Vignos, 1983). Standing or walking for a minimum of 2 to 3 hours daily is highly recommended (Siegel, 1978; Ziter & Allsop, 1976). High resistance and eccentric exercise should be avoided (Ansved, 2001).

Breathing exercises have been shown to slow the loss of vital capacity and forced expiratory flow rate (Koessler et al., 2001; Rodillo et al., 1989). Game activities such as inflating balloons or using blow-bottles to maintain pulmonary function can easily be included in a home program and will decrease the severity of symptoms during episodes of colds or other pulmonary infections.

The use of electrical stimulation in DMD has also been suggested as a means of slowing the progression of weakness and improving function. Scott and co-workers (1986) studied the effect of low-frequency electrical stimulation on the tibialis anterior muscle of 16 children with DMD. A 47% increase in maximum voluntary contraction was observed in younger children following a stimulation protocol used for 8 weeks, with little change noted in older children. The authors concluded that the results were encouraging and that further study on muscle groups used for functional activities was needed.

One of the primary considerations in the early management program of the young school-age child is to retard the development of contractures. Contractures have not been shown to be preventable, but the progression can be slowed with positioning and an ROM program (Hyde et al., 2000; Scott et al., 1981; Seeger et al., 1985; Wong & Wade, 1995).

The initial ROM program should include stretching the gastrocnemius-soleus and tensor fasciae latae. Progressive contracture of the gastrocnemius-soleus and tensor fasciae latae corresponds to gait deviations of toe-walking and an increased base of support. Stretching for the gastrocnemius-soleus can be done using a standing runner’s stretch. The child stands at a supportive surface,
places one leg back at a time with the knee straight, and leans forward. The position also assists with maintenance of hip flexor flexibility; however, specific stretching for the hip flexors should be included when any limitation is noted. Having the child lie supine with one thigh off the edge of a mat or bed and the other held to the chest (Thomas test position) can be used to stretch the hip flexors initially. An alternative method is discussed later for use as progression of hip extensor weakness evolves. A standing stretch for the tensor is accomplished by having the child stand with one side toward the supportive surface with the feet away from the wall and with the knee kept straight while leaning sideways toward the supportive surface.

A home ROM program should be emphasized for the young child and the family instructed in the stretching exercises. There is lack of agreement as to the frequency and duration of the stretching program. Suggested frequency of the program varies from once daily (Gardner-Medwin, 1980; Miller & Dunn, 1982; Scott et al., 1981) to twice daily (Vignos et al., 1963; Ziter & Allsop, 1976), and duration from 1 repetition up to 10 (Vignos et al., 1963). Other authors have suggested a time frame of 10 (Ziter & Allsop, 1976) to 20 (Gardner-Medwin, 1980) minutes to complete the stretching exercises. As a general recommendation, each movement should be repeated for 5 to 10 repetitions with a 10-second hold in the stretched position. The stretch should be done slowly and should not be painful. Increased risk of injury with the loss of myofibrils and replacement by connective tissue is present because of decreased muscle elasticity, and caution in using excessive passive force is advised. Reassessment of the contracture progression should be used as the final guide to stretching frequency and duration.

The ROM program can often be supplemented as part of the physical education program at school. Special instruction should be provided to the physical education teacher to develop an adapted program, particularly if the teacher does not have an adapted physical education endorsement. General physical education activities will also require modification for the child’s participation and should not be exhaustive. Physical fitness test activities such as push-ups, sit-ups, or timed running for long time periods should be modified or excluded to avoid fatigue or overwork weakness.

Night splints are helpful to slow the progression of ankle contractures. Scott and associates (1981) studied the efficacy of night splints and a home ROM program in a group of 59 boys diagnosed with MD ranging in age from 4 to 12 years. The subjects were categorized into three groups based on compliance with splint wear and use of stretching. The group that followed through on the daily passive stretching program and use of the below-knee splints over the 2 years of the study demonstrated significantly less progression of Achilles tendon contractures and less deterioration in functional skills, leading to a longer period of independent walking. Boys in the group that did not follow through on the stretching or splint program lost independent walking at a younger age. In a randomized study comparing the effect of ROM exercise with ROM and night splints, the combined intervention was found to be 23% more effective in slowing the progression of posterior calf contracture than ROM alone (Hyde et al., 2000). Similar findings were reported in a study by Seeger and colleagues (1985) that compared the use of night splints, stretching, and surgery.

The use of prone positioning at night to slow progression of the hip and knee flexion contractures may be possible if tolerated by the child. The recommendation to have the child sleep prone with the ankles off the edge of the bed has not been shown to affect the progression of the contractures but theoretically is sound.

Scoliosis is not common when the child is ambulatory, but the spine should be checked routinely (Wilkins & Gibson, 1976; Ziter & Allsop, 1976). Alignment of the spine should be closely monitored as weakness progresses to the stage of making walking difficult. Postural analysis using the forward bend test is recommended to monitor spinal alignment for scoliosis. Presence of a rib hump with the forward bend test verifies a structural versus functional curve of the spine. Amendt and colleagues (1990) have demonstrated that a rib hump measuring at least 5° of inclination with a scoliometer is a reliable method and correlated to radiographic assessment. The study by Amendt and colleagues (1990) was, however, not inclusive of children with DMD but demonstrates an objective method of noninvasive screening for scoliosis. Orthopedic referral is indicated if a rib hump is documented.

Falls and complaints of fatigue while walking become increasingly more frequent as the child reaches age 8 to 10 years. Guarding during stair climbing or during general walking should be considered to ensure safety as balance becomes tenuous. A manual wheelchair with appropriate fit and accessories will allow for limited mobility as walking becomes more difficult. As progression of weakness in the trunk and hip girdle begins to make walking difficult, a similar amount of weakness of the shoulder girdle musculature is also present, making propulsion of a manual wheelchair difficult except on level and smooth surfaces such as linoleum. A motorized scooter should be considered to provide the child with independence, provided that access is available in the home and school (Fig. 15-4).

Information should be made available to families concerning recreational activities provided through the local chapter of the MDA or other groups. Summer MDA camp
is a wonderful experience for most children, and a support group is often developed for the child or family through participation in MDA or other group activities that provide not only physical but also emotional support.

**Adolescent Period**

Adolescence marks a time of significant disability progression as a result of the combined impact of muscle weakness and development of contractures. Walking is lost as a means of mobility, and increasing difficulty in general mobility with transfers is seen. Use of a manual wheelchair or powered mobility becomes necessary during adolescence. If powered mobility is used, assistance with finances for purchase of equipment or home modifications for access is typically needed, with coordination through a social worker or MDA patient services coordinator.

Changes in physical capacity such as muscle strength and pulmonary function using the EK Scale have been reported by Steffensen and colleagues (2002) for adolescents. Muscle weakness leads to increasing difficulty with ADL, including dressing, transfers, bathing, grooming, and feeding, and subsequent increasing involvement of the physical therapist and occupational therapist. Decisions regarding possible surgical intervention are also considered for the management of scoliosis or contractures or to prolong walking with the use of orthoses.

As muscle weakness becomes more pronounced in the trunk and hip musculature, and contractures of the hip flexors, tensor fasciae latae, and gastrocnemius-soleus progress, walking becomes increasingly difficult until cessation of independent walking occurs, usually by age 10 to 12 (Brooke, 1986). If orthoses are used to maintain a standing or walking program, they should be initiated before the child reaches the stage of being nonambulatory.

Various methods to predict the termination of walking have been reported including 50% reduction in leg strength (Scott et al., 1982; Vignos & Archibald, 1960), manual muscle test grade below grade 3 for hip extensors or below grade 4 for ankle dorsiflexors (McDonald et al., 1995) or inability to climb steps. Brooke and colleagues (1989) reported the cessation of unassisted walking within 2.4 years (range of 1.2–4.1 years) when 5 to 12 seconds were required to climb four standard steps or within 1.5 years (range 0.6–2.2 years) when greater than 12 seconds were required. An alternative method, shown in Figure 15-5, has been suggested by Siegel (1977). The knee extension lag while sitting and the hip extension lag while prone are assessed to predict the cessation of independent walking. If the combined lag is greater than 90°, the termination of independent ambulation is within a few months. Monitoring and management of contractures becomes a key element in maintaining walking when the older child spends more time in a sitting position. Because the hip extensor musculature is significantly weak in the early stage of the disease and weakness of the quadriceps muscles becomes pronounced by age 8 to 10 years, inability to maintain the center of gravity behind the hip joint or in front of the knee joint during stance will lead to loss of the ability to stand.
Manual stretching of the hip flexors, tensor fasciae latae, and heel cords is necessary because the older child demonstrates difficulty in carrying out the exercises without assistance. Prone lying will help retard the development of hip and knee flexor contractures, but stretching of the hip flexors and tensor using the method shown in Figure 15-6 is recommended. The leg is initially positioned in abduction and then moved into maximal hip extension and then hip adduction. The knee can be extended to provide greater stretch for the iliotibial band and tensor.

Figure 15-6 Prone stretching of the hip flexor, iliotibial band, and tensor fasciae latae. The hip is first positioned in abduction and then moved into maximal hip extension and then hip adduction. The knee can be extended to provide greater stretch for the iliotibial band and tensor.

Continuation of Standing or Walking
The use of orthoses for a standing program or continuation of supported walking is not appropriate for all individuals; in fact, it should be considered a personal rather than therapeutic decision. Although a standing program may be useful to slow the progression of contractures, a braced walking program has little long-term functional or practical application because the child will eventually use a wheelchair. Continuation of standing through use of a standing frame, knee imm mobilizers, or KAFOs is a goal at our facility to address the issue of decreased bone mineral density (McDonald et al., 2002) and subsequent increased risk of fracture (Bianchi et al., 2003). Because surgery is often required in addition to orthotics for prolonged ambulation, both the parents and adolescent must agree in the management decision. Prolongation of ambulation through surgery and orthotics is not a common goal at our facility. Limited resources are more typically used for power mobility equipment, adaptive equipment, or environmental adaptations.

Prognostic factors for success that should be considered in making the decision to use orthoses to prolong walking include the residual muscle strength (approximately 50%) (Vignos, 1983); absence of severe contractures (Spencer & Vignos, 1962); timely application of the braces (Bach & McKeon, 1991; Spencer & Vignos, 1962); residual walking ability (Vignos & Archibald, 1960); and motivation of the child and family (Bowker & Halpin, 1978). The degree of mental impairment and obesity should also be considered. The timely use of orthoses has been shown to prolong walking (Bach & McKeon, 1991; Bowker & Halpin, 1978; Heckmatt et al., 1985) and to increase the child’s longevity (Bach & McKeon, 1991; Miller & Dunn, 1982; Vignos et al., 1963).

If the decision to use orthoses to prolong standing or walking is made, KAFOs should be prescribed (Fig. 15-7) (Bakker et al., 2000; Bowker & Halpin, 1978; Siegel, 1975). Ankle-foot orthoses are appropriate for positioning but do not provide the knee stability required to avoid falls while walking. Although the use of AFOs to control equinus is common for other diagnoses such as cerebral palsy, with DMD the orthoses often interfere with the use of an ankle strategy and the preserved distal strength that is needed for standing balance and walking. A reciprocating or wheeled walker may be helpful when assistance for balance is needed. Assistive devices such as a standard walker, crutches, or canes are seldom functional because of the degree of proximal shoulder girdle and upper extremity muscle weakness. Standby assistance should be provided when KAFOs are used, owing to the risk of injury from falls. Closer guarding and increased assistance will be needed as the weakness progresses. Transfers to and from standing are dependent because the knee joints of the KAFO must be locked to provide stability. The KAFOs can be used for continuation of a standing program even after walking is no longer possible.

Surgical intervention is commonly needed in conjunction with the use of braced walking as contractures progress. Documented indications for surgery include ankle plantar flexion contractures of greater than 10°, iliotibial band contractures greater than 20°, or knee-hip flexion contractures greater than 20° but less than 45° (Bowker & Halpin, 1978). Subcutaneous tenotomy of the Achilles tendon is often performed to correct equinus and also to decrease the subluxing force placed across the hip joint when the leg is in the sagittal plane during the maneuver.
tendon and fasciotomy of the iliotibial bands are the most commonly reported surgical procedures (Bach & McKeon, 1991; Bowker & Halpin, 1978; Hsu, 1995; Siegel et al., 1972; Vignos, 1983). Transfer of the posterior tibialis tendon is occasionally used for correction of the equinovarus foot posture (Hsu, 1995; Scher & Mubarak, 2002).

An intensive postoperative management program is essential to retard the effects of immobilization (Siegel et al., 1986). Standing in the plaster casts can be done on the first or second postoperative day. Gait training is begun as tolerated, and general conditioning exercises for the hips, trunk, and upper extremities are recommended. Breathing exercises should also be stressed. A smooth transition from the casts to bracing is ensured by having the child fitted for the KAFOs before hospitalization for surgery.

Standing pivot transfers must eventually be replaced by one- or two-person lifts or use of equipment as a result of the development of knee and hip flexion contractures and pronounced weakness of the lower extremities. Transfers to and from the wheelchair, toilet, tub, car, and furniture usually become dependent by age 12 to 14. A sliding board, manual lift, or hydraulic lift can be used during transfers. Proper instruction for transfers is needed because the degree of trunk muscle weakness makes sitting balance tenuous by this stage. If the caregiver is using manual lifting for transfers, he or she should be observed for and instructed in proper body mechanics and safety. A hydraulic lift can be used for transfers to and from the wheelchair, particularly when the adolescent is large or obese or when the caregiver cannot safely perform a manual lift. A U-style sling should be used with the lift to provide adequate head and trunk support during transfers. A tub lift or bath bench for bathing will be needed and a wheeled commode-shower chair should be considered depending on bathroom accessibility.

**Mobility and Spinal Alignment**

A power scooter as shown in Figure 15-4 should be considered as an initial power wheelchair prescription for the child who is hesitant to use a power wheelchair when walking is no longer possible. The scooter is often more easily accepted by the child and may be used for transition to a standard power wheelchair. If a power scooter is initially used, transition to a power wheelchair will be necessary when the adolescent is seen propping on the arm rests for trunk control. Asymmetric sitting postures must be aggressively managed owing to the correlation of increased sitting time and poor sitting posture with the onset of scoliosis. When limited resources are an issue, which is typically the case for children in managed care, a power wheelchair should be acquired without consideration for a scooter.

A manual wheelchair will be needed if nonaccessible areas for a powered wheelchair are encountered in the usual environment. Architectural barriers in the home or inability to transport a power wheelchair may also necessitate the use of a manual wheelchair.

Fit of the wheelchair must be closely monitored to provide adequate support. The reader should refer to Chapter 33 for information on wheelchairs and postural support systems. Special attention should be given to alignment of the spine and pelvis and to the need for customized accessories or modifications. Accessories to be considered for the manual or power wheelchair prescription should include a solid back and seat, lateral trunk supports, lumbar support, adductor pads, seat belt, and chest strap. The footrests should be modified to support the ankle in a neutral position. Additional items that may be appropriate include a tray; head support, if needed; or coated push rims, if the child has the strength to propel the wheelchair. A reclining back will allow a position change while sitting in the wheelchair and will help deter flexion contracture formation at the hip, or a tilt-in-space reclining option can be considered to allow for pressure...
Muscular Dystrophy and Spinal Muscular Atrophy

Exercise and Custom Equipment

With the cessation of walking in late childhood or early adolescence, the emphasis of an exercise program should shift from the lower extremities to active-assistive and active exercises of the upper extremities. More important, however, active exercise should be encouraged by having the adolescent assist as much as possible in ADL such as grooming, upper body dressing, and feeding through consultation with an occupational therapist. Key muscle groups for maintenance of strength for transfers include the shoulder depressors and triceps. The shoulder flexor and abductor and elbow flexor muscle groups are key areas for exercises to maintain routine ADL such as self-feeding and hygiene. Weakness of the upper arm musculature by 16 years of age makes ADL such as dressing, feeding, or hygiene extremely difficult.

The ROM program will require further modification as the adolescent becomes nonambulatory. Stretching of the aforementioned lower extremity joints should be continued with stretching of the shoulder and elbow included. Limitation in shoulder flexion and abduction, elbow extension, forearm supination, and wrist extension are most common.

The family will need to consider additional equipment or home modifications as their child reaches adolescence. A van with a lift or ramp will be needed to transport a power wheelchair. Modification of the bathroom can significantly assist the family by using a wheeled commode chair for toileting and a bath chair and handheld shower for bathing. A tub lift is a second option for bathing. A urinal should be available at home and school to decrease the frequency of transfers to the toilet. Modifications of the bed are also frequently required because the adolescent is unable to change position. An airflow mattress, egg-crate foam cushion, or hospital bed are all possibilities to be considered. A positioning program to include position changes at night is necessary for adolescents who are thin to provide comfort and ensure against skin breakdown. Customized foam wedges fabricated by the therapist may also be helpful in positioning at night.

Transition to Adulthood

The transition to adulthood marks a time of continued progression of disability with greater reliance on assistive technology for environmental access and increased need for assistance to carry out routine ADL (Stuberg, 2001). Mobility using a power wheelchair is necessary because upper extremity and truncal weakness will typically not allow use of a motorized scooter. Assistance for ADL, including dressing, transfers, and bathing, is now required. Hygiene about the face and feeding become increasingly difficult but usually remain manageable. Many social issues also arise with the completion of educational programming and transition to a prevocational, vocational, or home environment on a more full-time basis. Another major issue that requires thoughtful consideration by the family, individual, and management team is the utilization of assisted ventilation with progressive respiratory involvement at the terminal stage of the disease.

All transfers require assistance during late adolescence and by adulthood typically require use of a hydraulic or other type of mechanical lift. A high-backed sling seat is indicated because head and trunk control is minimal with the progressive weakness.
A power recline feature on the power wheelchair may be desirable, depending on accessibility and family choice, if funding is available. If not, a regular schedule for pressure relief through lateral weight shifting with assistance is needed. A properly fitted and well-tolerated cushion to avoid skin breakdown becomes an important area of intervention with loss of the ability to weight shift in the wheelchair. Skin breakdown is not a typical problem in DMD, but a cushion should be considered. A Jay Medical cushion (Jay Medical, Boulder, CO) is often well tolerated and provides a firm base of support to control pelvic obliquity, yet the gel inserts can be adjusted to allow for adequate pressure distribution. A customized insert will be needed if deformity becomes severe (e.g., severe scoliosis without surgical stabilization).

A ball-bearing feeder may be required to assist arm movement when progression of upper extremity weakness makes independent feeding difficult (Chyatte et al., 1965). The device can also be used to assist with general use of the arms in conjunction with activities at a table, such as when using a computer. Coordination of planning with an occupational therapist to address feeding and dressing issues is needed to identify solutions to increased dependence in feeding, dressing, and hygiene.

To maintain independence in environmental access, consideration should be given for using environmental control devices. An environmental control unit included on the power wheelchair can be used to independently access the lights, telephone, television, motors on doors, or a computer, to name just a few applications. Computer access for vocational applications such as word processing or avocational activities such as games is available.

Breathing exercises, postural drainage, or intermittent pressure breathing treatments should be included in the management program based on results of pulmonary evaluation (Rodillo et al., 1989). Specific tests of pulmonary function that document respiratory status include forced vital capacity (FVC = amount of air expired following a maximal inspiration) and peak expiratory flow rate (highest flow rate sustained for 10 ms during maximal expiration). Continuous positive airway pressure (CPAP) is recommended when FVC is below 30% of age-adjusted norm values (Lyager et al., 1995; Steffensen et al., 2002). Assisted ventilation with tracheostomy is recommended when respiratory insufficiency is present with abnormal blood gas levels during the day or night (Steffensen et al., 2002). In addition to the breathing exercises and assisted coughing, the family and caregivers should be instructed in the technique of postural drainage.

Close monitoring of respiratory function should become routine with increasing age because respiratory failure or pulmonary infection is the major contributing factor to death in 75% of children with DMD (Gilroy & Holliday, 1982). Longevity in DMD can be significantly prolonged by assisted ventilation (Bach et al., 1987; Curran & Colbert, 1989; Eagle et al., 2002). A 1997 survey of MDA-sponsored clinics reported that 88% of clinic directors offered noninvasive ventilatory aid for acute respiratory failure (Bach & Chaudhry, 2000). Bach and Chaudhry (2000) stressed the need for health care professionals to explore attitudes toward mechanical ventilation because our perceived impression of patient desires may often be incorrect. The use of daytime intermittent positive-pressure ventilation via mask or nasal cannula, nocturnal bilevel positive airway pressure (BiPAP), CPAP, negative pressure ventilators, and suctioning should be considered for the chronic hypoventilation related to weakness of respiratory musculature (Bach & Chaudhry, 2000).

A power-controlled bed to allow elevation of the head for respiratory management should be considered. Use of a bed with elevating capability also allows for greater ease in transfers, and height adjustment promotes use of proper body mechanics by family members for activities that require assistance such as dressing. Mattress selection should also be reviewed with the family because an airflow mattress may be needed when increasing dependence for bed mobility is encountered. Use of an airflow mattress may decrease the frequency of need for turning and repositioning at night. If sitting in a wheelchair is no longer tolerated in the later stages of the disease, elevation of the head of the bed becomes beneficial for reading or watching television. An easel will be required for reading.

Although it may be assumed by care providers that the quality of life and therefore satisfaction are significantly reduced for severely disabled individuals with DMD, this notion may be incorrect. In a survey of 82 ventilator-assisted individuals with DMD, Bach and colleagues (1991) concluded that the vast majority of individuals had a positive affect and were satisfied with life despite the physical dependence. Furthermore, it was found in a survey of 273 physically intact health care professionals that they significantly underestimated patient life satisfaction scores, and therefore they may make patient management recommendations based on their attitudes rather than the patient’s wishes. Bach and colleagues (1991) strongly recommended that we as professionals need to constantly inquire and objectively assess family and individual needs when interacting to provide therapeutic programs. Curran and Colbert (1989) have reported an average increase in longevity from 19 years 9 months to 25 years 9 months in individuals who use ventilatory assistance.

Respiratory insufficiency is a hallmark sign of the pre-terminal stage of DMD (Newsom-Davis, 1980). Progressive muscular weakness results in decreased ventilatory volumes...
caused by restriction of chest wall excursion. Coordination of care with the respiratory therapist is essential when clinical findings of respiratory muscle weakness, inability to cough, or chest wall restrictions are observed (Burke et al., 1971). Severe oxygen desaturation leading to a comatose state is evidence of the terminal stage of the disease.

Members of the team often become involved in answering questions regarding death. The physical therapist should be aware of the stages of disease progression and especially the preterminal signs to avoid making inappropriate comments concerning prognosis. Often little needs to be said, but rather a good listening ear is needed to help the family work through the crisis that is ever pending. It is often a comfort to individuals with DMD or family members that the end may come as a sleep without wakening. The person with DMD and his family members may indicate the need for additional support, but if issues are not being resolved adequately by the support that is available, consideration for involvement by a psychologist, counselor, clergy member, MDA support group, or other trained professional is indicated. Literature is available through the MDA to comfort family members, and texts are available if the family is interested (Charash, 1987; Ringel, 1987).

**Becker Muscular Dystrophy**

BMD, a more slowly progressive variant of DMD, has an incidence of about 1 in 20,000 births and a prevalence of 2 to 3 cases per 100,000 population (Emery, 1993). The impairments and participation restrictions of BMD closely resemble those of DMD; however, the progression is significantly slower, with a longevity into the forties (Emery & Skinner, 1976; Gilroy & Hollliday, 1982). The genetic defect for BMD is located on the same gene as that for DMD only in a different area; therefore, dystrophin is present in reduced amounts or abnormal size rather than completely absent as in DMD, which may explain the slower progression of clinical symptoms (Liechti-Gallati et al., 1989).

Initial clinical symptoms are typically not identified in boys with BMD before late childhood or early adolescence. Emery and Skinner (1976) found the mean age at onset of symptoms to be 11 years, inability to walk at 27 years, and death at 42 years. The authors pointed out, however, that the range of walking cessation is very wide. Perhaps one of the best functional discriminators between BMD and DMD is that 97% of adolescents with DMD are using a wheelchair for mobility by age 11 years, whereas 97% of adolescents with BMD are still walking (Emery & Skinner, 1976). Another discriminator is the frequent complaint of muscle cramping in individuals with BMD that is rarely reported in DMD (Dubowitz, 1992).

The impairments of BMD are the same as in DMD, although less severe, and the initial clinical signs include frequent falls and clumsiness in the mid- to late teens. The pattern of weakness is the same as in DMD, and pseudohypertrophy of the calves may be present. The incidence of contracture, scoliosis, and other skeletal deformities is lower in BMD. Although not as severe as in DMD, hip, knee, and ankle plantar flexor muscle contractures can be present when walking is no longer possible. The use of night splints to maintain ankle dorsiflexion ROM is often indicated, along with a home program of heel cord stretching. Significant disability will develop by the midtwenties, requiring the use of power mobility and consideration for use of orthoses to maintain walking. KAFOs can also be used to prolong walking; however, braced ambulation will not be functional for community access but rather as a means of exercise. The general goals and management procedures outlined in the section on DMD are the same for BMD, including the progression from walking to use of power mobility.

Because the person with BMD lives much longer than someone with DMD, transition planning following school and assistance with living arrangements into adulthood become major issues. Vocational or avocational choices should be made with the disease progression and disability level in mind. Vocational rehabilitation services should be initiated before completion of high school to allow adequate time for evaluation. Governmental support through Medicaid, Social Security benefits, or other sources may be needed to offset expenses to allow for independent living because adaptive equipment and an attendant will be needed. Ongoing medical services are available through the MDA. No data are available regarding the number of individuals who go to college or become employed following high school, but with the assistive technology available to promote independence, either option can be explored.

**Congenital Muscular Dystrophy**

Congenital myopathies as a diagnostic category consist of many diseases, including congenital MD. Congenital MD is a heterogeneous group of muscle disorders with onset in utero or during the first year of life. Reported forms of congenital MD are (1) congenital MD with central nervous system (CNS) disease (Fukuyama syndrome, Walker-Warburg disease, and muscle-eye-brain disease), (2) merosin-deficient congenital MD, (3) integrin-deficient congenital MD, and (4) congenital MD with normal merosin. Fukuyama, merosin-deficient, and normal merosin forms will be discussed. The reader should refer to the review article by Voit (1998) for additional information. Another valuable resource for information is the
Online Mendelian Inheritance of Man (OMIM) website of the National Center for Biotechnology. The mode of inheritance in congenital MD is reported as autosomal recessive (Emery, 2002; Muscular Dystrophy Association, 2000). Although all forms are rare, the range of severity and disability varies significantly among types.

In congenital MD with associated CNS disease (Fukuyama type), mental retardation and seizures are common along with moderate to severe hypotonia at birth and the presence of contractures (Fukuyama et al., 1981). Magnetic resonance imaging reveals nonspecific cerebral malformations and occasionally lissencephaly as pathologic features of the CNS disease. Contractures typically involve the lower extremities (hips and knees) and elbows. Other commonly reported dysmorphic features include congenital dislocation of the hips, pectus excavatum, pes cavus, kyphoscoliosis, and an unusually long face. Weakness of the extraocular muscles, optic atrophy, and nystagmus have been reported (Brooke, 1986). Children with this type of MD rarely attain the ability to walk (Emery, 2002). The genetic defect is at chromosome 9q31-q33 with speculation that the missing protein is “fukutin.”

The early management program in children with congenital MD with nervous system disease should focus on family instruction, developmental activities to address delays in gross motor skill development, and aggressive management of contractures. Attention to positioning is necessary to guard against secondary deformity resulting from gravitational effects on the trunk with presence of moderate to severe hypotonia. Early intervention by an occupational therapist to address feeding and oral motor control issues is commonly coordinated with physical therapy. Impaired respiratory function and pulmonary complications are hallmark features of congenital MD. The family should be instructed in chest physical therapy, such as postural drainage, and consultation with a respiratory therapist may be needed on an ongoing basis.

Because many children with congenital MD and associated nervous system disease do not attain walking, maximizing functional skills in sitting becomes a primary goal of the physical therapy management program as the child ages. Therapeutic exercise to improve head and trunk control should be aggressively addressed with use of adaptive equipment to slow the progression of spinal deformity and contractures and to maximize access to the environment. Because mental retardation is common, power mobility may not be an option. Additional management issues for children with significant hypotonia are discussed later in the chapter in the section on acute SMA. In congenital MD with merosin deficiency, the typical clinical presentation includes hypotonia and weakness, contractures, normal intelligence, seizures (20%), and a delay in acquisition of motor milestones. Infants demonstrate a delay in walking, with acquisition of walking ranging from 13 months to 6 years (North et al., 1996). Progressive contractures may be present, and in severe cases, the children may never walk. Longevity ranges from 15 to 30 years. Peporago and associates (1998) reported on a cohort of 22 children with merosin-deficient congenital MD. All children demonstrated severe floppiness at birth, normal intelligence, and delay in achievement of motor milestones. Merosin-deficient congenital MD is due to a defect at chromosome 6q22. Muscle weakness and contractures are the primary impairment in merosin-deficient congenital MD. Muscles innervated by the cranial nerves may be involved, requiring a feeding program that is coordinated with occupational therapy. Contractures must be managed aggressively with a home ROM program including manual stretching, positioning, and splinting. Because many of the children have a potential to develop walking, the ankle plantar flexion contractures may require orthopedic intervention if the contractures cannot be managed conservatively.

Activity limitations, such as delayed acquisition of gross motor skills, should not be managed by direct service therapy programs because a slower rate of skill progression is expected. Because these children vary in their rate of motor skill development, information can be provided to the family concerning probable rates of motor skill acquisition, but unrealistic therapeutic expectations should be avoided. Because there is a wide range of functional deficits in children with congenital MD, care must be taken in predicting functional gross motor outcomes or level of participation at home and school.

CHILDHOOD-ONSET FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY

Facioscapulohumeral MD is rare, with an incidence of 3 to 10 cases per million births (Stevenson et al., 1990). The disorder is inherited as autosomal dominant or recessive with the genetic defect on chromosome 4q35. The disorder affects males and females equally. Childhood-onset facioscapulohumeral MD typically results in the onset of clinical signs within the first 2 years but without significant impairment or disability until later in the first decade. Contractures are seldom a problem.

Infancy and Preschool-Age Period

The impairment of muscle weakness about the face and shoulder girdle is typically the only prominent feature of the disease during the infant and preschool-age period. Parents report that the child may sleep with the eyes par-
tially open, and on physical examination weakness of the facial musculature is predominant. Children are frequently unable to whistle, and drinking with a straw may be difficult. When asked to purse the lips together and puff the cheeks out, the child is unable to maintain the cheeks out when even the slightest pressure is applied. The child’s smile is also masked because of the weakness, thereby hindering communication as a result of inconsistency between what is spoken and the affect displayed.

Children with childhood-onset facioscapulohumeral MD typically develop independence in walking without significant delay. An excessive lordotic posture during walking is a classic clinical feature with progression of weakness. The scapulae are widely abducted and outwardly rotated, giving evidence of the degree of interscapular muscle weakness.

**School-Age Period**

Progressive disability occurs during the school-age period, with weakness becoming more generalized throughout the trunk, shoulder, and pelvic girdle musculature. Progression of childhood-onset facioscapulohumeral MD is more insidious than the adult form, and independent walking may be lost by the end of the first decade (Gardner-Medwin, 1980).

The severe winging of the scapula, a hallmark feature of the adult form of the disease, becomes more prominent with age in activities such as reaching overhead. Management should focus on instruction to the child and family on activities to avoid that may cause fatigue and on guarding against heavily resisted upper extremity activity. Studies of adults with facioscapulohumeral MD comparing dominant with nondominant arm strength have shown that overuse and perhaps just consistent use of the dominant arm play a significant role in progression of muscle weakness (Brouwer et al., 1992; Johnson & Braddom, 1971).

As weakness of the hip and knee extensors progresses, the use of KAFOs should be considered for assisted walking and transfers. When walking becomes increasingly difficult, power mobility using a scooter or power wheelchair should be considered because the degree of upper extremity weakness will not allow independence in propulsion of a manual wheelchair.

**Transition to Adulthood**

No specific prognostic information on the longevity of individuals with childhood-onset facioscapulohumeral MD is available, and therefore transition planning from the educational environment should be a goal of the therapy program. If severe weakness is present and significant assistance from the family is needed, individuals may not desire to plan for living outside the family home. If independent living is desired, coordination of planning with an attendant will be necessary and evaluation for accessibility issues will need to be completed. Assistance through vocational rehabilitation services should be coordinated with transition planning if vocational goals are identified.

**CONGENITAL MYOTONIC MUSCULAR DYSTROPHY**

Myotonic MD is the most common adult-onset neuromuscular disease, with an incidence of 1 in 8000 births (Harper, 1989). Congenital myotonic MD is rare and demonstrates severe clinical features of the adult-onset diagnosis. Inheritance is reported as autosomal dominant with genetic defect of chromosome 19q13.3 affecting males and females equally. More recently a second form of myotonic MD (proximal myotonic myopathy, or PROMM) has been reported and linked to chromosome 3q21 (Ricker, 2000). Children with congenital myotonic MD are almost exclusively born to mothers with myotonic MD who have the chromosome 19 defect. Approximately 25% of children born to mothers with myotonic MD will have congenital myotonic MD (Harper, 1989). Most children demonstrate severe hypotonia and weakness at birth; however, a few children first have only signs of mental retardation by age 5 years and no significant motor impairment as infants. Because the children who initially have only mental impairment follow a progression of motor impairment similar to that of adult-onset myotonic MD, the infancy-onset form is discussed in this section of the chapter.

Mental retardation in congenital myotonic MD is common, with an average IQ of 65 typically reported (Harper, 1975; Roig et al., 1994). There is no evidence of progressive deterioration of mental function. A study by Rutherford and co-workers (1989) including 14 children provided prognostic information regarding survival and the relationship to mechanical ventilation at birth. No infant in the study survived who required mechanical ventilation for longer than 4 weeks.

**Infancy**

If the child survives the early weeks of life, the prognosis is one of steady improvement in motor function over the first decade, with most children developing independent walking (Harper, 1975, 1989; Roig et al., 1994). A follow-up study by O’Brien and Harper (1984) of 46 children reported only 4 children who died outside the neonatal period at ages 4, 18, 19, and 22 years. Four additional children demonstrated significant disability associated with a poor prognosis, and none was older than age 30 years. In
a study of 115 children with congenital MD, Reardon and colleagues (1993) reported that 25% of the children lived to age 18 months, and of those who survived infancy, 50% lived into their mid-30s.

Severe weakness and partial paralysis of the diaphragm at birth are clinical features that often suggest the diagnosis of congenital myotonic MD. Myotonia (delay in relaxation after muscular contraction), a hallmark feature of adult myotonic MD, is typically not evident at birth in the congenital form but rather develops by 3 to 5 years of age (Brooke, 1986). Myotonia in congenital myotonic MD is typically not considered to be a significant impairment or a cause of functional limitations in comparison with the degree of weakness that is present. The symptoms of myotonia, however, are increased with fatigue, cold, or stress (Siegel, 1986). Typical facial features include a short median part of the upper lip, which gives the mouth an inverted-V shape. Facial movements are limited, with muscles innervated by the cranial nerves involved in the severe weakness pattern. Severe respiratory impairment is prominent in the newborn period, requiring resuscitation and assisted ventilation in most cases. Talipes equinovarus contractures are reported in over 50% of children, and a general pattern of arthrogryposis occurs in less than 5% (Harper, 1989).

Progressive improvement in gross motor skills can be expected if the child survives the newborn period. The presence and degree of intellectual impairment become a major factor in the progression of milestone acquisition. The development of hip abduction and external rotation contractures should be closely monitored if leg movement and habitual positioning favor the development of this secondary impairment.

Harper (1989), in a cohort of 70 children with congenital myotonic dystrophy, reported that hypotonia is rarely prominent beyond age 3 to 4 years. Children typically develop walking, but further motor impairment follows the clinical progression of adult-onset disease without definitive data being available into adulthood to document disease progression.

Consultation with a respiratory therapist on pulmonary care will be needed until the infant is weaned from assisted ventilation. Feeding may require the use of a nasogastric tube during the newborn period or early infancy, and initiation of a feeding program should be coordinated with an occupational therapist. A swallowing study may be indicated to evaluate potential for aspiration when the feeding program is initiated. If the newborn survives early respiratory difficulties, progressive improvement in pulmonary function is usually seen without need for ongoing intervention.

Talipes equinovarus contractures should be aggressively managed in infancy with casting, taping, and exercises but may ultimately require orthopedic intervention as they have been shown to significantly delay walking (Reardon et al., 1993). Ankle-foot orthoses or night splints may be indicated based on individual needs. In addition to home instruction for ROM activities to manage contractures, the family should be provided with a general program of activities to promote gross motor skill development. Because the natural progression of the disease is improvement of motor function, consultation rather than a direct service program is indicated, unless surgical intervention for contracture management is required.

School-Age Period
Consultation for development of adaptive physical education activities will be needed during the school-age period. Other physical therapy–related activities will depend on the use of orthoses and progression of gross motor skill development. Specific therapeutic exercise programs for strengthening have not been reported but may be indicated in addition to ROM activities.

Transition to Adulthood
The natural progression of myotonic MD is insidious weakness of the distal upper and lower extremity musculature and progressive increase in myotonia, leading to increasing disability. Children with congenital myotonic MD will demonstrate progression in the disease as described for adults, but typically at an earlier stage, usually by the middle of the second decade. The reader should refer to references on adult myotonic MD for further information on clinical course and management (Brooke, 1986; Harper, 1989; Siegel, 1986).

Emery-Dreifuss Muscular Dystrophy
Emery-Dreifuss MD1 (EDMD1) is inherited as an X-linked recessive disorder at gene locus Xq28 resulting in the absence of the protein emerin. Emery-Dreifuss MD2 (EDMD2) is also inherited, but as an autosomal dominant or recessive disorder with a defect on chromosome 1q21.2 and defect of the protein lamin.

The clinical features of EDMD1 vary widely and include contracture of the posterior neck, elbow, and ankle joints. A humeroperoneal pattern of muscle weakness is observed with usual onset in the teen years, but ranging from neonatal to the third decade. Later progression of the weakness to the legs is reported. Contracture is typically seen before the onset of weakness. The onset of EDMD2 is most common in the first or second decade and a similar pattern of contracture to EMMD1 is seen. The humeroperoneal weakness is more prominent, especially
affecting the elbow flexors (Brooke, 1986). Cardiac abnormalities are more common in EDMD1, with sudden death (in persons ranging in age from 25 to 56 years) reported by Pinelli and colleagues (1987) in a large family cohort with bradyarrhythmias. Physical therapy management is limited during the childhood period because disability is not common. Independent walking is typically maintained into adulthood without significant disability. A ROM program for contracture prevention is advised, but orthopedic intervention is common to correct the Achilles tendon contractures (Shapiro & Specht, 1991). Holter monitoring is advised when cardiac abnormalities are identified, and pacemakers are used to control rhythm.

**SPINAL MUSCULAR ATROPHY**

Classification of SMA into four groups is based on clinical presentation and progression (Table 15-2). Types I and II are commonly referred to as acute and chronic Werdnig-Hoffmann disease, respectively, type III as Kugelberg-Welander disease, and type IV as adult-onset SMA.

**DIAGNOSIS AND PATHOPHYSIOLOGY**

SMA comprises the second most common group of fatal recessive diseases after cystic fibrosis (Semprini, 2001). The pathologic feature of SMA is abnormality of the large anterior horn cells in the spinal cord. The number of cells is reduced, and progressive degeneration of the remaining cells is correlated with loss in function.

The diagnosis of SMA is confirmed by clinical examination and laboratory procedures, including electromyography, muscle biopsy, and genetic testing (Brooke, 1986; MacKenzie et al., 1994). Electromyographic findings include fibrillation and fasciculation potentials. Nerve conduction velocities are normal. Muscle biopsy demonstrates changes that are typical of a disease involving denervation (i.e., large groups of atrophic fibers are dispersed among groups of normal or hypertrophic fibers). The absence of fibrosis around the atrophic groups on the muscle biopsy helps delineate SMA from DMD.

SMA is typically inherited as autosomal recessive with the genetic defect on chromosome 5q11.2-13 (Semprini, 2001). The gene for SMA, termed *survival motor neuron* (SMN), was discovered in 1994 by MacKenzie and colleagues (1994) on chromosome 5q13 and is responsible for the production of a protein bearing the same name. The SMN protein is involved in maintenance of the anterior horn cell and when missing results in a lack of survival of the cell, leading to apoptosis (programmed cell death). In addition to the SMN gene, another defect of the gene in a near locus results in the lack of formation of neuronal apoptosis inhibitory protein (NAIP), which has also been shown to play a role in SMA. Variation in the amount of NAIP protein may explain the premature cell death seen in SMA, in association with the SMN gene defect (Robinson, 1995).

The incidence of Werdnig-Hoffmann disease is 1 in 10,000 live births (Semprini, 2001), and the incidence of Kugelberg-Welander disease is reported as 6 cases per 100,000 live births (Winsor et al., 1971). Reported incidence of the other forms is variable because of the inconsistency of applying classification criteria.

One criterion for classification of SMA is the level of functional ability (Dubowitz, 1989). The more typical classification criteria are multifactorial and involve age at onset of the first clinical signs, pattern of muscle involvement, age at death, and genetic evidence (Pearn, 1980).

SMA is a heterogeneous disorder containing several different clinical presentations and rates of progression. Progressive SMA of early childhood (type I) was first reported by Werdnig and Hoffmann in the late 1800s (Hoffmann, 1893; Werdnig, 1894). A more slowly progressive form of SMA (type III) with onset usually be-

**TABLE 15-2**

<table>
<thead>
<tr>
<th>TYPE</th>
<th>ONSET</th>
<th>INHERITANCE</th>
<th>COURSE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Childhood-onset, type I,</td>
<td>0–3 mo</td>
<td>Recessive</td>
<td>Rapidly progressive; severe hypotonia; death within first year</td>
</tr>
<tr>
<td>Werdnig-Hoffmann (acute)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Childhood-onset, type II,</td>
<td>3 mo–4 yr</td>
<td>Recessive</td>
<td>Rapid progression that stabilizes; moderate to severe hypotonia; shortened life span</td>
</tr>
<tr>
<td>Werdnig-Hoffmann (chronic)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Juvenile-onset, type III,</td>
<td>5–10 yr</td>
<td>Recessive</td>
<td>Slowly progressive; mild impairment</td>
</tr>
<tr>
<td>Kugelberg-Welander</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
widely reported in the literature that all children with the development of scoliosis and often contractures. It is breathing by use of the abdominal musculature is typical. Milder forms of SMA appear alert and responsive. Respiratory myotonic or facioscapulohumeral MD, children with acute SMA (Marshall, 1984). Unlike the faces of children with most commonly reported in children with acute-onset fasciculations, including fasciculations of the tongue, are rare involvement with juvenile onset. Contractures may be inconsistently involved in childhood-onset SMA, with cerebellar and optic atrophy.

No cure or treatment is available for SMA, but physical therapy is commonly advocated (Marshall, 1984; Watt & Greenhill, 1984). Poor prognosticators for long-term survival include early age at onset (before 4 months of age) that is often noted as weak fetal movement; fasciculations of the tongue in infancy; and severe, generalized weakness, particularly of the trunk and proximal musculature (Gamstorp, 1967). More recent evidence has shown that children with severe onset of symptoms may survive up to their fifth birthday (Borkowska et al., 2002).

**Acute Childhood Spinal Muscular Atrophy (Type I)**

**Impairments, Activity Limitations, and Participation Restrictions**

The primary impairment in all forms of SMA is muscle weakness secondary to progressive loss of anterior horn cells in the spinal cord. Weakness is particularly pronounced in the acute and chronic childhood forms (types I and II) within the first 4 months. The cranial nerves are inconsistently involved in childhood-onset SMA, with rare involvement with juvenile onset. Contractures may be a primary impairment in acute-onset SMA, with reports of talipes equinovarus or other intrauterine deformities secondary to limited fetal movement. Muscle fasciculations, including fasciculations of the tongue, are most commonly reported in children with acute-onset SMA (Marshall, 1984). Unlike the faces of children with myotonic or facioscapulohumeral MD, children with acute childhood SMA appear alert and responsive. Respiratory distress is present early, and significant effort to augment breathing by use of the abdominal musculature is typical.

Secondary impairments in acute-onset SMA include the development of scoliosis and often contractures. It is widely reported in the literature that all children with SMA develop scoliosis, usually requiring surgical intervention (Granata et al., 1989c; Lonstein, 1989). Other secondary impairments include decreased respiratory capacity and easy fatigability. Because only the passage of time will allow differentiation of children with acute versus chronic childhood SMA, treatment should begin early with a focus on feeding, ROM, positioning, respiratory care, and selected developmental activities.

**Infancy**

In acute childhood SMA, weak or absent fetal movement during the last months of pregnancy is commonly reported by the mother. Significant weakness is present at birth or develops within the first 4 months, which manifests as inability to perform antigravity movements with the pelvic or shoulder girdle musculature and typical posturing in a gravity-dependent position (Fig. 15-8). The proximal musculature of the neck, trunk, and pelvic and shoulder girdles demonstrates the greatest weakness. Limited antigravity movement of distal upper and lower extremity musculature is present, and a positioning program in the newborn period or at the onset of symptoms is necessary. Use of wedges should be considered to avoid supine positioning in the presence of respiratory distress. If the supine position is used, rolled towels or bolsters are needed to keep the upper extremities positioned in midline and to prevent lower extremity abduction and external rotation. The side-lying position allows midline head and hand use for play without having to work against gravity. Prone positioning on wedges should be limited or not used, owing to the effort required for head righting to interact with the environment.

Respiratory care is a central focus of the habilitation program in acute childhood SMA. Children frequently require intubation for respiratory distress and tracheoscopy. Coordination with nurses and respiratory therapists on a program that includes suctioning, assisted coughing, and postural drainage is necessary. The use of supported sitting should be closely monitored for spinal alignment and respiratory response. Use of an elastic binder around the abdomen in sitting may be useful for children who demonstrate a marked reduction in oxygen saturation when seated.

ROM exercises should be carried out to ensure maintenance of flexibility and comfort. Flexion contractures of the hips, knees and elbows, hip abductors, ankle plantar flexors, and positional torticollis are deformities that can be avoided with a comprehensive ROM and positioning program (Binder, 1989). The exercise program should also include limited activities for strengthening, such as lightweight toys or rattles with Velcro straps around the wrists or mobiles positioned close to the hands for easy access.
The use of hammocks has also been advocated to provide the child with the opportunity for movement with only slight movements of the body (Eng, 1989a, 1989b). Developmental activities such as the use of supported sitting for the development of head control should be of short duration to avoid fatigue.

Head control fails to develop or is significantly impaired in acute childhood SMA. The child is unable to lift the head from a prone position to clear the airway. Early developmental postures such as prone on elbows are not attained. The use of developmental exercise in acute childhood SMA is controversial but should be considered if the child tolerates the activities, because a few children with chronic childhood SMA exhibit clinical signs of weakness in the first year of life and as a result may be misdiagnosed as having acute SMA.

In conjunction with an occupational therapist, a feeding program that is safe and not excessively exhausting should be implemented. Small, frequent feedings may be necessary, and breast feeding may be difficult (Eng, 1989b). Special care with feeding is necessary to avoid aspiration and secondary respiratory problems.

Although death secondary to pneumonia or other respiratory complications is typical within a few months to a few years in acute childhood SMA, the child’s death is usually not a struggle, owing to the degree of weakness and apnea (Eng, 1989b). The mean age of death is reported to be 6 months, with a range from 1 to 21 months reported by Merlini and associates (1989). However, with the use of ventilatory assistance, Bach and colleagues (2002) have reported survival up to 42 months. Counseling and support for the parents and family is an extremely important component in the management of these children.

**Chronic Childhood Spinal Muscular Atrophy (Type II)**

Impairments, Activity Limitations, and Participation Restrictions

The onset of significant weakness in chronic childhood SMA usually appears within the first year, with the course of the disease widely variable. Pearn and colleagues (1978) reported that of a cohort of 141 children, 95% demonstrated clinical signs before age 3 years. Forty-six percent never walked (even with orthotics), 38% were able to walk unaided at some stage, and the median age at death exceeded 10 years.
Eng (1989b) has reported three separate subgroupings within type II based on the pattern of presentation and progression. In the most severely involved group, the children never developed the ability to sit alone and respiratory capacity was significantly reduced. In the intermediate group, the children sat alone but never developed the ability to walk and demonstrated a regression of forced vital capacity to 45% by age 10 years. In the final group, independent walking was attained but half of the children lost this ability toward the end of the first decade. Interestingly, in the group of patients who remained ambulatory in the study by Eng (1989b), forced vital capacity was maintained at 90% as compared with 65% for those who lost independent walking during the first decade. The results of the study led Eng to conclude that forced vital capacity may be a physiologic predictor of walking duration.

Contractures are infrequently an impairment in chronic SMA. The distribution of weakness is similar to acute childhood SMA with primary proximal involvement, but to a much less severe degree. Weakness is usually greatest in the hip and knee extensors and trunk musculature. Involvement of the distal musculature appears later in the course of the disease and is less severe than the proximal involvement. Involvement of the cranial nerves has been reported but is not considered to be a typical feature of SMA other than in the acute childhood form. Fasciculations of the tongue have been reported in approximately one half of the children.

Infancy
Because the clinical presentation and progression of chronic childhood SMA are highly variable, the management program must address the major impairments, activity limitations, and participation restrictions as they are manifested. Approximately 15% of children have impairments within the first 3 months, and the remaining children have impairments by 18 months of age (Merlini et al., 1989). The program for the newborn with chronic SMA should be similar to that for children with acute SMA. Some children may develop the ability to stand, but few are able to use walking as a primary means of mobility.

Sitting posture is an area of primary concern in the management program with children who demonstrate significant weakness, requiring external head and trunk support in antigravity positions. A molded sitting support orthosis as shown in Figure 15-9 provides optimal contouring of the torso for support in sitting, or a thoracolumbosacral orthosis (TLSO, or “body jacket”) can also be used. Developmental activities provided on an ongoing basis are indicated to develop gross motor skills. Therapy sessions should be kept short to avoid fatigue and should emphasize selected developmental areas during each session because tolerance to handling in multiple positions is usually limited. Swimming has been reported to be beneficial in maintaining muscle strength and functional skills (Cunha, et al., 1996) Instruction to the family in the use of adaptive equipment for proper positioning is crucial in slowing the deforming effects of gravity on the spine when the child is sitting or standing.

If the child is not standing by the age of 16 to 18 months, adaptive equipment for standing should be considered. The rate of fracture in SMA has been reported to range from 12% to 15%, and weight bearing has been shown to decrease the frequency of lower extremity fractures (Ballestrazzi et al., 1989). A supine stander is recommended for children without adequate head control. Orthopedic consultation for a corset or TLSO should be considered for use in standing to maintain trunk alignment if the adaptive equipment does not provide adequate control.

Preschool-Age and School-Age Period
In the toddler, orthotics for standing might be considered (lightweight KAFOs); however, the progression of weakness may make walking an unrealistic goal. In a report of promotion of walking in 12 children with intermediate
SMA (ages 13 months to 3 years), Granata and associates (1989a) described success in attaining assisted ambulation, with 58% of the children using orthoses. Although only a small number of children were studied, these investigators also reported less severe scoliotic curves in the children who used the orthoses in comparison with a control group of children with SMA.

If a walking program is initiated, training in the parallel bars followed by use of a walker or other device to allow greater independence is desired. Close monitoring of safety with supported walking is necessary owing to the degree of weakness present and the potential for serious injury from a fall. The incidence of hip dislocation and contractures has also been reported as less when a supported walking program is used (Granata et al., 1989b).

Independence with mobility other than walking is a primary goal for the child who will not develop independence in walking or when walking is no longer possible (Jones et al., 2003). Because most power scooters do not provide adequate trunk control, use of a power wheelchair is indicated. If an orthopedic appliance is not used to support the trunk, close attention to fit is needed with use of lateral trunk supports and a trunk harness. Consideration should also be given to changing the side of the joystick every 6 months to avoid a pattern of leaning to one side. Prognosis for children with chronic childhood SMA is dependent on frequency and severity of pulmonary complications. Severe contractures as a result of prolonged sitting and progression of scoliosis are common, necessitating implementation of a consistent ROM program. Surgical intervention for spinal stabilization is an option if pulmonary function testing indicates a good prognosis for survival of the surgical intervention.

**Transition to Adulthood**

Survival into adulthood is extremely variable in chronic childhood SMA and depends on the progression of muscle weakness and secondary deformities. Because of the significant degree of muscle weakness, assistance is typically required for transfers and many ADL. An attendant or family member is needed to provide assistance for general ADL. Intelligence is rarely affected, and therefore vocational goals in areas of interest should be explored through vocational rehabilitation services.

An aggressive program of pulmonary care is required, including breathing exercises and postural drainage. Forced vital capacity has been shown to decrease about 1.1% per year, but mechanical ventilation is seldom needed (Steffensen et al., 2002). The ROM program should also be continued to control progression of the contractures unless a pattern of stability is recorded.

**Juvenile-Onset Spinal Muscular Atrophy (Type III)**

Juvenile-onset SMA may demonstrate symptoms of weakness within the first year of life in the proximal hip and shoulder girdle musculature, but more typically the onset is later in the first decade. Rarely are bulbar signs seen with the disease. Calf pseudohypertrophy is reported in approximately 10% of the cases. Fasciculations are noted in about half of the patients, and minimyoclonus may be a primary impairment noted on examination but rarely interferes with function (Brooke, 1986; Dorscher et al., 1991).

**Impairments, Activity Limitations, and Participation Restrictions**

In a study by Dorscher and colleagues (1991) reviewing the status of 31 patients with Kugelberg-Welander disease, proximal lower extremity weakness was the most common impairment reported. Secondary impairments included postural compensations resulting from the muscle weakness, contractures, and occasionally scoliosis. An increased lumbar lordosis and compensated Trendelenburg gait pattern are common postural compensations for proximal muscle weakness of the lower extremities. Ankle plantar flexion contractures are occasionally reported but not with the frequency seen in DMD, which aids differentiation of the two diseases. Scoliosis was reported in about 20% of patients by Dorscher and colleagues (1991) but was reported in all patients by other researchers (Granata et al., 1989b). In adolescents with type III SMA the incidence of scoliosis and its severity are related to the degree of weakness and functional status. Individuals who maintain independent walking have a lower incidence of scoliosis and less severe curves if scoliosis develops.

**School-Age Period**

A similar clinical presentation to DMD is seen in juvenile-onset SMA. The initial disability usually becomes apparent within the first decade and includes difficulty in arising from the floor, climbing stairs, and keeping up with peers during play. A waddling gait, which becomes more pronounced with attempts at running, will also be observed. Unlike DMD, no significant disability of upper extremity function is usually noted and proximal upper extremity strength is well preserved. Walking can usually be maintained lifelong as the primary means of ambulation. In those cases when weakness is noted before 2 years of age, however, a wheelchair or scooter may ultimately be required for mobility over long distances.

Management for the adolescent with juvenile-onset SMA is consistent with the concepts previously presented in this chapter. ROM exercises should be prescribed as
appropriate, and selected strengthening exercises may be indicated to maintain functional skills. Adaptive equipment for mobility is not usually indicated, but a power scooter for long-distance mobility may be needed in certain cases. If performance of ADL becomes a problem, collaboration with an occupational therapist to address concerns may also be needed.

Transition to Adulthood
Difficulty in ADL that requires lifting of moderately heavy objects overhead can be expected, and vocational activities that involve manual labor are not recommended. Because the life span is not significantly shortened, vocational planning is needed. No significant disability requiring adaptive equipment or environmental access is usually required until later in adulthood.

SUMMARY

Muscle weakness and contracture are hallmark features of the childhood forms of muscular dystrophy and spinal muscle atrophy. A background knowledge of therapeutic exercise, functional use of orthoses and adaptive equipment, and strategies to minimize disabilities secondary to these impairments allow the physical therapist to bring unique information and skills to the management team.

Many of the disorders significantly reduce longevity. Therefore, the patient’s quality of life and attention to how the family copes with the stress should be included in the team’s intervention program. Providing the children and families with support and realistic expectations is an ongoing challenge. Support groups or contact with another family that has had a similar experience can often help the family work through crisis periods, particularly when extended family support is not available.

Through the combined perspectives and innovative solutions of team members, a comprehensive program can be provided that takes into consideration the multifaceted demands of each individual and family. A philosophy of using a family-centered approach to care will help ensure that needs are met to the best of the team’s ability.

CASE STUDIES

Each of the two cases that are presented began before development of the Guide to Physical Therapist Practice (APTA, 2001). However, the reports are presented with reference to the guide to assist the practitioner in application of the Guide to clinical practice.

DONALD

Donald is from a family with six siblings (three brothers and three sisters). Three of the four boys were diagnosed with DMD. No family history of neuromuscular disease had been reported previously, and diagnosis followed medical examination of Donald’s older brother at age 5 years for clumsiness and frequent falls. Donald was 3 years of age at the time of diagnosis.

At the time of diagnosis Donald’s management program would be included in Musculoskeletal Practice Pattern C: Impaired Muscle Performance of the Guide to Physical Therapist Practice (APTA, 2001). He exhibited no significant gait deviations but had mild shoulder girdle and trunk flexor muscle weakness, evidence of a Gowers sign after the third attempt to rise to standing from the floor, and pseudohypertrophy of the posterior calf musculature.

A physical therapy examination at age 8 years revealed a gait pattern typical for DMD as previously described. No significant participation restrictions were noted, and impairments were only minimal. Donald was independent on stairs using a handrail but demonstrated a two-foot-per-step progression. Functional status corresponded to grade 2 on the scale published by Vignos and associates (1963) (see Box 15-1). ROM was within normal limits, with the exception of mild limitation of ankle dorsiflexion with the knee in extension. Muscle strength was quantified with manual muscle testing and recorded as fair plus in the shoulder and hip girdle musculature, poor in the abdominals, and good minus in the intermediate and distal upper and lower extremities. A home program was provided that included daily ROM of the posterior calf musculature and instruction on general activities to avoid excessive fatigue. Services were coordinated by the local MDA clinic with a follow-up visit every 6 months.

Donald’s initial disability was related to independent mobility with progressive loss of walking, which by age 12 involved inability to climb stairs and increased frequency of falls with attempts to walk on uneven surfaces. Furniture and walls were commonly used for balance. He was independent in scooting on the floor and used crawling for additional mobility at home. Although he was not able to stand from the middle of the floor, he was able to pull up to standing at a supportive surface. Night splints were initiated to augment the ROM program for the ankle plantar flexion contractures, and a manual wheelchair was provided for assistance with long-distance mobility. Muscle strength demonstrated progressive decline, with manual muscle testing measuring poor grades for the proximal hip and shoulder girdle musculature, fair plus grades for knee extension, and a Vignos scale rating of 5.
Mild tightness of the iliotibial band was present, and limitation of full hip and knee extension was noted. The ROM program was expanded to include the additional areas of tightness. With the progression of impairments beyond muscle function, a shift in Musculoskeletal Practice Pattern C to D: Impaired Joint Mobility, Muscle Function, Muscle Performance, and Range of Motion Associated with Capsular Restriction would be indicated.

By age 15, Donald was walking only short distances, and primarily for mobility within the home. A three-wheeled motorized scooter was provided for distance mobility, and Donald was independent in all transfers from the scooter. A raised toilet seat and tub bench were provided for the bathroom. He received adapted physical education and consultative physical therapy as a related service in the educational setting. He was followed through the MDA clinic, and Donald’s home program was augmented by a school program, including standing using a prone stander, ROM exercises three times per week, and adaptive physical education activities for general mobility and upper and lower extremity strengthening.

Progressive weakness and flexion contractures at the hip and knee resulted in the loss of walking when Donald was 17. It should be noted that this is exceptionally late for the loss of walking because 10 to 11 years is more typically reported (Brooke, 1986). Because Donald’s older brother had died following complications from a fracture resulting from a fall while wearing KAFOs, Donald and the family decided against continuation of a walking program using orthoses. A daily standing program at school was maintained until progression of the contractures resulted in a need to discontinue the program because orthopedic intervention was not desired by the family.

At age 26, Donald was at stage 8 on the Vignos Functional Rating Scale (see Box 15-1). He was living in an apartment with a full-time home health aide. The scoliosis that was documented 6 years earlier had not progressed, nor was any intervention required. Donald used a power wheelchair with joystick control as shown in Figure 15-10 owing to progression of upper extremity weakness and a need to provide greater trunk support in sitting. Assistance was required for all transfers, for bathing, and for dressing. A tub bench was used. Donald was independent with eating and personal hygiene such as brushing his teeth. He required assistance for bed mobility. The sliding board transfer demonstrated in Figure 15-11 became too difficult at age 23, requiring use of a manual lift or Hoyer for all transfers.

Donald assisted as an aide for an art teacher at a school for children with multiple disabilities (Fig. 15-12) until age 23, when his upper extremity weakness progressed to the point where he decided to stop working. He peace-
Fully passed away at the age of 28 while sleeping at home following hospitalization for a bout of pneumonia.

**DEREK**

Derek is 9 years, 3 months of age, with diagnosis of a dystrophinopathy and probable Duchenne muscular dystrophy. History includes an unremarkable pregnancy and term birth. Derek weighed 8 lb 13 oz at birth and went home within 3 days. He has an older sister.

Derek’s parents first began to have questions regarding his development at 17 months of age because of his delay in walking. His parents also reported what they perceived as clumsiness in his attempts to walk. Derek’s pediatrician did not share the parents’ concern regarding his delay or clumsiness in walking.

Following repeated expressions of concern by the family, Derek’s pediatrician referred him for physical therapy services at age 5 because he was not able to keep up with peers, especially in activities that required running, jumping, or balance. He was seen by a physical therapist in a hospital setting for coordination and strengthening exercises. After almost 6 months of intervention, the physical therapist recommended further diagnostic testing as manual muscle test scores demonstrated a decline in strength despite the strengthening exercises. He was subsequently referred to the local MDA-sponsored neuromuscular clinic for further examination.

At Derek’s initial visit to the neuromuscular clinic, his standing posture was found to be mildly lordotic, his gait exhibited increased lateral trunk sway toward the stance phase leg, he had difficulty hopping on one foot, he exhibited a positive Gowers sign when rising from the floor, and he needed to use a handrail when ascending or descending stairs. The physical examination found a pattern of mild proximal muscle weakness with most manual muscle test grades in the 4/5 range proximally and 5/5 distally. Creatine phosphate kinase testing resulted in a value of 20,009 IU/L (normal value 25–204 IU/L). DNA analysis found a deletion of exon 45 of the X chromosome, which resulted in a diagnosis of dystrophinopathy. A muscle biopsy to test for the presence or absence of dystrophin has not yet been done to definitively differentiate between Duchenne and Becker muscular dystrophy.

Prednisone was prescribed for Derek following the initial clinic visit with a dosage of 15 mg/day that was increased to 20 mg/day at the age of 9 years. Night splints (Fig. 15-13) and a home program for stretching of the posterior calf musculature were also initiated at the initial clinic visit with discontinuation of outpatient physical therapy services. His ankle dorsiflexion range of motion, which was at neutral at the initial visit, has improved to 10° with the knee extended and has remained stable through use of the night splints and stretching program. Improved muscle strength was recorded following the initiation of the prednisone regimen. Changes in muscle strength as documented with handheld dynamometry are shown in Table 15-3.

Currently, Derek is 9 years old and is in the fourth grade at his community elementary school. He receives consultative physical therapy at his school for input regarding his adapted physical education program and to address issues related to accessibility and fatigability during the school day. He is independently ambulatory with only mild gait deviations, which become more prominent when he attempts to run. He is independent in
stair climbing using a handrail and is able to transfer from the floor to standing using a Gowers maneuver. His home program of heelcord stretching (five repetitions of a standing runner’s stretch carried out daily (see Fig. 15-14), and night splint use are jointly monitored by the physical therapist at school and through the MDA clinic. At this point in time, he has no restriction on activity other than to avoid fatigue.

**ACKNOWLEDGMENTS**

My personal thanks goes to the children and their families who contributed to my knowledge of MD and SMA that made the writing of this chapter possible. Many challenges, accomplishments, disappointments, joys, and tears have paved the way. Special thanks to Donald and Derek and their families for sharing their stories, and to Becky, who first inspired me to take this path of clinical work.

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APPENDIX

Clinical Protocol for Functional Testing in Duchenne Muscular Dystrophy*

A. Pulmonary
   1. Forced vital capacity.
   2. Maximum voluntary ventilation.
B. Functional grade (arms and shoulders). Select one.
   1. Starting with arms at the sides, the patient can abduct the arms in a full circle until they touch above the head.
   2. Can raise arms above head only by flexing the elbow (i.e., shortening the circumference of the movement) or using accessory muscles.
If 1 or 2 is entered above, how many kilograms of weight can be placed on a shelf above eye level, using one hand?
   3. Cannot raise hands above head but can raise an 8 oz glass of water to mouth (using both hands if necessary).
   4. Can raise hands to mouth but cannot raise an 8 oz glass of water to mouth.
   5. Cannot raise hands to mouth but can use hands to hold pen or pick up pennies from the table.
   6. Cannot raise hands to mouth and has no useful function of hands.
C. Pulmonary.
   1. Maximum expiratory pressure.
D. Time to perform functions. Enter time in seconds. T = tried but failed to complete by time limit of 120 seconds.
   1. Standing from lying supine.
   2. Climbing four standard stairs (beginning and ending standing with arms at sides).
   3. Running or walking 30 feet (as fast as is compatible with safety).
   4. Standing from sitting on chair (chair height should allow feet to touch floor).
   5. Propelling a wheelchair 30 feet.
   6. Putting on a T-shirt (sitting in chair).
   7. Cutting a 3 \times 3\text{-inch} premarked square from a piece of paper with safety scissors (lines do not need to be followed precisely).
E. Functional grade (hips and legs). Select one.
   1. Walks and climbs stairs without assistance.
   2. Walks and climbs stairs with aid of railing.
   3. Walks and climbs stairs slowly with aid of railing (over 12 seconds for four standard stairs).
   4. Walks unassisted and raises from chair but cannot climb stairs.
   5. Walks unassisted but cannot rise from chair or climb stairs.
   6. Walks only with assistance or walks independently with long leg braces.
   7. Walks in long leg braces but requires assistance for balance.
   8. Stands in long leg braces but is unable to walk even with assistance.
  10. Is confined to bed.