

is damaged or the nature of the injury. The response is marked by vascular, chemical, and cellular events, with the ultimate purpose being to prepare the area for repair. The primary objective of the vascular response to injury is to mobilize and transport the body's defenses (white blood cells).

Vasoconstriction occurs initially along with reduced fluid flow through the injured area, resulting from development of fibrinogen clots in the tissue spaces and lymphatic channels (which prevents the spread of bacteria and toxins). Vasoconstriction allows for the white blood cells to migrate to the periphery of the vessel in a process called *margination* (see Chapter 6). These white blood cells eventually adhere to the walls of the damaged capillary, a process called *pavementing*.

Shortly after the injury and vasoconstriction, vasodilation of the local blood vessels occurs. The increased blood flow is accompanied by increased permeability of the small blood vessels. The permeability changes occur secondary to direct trauma to the vessels and to the presence of chemical mediators such as histamines, serotonin, and bradykinins.

The increased permeability allows for the white blood cells to squeeze through the blood vessel wall. This process is called *diapedesis* (see Fig. 6-12). The increased blood volume and vessel permeability also result in a significant transfer of fluid into the injured area. The fluid shift occurs because of the heightened intravascular hydrostatic pressure and the altered osmotic pressure gradient (as larger molecules escape into the tissues).

Once beyond the blood vessel walls the white blood cells are guided to the site of injury by a process called *chemotaxis* (see Fig. 6-12). Numerous elements of the damaged tissue (i.e., bacterial toxins and tissue polysaccharides) draw the white blood cells to the area of highest concentration of these elements.

Upon arriving at the site of damage, the white blood cells begin to clean up the area by the process of *phagocytosis*. The neutrophils, monocytes, and macrophages recognize, engulf, and digest debris, necrotic tissue, red blood cells, and proteins to prepare the area for repair and growth of new tissue.

The cardinal signs of acute inflammation are listed in Table 6-3. Accompanying clinical findings include increased muscle tone or spasm and loss of function. Movements of the involved area are generally slow and guarded. Cyriax describes two components of passive movement testing that also suggest acute inflammation: a spasm end feel and pain reported before resistance is noted by the practitioner as the limb is moved passively.<sup>29</sup>

If surgery is not indicated, the most effective interventions for acute inflammation are pharmacotherapy and physical therapy. Salicylates (except aspirin) and nonsteroidal antiinflammatory drugs (NSAIDs; see Chapter 5; see also Tables 5-1 and 5-2) are the most commonly administered medications for pain and inflammation.

The antiinflammatory effect is attained chiefly by inhibition of the biosynthesis of prostaglandins. Other antiinflammatory mechanisms include decreasing the release of chemical mediators from granulocytes, basophils, and mast cells; decreasing the sensitivity of vessels to brady-

kinin and histamine; and reversing or controlling the degree of vasodilation.

### SPECIAL IMPLICATIONS FOR THE THERAPIST

22-1

#### ***Biologic Response to Trauma***

See the section on Specific Tissue or Organ Repair, Chapter 6, and Special Implications for the Therapist: Specific Tissue or Organ Repair in Chapter 6.

## AGING AND THE MUSCULOSKELETAL SYSTEM

Much has been written about the effects of aging on the musculoskeletal system, especially in light of exercise as an effective intervention for so many diseases and conditions. Participation in a regular exercise program is an effective intervention to reduce or prevent a number of functional declines associated with aging.

Endurance training can help maintain and improve various aspects of cardiovascular function (as measured by maximal  $\text{Vo}_{\text{2}}$ , cardiac output, and arteriovenous oxygen difference) and enhance submaximal performance. Importantly, reductions in risk factors associated with disease states (e.g., heart disease, diabetes) improve health status and contribute to an increase in life expectancy.<sup>9,10</sup>

Strength training helps offset the loss in muscle mass and strength typically associated with normal aging. Additional benefits from regular exercise include improved bone health and therefore reduction in risk for osteoporosis; improved postural stability, thereby reducing the risk of falling and associated injuries and fractures; and increased flexibility and range of motion.

Although not as abundant, evidence also suggests that involvement in regular exercise also can provide a number of psychologic benefits related to preserved cognitive function, alleviation of depressive symptoms and behavior, and an improved concept of personal control and self-determination.<sup>9,10</sup>

Sports-related injuries among people born between 1946 and 1964, now referred to as "boomeritis,"<sup>7</sup> are on the increase as older adults continue to participate actively in sports of all kinds. Physical therapists can provide valuable preventive education regarding the aging process as it relates to the musculoskeletal system and exercise. This presentation is a brief summary of the findings to date; more in-depth discussion is available.<sup>34,50,70,91</sup>

### Muscle

#### **Sarcopenia**

**Overview and Definition.** Age-related loss in muscle mass, strength, and endurance accompanied by changes in the metabolic quality of skeletal muscle is termed *sarcopenia*. Sarcopenia involves both the reduction of muscle mass and/or function as well as the impairment of the muscle's capacity to regenerate.<sup>35</sup>

Muscle mass is lost at a rate of 4% to 6% per decade starting at age 40 in women and age 60 in men.<sup>54</sup> The greatest decline in both men and women occurs with inactivity, acute illness, and after age 70, at which time the mean loss of muscle mass has been measured as 1% per year.<sup>54</sup> At all ages, females appear to be more vulnerable to loss of lean tissue than males; however, in men and women, muscle strength can be maintained through exercise well into the eighth decade.<sup>43,58</sup>

**Etiology.** The etiology is multifactorial, involving changes in muscle metabolism, endocrine changes (e.g., low testosterone levels), nutritional factors, and mitochondrial and genetic factors.<sup>69,73</sup> It remains uncertain how much age-related loss of muscle function is an inevitable consequence of aging, nutritional status, or dysregulation of neurologic, hormonal, and/or immunologic homeostasis.

Likewise, it remains unknown how much sarcopenia reflects a decline in physical activity and exercise capacity, and as part of a broad cycle, whether this decline is a function of age, lack of motivation, decline in neuromuscular function from disuse or loss of motoneurons, age-associated decreases in metabolism, or other factors such as anemia or high levels of inflammatory markers.<sup>23,95</sup>

**Pathogenesis.** The identification of the molecular chain able to reverse sarcopenia is a major goal of studies on human aging. Animal studies suggest that myofiber regeneration in sarcopenic muscle is halted at the point where reinnervation is critical for the final differentiation into mature myofibers.

Combined evidence points to a decreased capacity among motoneurons to innervate regenerating fibers. There are also changes observed in the expression of several cytokines known to play important roles in establishing and maintaining neuromuscular connectivity during development and adulthood.<sup>38</sup>

The decline in muscle mass previously thought to be the result of proteins' breaking down faster than they were being created and restored may be linked instead to other potential reasons such as diet and nutrition, the body's ability to use protein from food, and hormonal changes.<sup>53</sup> For example, inadequate dietary intake of protein also results in loss of skeletal muscle mass; the current recommended daily allowance (RDA) may not be adequate to completely meet the metabolic and physiologic needs of virtually all older people.<sup>21</sup>

Loss of muscle function appears to be due to decreased total fibers, decreased muscle fiber size, impaired excitation-contraction coupling mechanism, or decreased high-threshold motor units. For example, at midthigh, muscle accounts for 90% of the cross-sectional area in young, active adults. However, this same measurement taken in older adults is only 30%.<sup>46</sup>

Additionally, selective loss of motor unit number or atrophy (particularly after 70 years) of fast twitch (type IIa) muscle fibers<sup>98</sup> occurs. Some researchers suggest that no preferential loss of type I or type II muscle fibers occurs with age, but rather, both types are equally affected and type II fiber cross-sectional area is reduced, which accounts for the significant decrease in muscle strength.<sup>1</sup>

Other studies have shown that type II fibers are preferentially affected by aging and that fiber II atrophy is

associated with a decline in satellite cells (essential for skeletal muscle growth and repair).<sup>93</sup>

Other researchers hypothesize an extrinsic apoptotic pathway to explain how type II fiber-containing skeletal muscles may be more susceptible to muscle mass losses.<sup>74</sup> Several signaling pathways of skeletal muscle apoptosis are currently under intense investigation, with particular focus on the role played by mitochondria.<sup>65</sup> However it occurs, the clinical significance of this loss is that it leads to diminished strength and exercise capacity.<sup>90</sup>

**Effects of Sarcopenia.** From a clinical perspective, loss in muscle mass accounts for the age-associated decreases in basal metabolic rate contributing to metabolic disorders such as type 2 diabetes mellitus and osteoporosis and decreases in muscle strength and activity levels, which, in turn, are the cause of the decreased energy requirements of the aging adult.<sup>41</sup>

Loss of muscle mass (i.e., atrophy) and definition and loss of muscle function resulting in subsequent muscle weakness are implicated in difficulty accomplishing activities of daily living (e.g., rising from a chair, climbing stairs, carrying groceries), slow gait speed, impaired balance reactions, and increased risk of vertebral compression (and other) fractures. There does not appear to be a relationship between age-related sarcopenia and the bone mass loss also prevalent in the same age group.<sup>25</sup>

Aging workers notice increasing difficulty continuing a job they have previously performed without trouble. Slowing down of reflexes and coordination combined with loss of muscle mass and strength can make it difficult to remain in the same job or train for a new job.

By age 65, changes in the muscle mass, muscle weakness, and decreased levels of physical activity are evident in the increased numbers of falls and injuries. Injuries in an aging musculoskeletal system take longer to recover, contributing to further physical deconditioning, potentially creating additional comorbidities.

**Exercise and Sarcopenia.** (See also the Musculoskeletal System and Exercise in this chapter). Appropriate exercise can alter, slow, or even partially reverse some of the age-related physiologic changes that occur in skeletal muscle, including sarcopenia. Skeletal muscle adaptations in response to strength training in older adults occur with progressive resistance training (PRT) or high-intensity training (e.g., two to six sets of eight repetitions at approximately 80% of the person's one-repetition maximum).<sup>97</sup>

Understanding muscle fiber types and the impact of physical therapy interventions on muscle fiber type conversions is becoming increasingly important in today's evidence-based practice. An excellent review of these concepts is available.<sup>79</sup> We know, for example, that age-related changes can be counteracted and physical function improved by increased physical activity of a resistive nature.<sup>87</sup> Mechanical load on muscle can increase the cross-sectional area of the remaining fibers but does not restore fiber numbers characteristic of young muscle.<sup>1</sup>

Strength training has been shown to improve insulin-stimulated glucose uptake both in healthy older adults and in individuals with diabetes. Strength training also improves muscle strength in healthy adults and in those who have chronic diseases. Increased strength leads to

improved function and a decreased risk for falls, injuries, and fractures.<sup>32</sup> These results also promote increased independence and improved quality of life.<sup>101</sup>

Aging muscle may be resistant to insulin-like growth factor I (IGF-I); IGF-I promotes myoblast proliferation, differentiation, and protein assimilation in muscle through multiple signaling mechanisms. Exercise may be able to help aging muscle that is resistant to IGF-I by reversing this effect.<sup>1</sup>

High-resistance training exercise has been of significant benefit to sarcopenia.<sup>69</sup> In fact, after 6 months of exercise training, resistance exercise has been shown to reverse mitochondrial dysfunction for genes that are affected by both age and exercise.<sup>68</sup> Combinations of resistance exercise, aerobic exercise, and stretching have shown beneficial effects on sarcopenia, but the optimum regime for older adults remains unclear.<sup>49,60</sup>

Many older adults would like to be more physically active but do not have the experience or knowledge to develop and build up an exercise regimen without appropriate supervision such as the physical therapist can offer. Others have participated in athletics throughout adulthood and continue to train and remain in good health. The therapist can help educate older adults about the importance of maintaining strength training and endurance with the emphasis on strength, which decreases more rapidly than endurance.<sup>47</sup>

## Joint and Connective Tissue

At the same time as changes in bone and muscle are taking place, a progressive loss of flexibility and changes in connective tissue starts contributing to an increased incidence of joint problems beginning in middle age and progressing through old age. Loss of flexibility also contributes to increased risk of falls and other injuries. Connective or periarticular tissue, including fascia, articular cartilage, ligaments, and tendons, becomes less extensible with resultant decreased active and passive range of motion.

### Increased Stiffness and Decreased Flexibility

It is not clear whether this decreased flexibility occurs as a consequence of biologic aging, inactivity, degenerative disease, adhesion molecules, or a combination of all these factors.<sup>98</sup> One possible cause is related to fibrinogen, produced in the liver and converted to fibrin, which constantly circulates throughout the body to serve as a clotting mechanism (with superglue-like effects) should an injury occur.

Fibrinogen normally leaks out of the vasculature in small amounts into the intracellular space and then adheres to cellular structures, causing microfibrinous adhesions among the cells. Activity and movement normally break down these adhesions along with macrophagic activity to dissolve unused fibrinogen and fibrin.

In the aging process, less fibrinogen and fewer (less efficient) macrophages are available. These factors, along with less physical activity and movement, allow these microadhesions to accumulate in muscle and fascia, resulting in an increased sense of overall stiffness.

Others have shown that aging collagen has increased cross-links between molecules, increasing the mechanical stability of collagen but also contributing to increased tissue stiffness.<sup>76</sup> Increased collagen content in the endomysium of animal intramuscular connective tissue has been shown to correlate with increased stiffness of the whole muscle.<sup>4</sup>

Regardless of the exact physiologic mechanism for the gradual increase in stiffness associated with aging, physical activity has an important influence in alleviating stiffness. Further research is needed to understand how and what kind of physical activity influences or possibly prevents stiffness.<sup>70,99</sup>

### Changes in Articular Cartilage

Articular cartilage, which cushions the subchondral bone and provides a low-friction surface necessary for free movement, contains few cells, is aneural and avascular, and often starts to break down with increasing age.<sup>34</sup> The main proteoglycan in articular cartilage (aggrecan) binds with hyaluronan to form massive aggregates that expand the collagen matrix of the tissue to provide it with its compressive and tensile strength.

With age, proteoglycan aggregation is reduced and smaller proteoglycans are synthesized with an increase in keratin sulfate and reduced chondroitin sulfate content. The hydrophilic proteoglycans have been shown to become shorter in aged tissue and therefore lose their ability to hold water in the matrix.<sup>20</sup> Dehydrated articular cartilage may have a reduced ability to dissipate forces across the joint.<sup>70</sup>

Degeneration and thinning or damage of articular cartilage with loss of water content contribute to a significant increase in incidence of osteoarthritis with aging. By age 60, as much as 80% of the population shows evidence of such, although only about 15% present with symptoms.<sup>98</sup>

Knowledge of these changes has resulted in new interventions such as glucosamine-chondroitin supplementation and joint viscosupplementation for osteoarthritis (see Chapter 27). With or without a symptomatic presentation, educating adults about the importance of joint protection is an important role of the physical therapist.

### Tendons

Tendons exhibit a lower metabolic activity associated with aging that has implications for injury and healing in the aging population.<sup>2,3</sup> Also an age-related decrease occurs in the tensile strength of some tendons and ligament-bone interfaces, and loss of integrity of some joint capsules occurs. For example, rotator cuff impairment with loss of joint function is common in older people. A gradual loss of connective tissue resistance to calcium crystal formation occurs in the older adult, leading to an increase in the incidence of crystal-related arthropathies<sup>34</sup> (e.g., gout, pseudogout; see Chapter 27).

### Proprioception

Joint proprioception, described as sensations generated to increase awareness of joint orientation at rest and in motion, declines with age, especially in the knee and

ankle.<sup>72</sup> Joint proprioception provides both a sense of joint position and sense of joint movement. Mechanoreceptors located in the joint capsules, ligaments, muscles, tendons, and skin provide the sensory information needed for a sense of joint position.

The presence of osteoarthritis seems to make joint proprioception even worse, though it is unclear whether impaired joint sense promotes arthritic change or whether arthritic change causes the sensory loss. There is some evidence that proprioception may be reduced before the development of joint degenerative change.<sup>59</sup>

## Bone

The skeletal system serves numerous functions in the human body throughout the lifespan. Bone is the primary storage depot for calcium, phosphate, sodium, and magnesium. Bones are the hosts for the hemopoietic bone marrow (growth and development of elements of blood). Bones also serve important mechanical functions, such as protection of components of the nervous system and visceral organs; provision of rigid internal support for the trunk and extremities; and provision of attachment sites for numerous soft tissue structures.

Bone is remodeling constantly throughout life. While osteoclasts resorb the existing bone, new bone is being formed by osteoblasts. Three primary influences affect this remodeling process: (1) mechanical stresses; (2) calcium and phosphate levels in the extracellular fluid; and (3) hormonal levels of parathyroid hormone, calcitonin, vitamin D, Cortisol, growth hormone, thyroid hormone, and sex hormones.

Aging adversely affects the "quality" of human bone material, both the stiffness and strength of bone and its "toughness." These effects are caused by factors such as architectural changes, compositional changes, physicochemical changes, changes at the micromechanical level, and the degree of prior *in vivo* microdamage.<sup>102</sup>

The bone density of the skeleton reaches its peak during an adult's twenties and remains stable for about two decades. Around the time of menopause for women, resorption, the process by which bone is broken down and calcium is released from the bone for use by the body, increases, while formation, the bone-rebuilding process, fails to keep pace. This imbalance, which is triggered by declining estrogen levels, leads to rapid bone loss during the first decade after menopause, with moderate bone loss thereafter. In women with low peak bone mass, it can result in osteoporosis with the increased potential for vertebral, hip, or other fracture.<sup>51</sup>

The same progressive decrease of calcium can occur in men, only at a reduced and slowed rate. In women, loss occurs at a rate of approximately 1% per year after age 35 with acceleration especially during the first 5 years after menopause. Men lose 10% to 15% by age 70 years and 20% by age 80.

In women the loss is greater, amounting to about 20% by age 65 and 30% by age 80.<sup>5,84</sup> In both genders, by age 65, bone loss generally has progressed to a point where the older adult is predisposed to fractures, especially when other comorbidities exist (e.g., diabetes,

balance or vestibular impairment, renal impairment, immobilization).<sup>98</sup>

## THE MUSCULOSKELETAL SYSTEM AND EXERCISE

By the year 2030, 70 million people in the United States will be 65 years or older; people 85 years and older will be the fastest growing segment of the population. As more individuals live longer, the importance of exercise and physical activity to improve health, functional capacity, quality of life, and independence will increase in this country.<sup>9,10</sup>

Strength training is considered a promising intervention for reversing the loss of muscle function and the deterioration of muscle structure that is associated with advanced age. The capacity of older men and women to adapt to increased levels of physical activity is preserved, even in the most aged adult.<sup>19,92</sup>

For example, the relationship of exercise to insulin action is important because increased body fat (especially abdominal obesity) and decreased exercise is linked to the increased incidence of diabetes in the aging population.

Regularly performed exercise can affect nutritional needs and functional capacity in the older adult, contributing a preventive effect.<sup>38</sup> Combining knowledge of exercise principles with nutrition is important for all people but especially in the older adult population, disabled individuals, athletes, adolescents, and anyone with a medical condition, disease, or illness.<sup>100</sup>

## Muscle

Human muscles contain two different types of muscle fibers based on speeds of shortening and morphologic differences. Type I muscle fibers, known also as *slow oxidative slow twitch fibers*, are the fatigue-resistant red fibers. The red color is the result of high amounts of myoglobin and a high capillary content. Greater myoglobin and capillary content contributes to the increased oxidative capacity of red muscles compared to white muscles (type II).<sup>79</sup>

Type II fibers, or fast twitch fibers, have two different characteristics. Type IIa, which are bigger and faster than type I, are also fatigue resistant and are referred to as *fast oxidative fibers*. Type IIb fibers are the classic white fibers, which lack aerobic enzymes and therefore fatigue rapidly. Each muscle contains type I and type II fibers in various proportions.

The basic distribution of fiber types is thought to be an inherited characteristic. Although distribution varies among individuals, the average ratio of fast to slow twitch fibers is 50:50. Individuals trained in endurance activities usually have a higher proportion of slow twitch fibers, and those trained for high-intensity, high-speed activities have more high twitch fibers. The oxidative capacity of both fibers can be increased greatly by endurance training, but the glycolic capacity and contractile properties are not modified.<sup>81</sup>

Muscle function can be described in terms of strength and endurance, which is also how we focus training of muscle. Strength can be defined in several ways depending on the specific method of measurement but is usually related to the diameter of the muscle fiber, which has been consistently shown to increase with strength training.

Endurance can be measured as the ability to work over time; local muscle endurance is distinguished from general body endurance as the ability of an isolated muscle group to continue a prescribed task rather than the ability to continue an activity such as running, swimming, or jogging for an extended period of time.<sup>81</sup>

As a result of specificity of training and the need for maintaining muscular strength and endurance, and flexibility of the major muscle groups, a well-rounded training program including aerobic and resistance (strength and endurance) training and flexibility exercises is recommended.

### Strength Training

Strength training refers to exercise directed at improving the maximum force-generating capacity of muscle. There is evidence that strength training has a positive effect on aging skeletal muscle.<sup>97</sup>

Collectively, studies indicate that strength training in the older adult (1) produces substantial increases in the strength, mass, power, and quality of skeletal muscle; (2) can increase endurance performance; (3) normalizes blood pressure in those with high-normal values; (4) reduces insulin resistance; (5) decreases both total and intraabdominal fat; (6) increases resting metabolic rate in older men; (7) prevents the loss of bone mineral density with age; (8) reduces risk factors for falls; and (9) may reduce pain and improve function in those with osteoarthritis in the knee.

Significant strength gains are possible in all populations, including older adults, when exposed to an adequate strength training program. Strength gains occur from enhanced neuromuscular activation over the initial 8 weeks and from increased fiber density and hypertrophy during subsequent weeks.<sup>62</sup>

Considerable evidence exists that sarcopenia can be prevented, reduced, and reversed with prescriptive strength training programs that emphasize gradual, progressive, high-intensity resistance exercises (e.g., high load/low repetition) for the upper and lower extremities.<sup>13,77,79</sup>

Resistance training significantly increases muscle size and increases energy requirements and insulin action in adults over age 65 years. A program of once- or twice-weekly resistance exercise (carried out at a level described as "reasonably difficult" or "difficult") achieves muscle strength gains similar to 3 days/wk training in older adults and is associated with improved neuromuscular performance.<sup>88</sup> The goal is to design a program for each individual to provide the proper amount of physical activity and exercise to attain maximal benefit at the lowest risk.<sup>9,10</sup>

Strength training does not increase maximal oxygen uptake beyond normal (i.e., individuals attain the same maximal  $\text{Vo}_2$  before and after training).<sup>66,67</sup> In postmeno-

pausal women, muscle performance, muscle mass, and muscle composition are improved by hormone replacement therapy (HRT). The beneficial effects of HRT combined with high-impact physical training appear to exceed those of HRT alone.<sup>82,83,86</sup> Long-term results remain under investigation.

### Endurance Training

Endurance training refers to exercise directed at improving stamina (the duration that a person can maintain strenuous activity) and aerobic capacity ( $\text{Vo}_{\text{max}}$ ). Endurance training places a high metabolic demand on the muscle and will increase the oxidative capacity of all muscle fiber types.<sup>79</sup>

Endurance exercise can reverse the decline in physical conditioning associated with aging. An endurance training program using relatively modest intensity of training can reverse 100% of the loss of cardiovascular capacity, returning some healthy older adults to levels of aerobic power present in young adulthood. Even an older person who has failed to maintain fitness over time can benefit from an exercise program.<sup>66,67</sup>

In middle-aged adults, the mechanism responsible for decline in cardiovascular capacity appears to be a reduced plasticity of heart muscle; improved aerobic power after training appears to be directly related to peripheral oxygen extraction (i.e., the muscles' ability to take up and use oxygen).

Aerobic exercise results in improvements in functional capacity and reduced risk of developing type 2 diabetes mellitus in the older adult. Aerobic endurance training for fewer than 2 days/wk at less than 40% to 50%  $\text{Vo}_2$  and for less than 10 minutes is generally not a sufficient stimulus for developing and maintaining cardiovascular fitness in healthy adults.

### Joint

As discussed earlier, tendons, ligaments, and muscles around the joints have less water content, resulting in increased stiffness, with increasing age. Articular cartilage has less tensile strength and biochemical composition changes, often leading to osteoarthritis.<sup>75</sup>

Joint changes with deterioration of subchondral bone and atrophy of the synovium also can occur. Well-regulated exercise does not produce or exacerbate joint symptoms and actually may improve symptoms.<sup>26</sup> This concept is discussed in greater detail in the section on osteoarthritis (see Chapter 27).

### Bone

The relationship between bone mass and activity is well established. Complete immobilization and weightlessness result in rapid onset of accelerated bone resorption; bone mass recovers when activity resumes, but whether bone loss is completely reversible is unknown. Immobilization also leads to changes in collagen, ligaments, and the musculotendinous junction at the joint, causing reduced range of motion.

Osteopenia, osteomalacia, and osteoporosis affect the mineralization of bone matrix and can impact the bone

health of the aging adult. Older adults are at greater risk for osteoporosis-related fractures, both age related for all adults and postmenopause related for women. Fractures are discussed in more detail in Chapter 27.

### Specific Exercise Guidelines

Resistance training is an integral component in the comprehensive health program promoted by major health organizations such as the American Heart Association, American College of Sports Medicine, Surgeon General's office, and others. Population-specific guidelines have been published, and the current research indicates that for healthy people of all ages and many people with chronic diseases, single-set programs of up to 15 repetitions performed a minimum of twice per week are recommended.

Each workout session should consist of 8 to 10 different exercises that train the major muscle groups. Single-set programs are less time consuming and generally result in greater compliance. The goal of this type of program is to develop and maintain a significant amount of muscle mass, endurance, and strength to contribute to overall fitness and health.

Although age in itself is not a limiting factor to exercise training, a more gradual approach in applying prescriptive exercise at older ages may be necessary because exercise programs also can cause injury, especially in the presence of comorbidities such as arthritis, obesity, neurologic disease, postural instability, cardiovascular impairment, previous joint injuries, joint deformity, or other musculoskeletal complications, such as tendonitis or shoulder impingement syndrome. High-intensity resistance training (above 60% of the one-repetition maximum) has been demonstrated to cause large increases in strength in the older adult (older than 65 years).<sup>17,40</sup>

People with chronic diseases may have to limit range of motion for some exercises and use lighter weights with more repetitions.<sup>42,52</sup> Otherwise, older adults do not have to "take it easy" when performing exercise. The presence of heart disease, diabetes, cancer, or other comorbidities may require some initial progression in the prescribed program.<sup>97</sup>

Overall, therapists should pay careful attention to finding exercise intensities that are optimally suited to induce the desired training effects. The skeletal muscle of older people is more easily damaged with the loading that occurs during training compared with the skeletal muscle of younger adults. Care should be taken to monitor soreness and prevent muscle injuries after exercise.<sup>97</sup>

Recommendations for the quantity and quality of exercise for developing and maintaining cardiorespiratory and muscular fitness and flexibility in healthy adults also have been published. A certain combination of *frequency* (3 to 5 days/wk), *intensity* (55% or 65% to 90% of maximum heart rate or 40% or 50% to 85% of  $\text{V}\text{O}_{\text{2max}}$ ), and *duration* (20 to 60 minutes continuously or 10-minute bouts intermittently throughout the day) of exercise performed consistently over time has been found effective for producing a training effect.<sup>9,10</sup>

Fatigue, the inability to continue to maintain a given activity, may develop as a result of depletion of muscle and liver glycogen, decreases in blood glucose, dehydration, and increases in body temperature. In a strength training program for adults over 65 years, repeated maximum voluntary contractions resulting in fatigue may differ from those for younger populations. This may be relevant for designing optimal strength training programs for older adults specifically requiring closer supervision to ensure that each repetition is completed without substitution or incomplete range of motion and to adjust rest times between contractions. Alternatively, electrical stimulation may provide more consistent muscle activation during strength training in this age group.<sup>85</sup>

Exercise guidelines for the very old (older than 85 years) also have been published by the American College of Sports Medicine as follows: *frequency* of at least 2 days/wk, preferably 3 days; *intensity* of 40% to 60% of heart rate reserve; *duration* of at least 20 minutes. Walking, leg/arm ergometry, seated stepping machines, and water exercises are recommended.

Additional recommendations for resistance training include two to three sets of 8 to 12 repetitions performed with good form and through the entire range of motion for each exercise performed on each training day (one set may be sufficient); some standing postures with free weights and balance training should be included.<sup>9,10</sup> When 12 repetitions can be completed without difficulty (observe for increased respiration, extremity tremors, facial grimacing), the weight can be increased by 5% with a lower number of repetitions to begin a new training cycle.

## MUSCULOSKELETAL SYSTEM DISEASE

Although not nearly as common as traumatic and repetitive or overuse injuries, musculoskeletal system diseases are significant from the standpoint of disability, mortality, and cost in terms of health care dollars. The most serious of these diseases are cancer and infection. The pathogenesis of these two types of diseases illustrates the intricate interrelationships between the musculoskeletal system and other body systems.

The primary highways or communication networks connecting the musculoskeletal and other body systems are the circulatory and lymphatic systems. These pathways are the routes utilized by disease to travel from one system to another. In addition, these highways deliver the nutrients and other supplies needed by the musculoskeletal system.

### Cancer

Although primary malignant bone and soft tissue tumors are rare, metastatic disease of the musculoskeletal system is relatively common. Bone is one of the three most favored sites of solid tumor metastasis, indicating that the bone microenvironment provides fertile ground for the growth of many tumors. Although lung, breast, and prostate are the three primary sites responsible for most metastatic bone disease, tumors of the thyroid and kidney,

lymphoma, and melanoma also can metastasize to the skeletal system.

Cancer cells typically invade the thin-walled lymphatic channels, capillaries, and venules as opposed to the thicker-walled arterioles and arteries. Once the cancer cells enter the bloodstream, they must lodge in the vascular network of the host tissue before the secondary cancer can develop.

Organs with extensive circulatory or lymphatic systems, like the lungs and liver, are the most common sites of metastasis. Of the other potential sites of metastasis, the axial skeleton is among the most common. The blood supply to the axial skeleton is extensive compared with that to the distal components of the extremities, and the spinal blood flow through the thin-walled, valveless veins is slow and sluggish. This gives the circulating cancer cells ample opportunity to attach to the vessels' endothelia. The bony thorax, lumbar spine, and pelvis are the most common components of the axial skeleton for seeding of cancer to occur, and the vertebral bodies, because of the extensive venous plexus, appear to be the initial site for development of disease.

As with primary bone tumors, the major manifestation of metastatic bone cancer is pain, especially on weight bearing and at night. The pain can be caused by stretching of the periosteum or irritation of a nerve root or spinal cord, or can be secondary to bone collapse (pathologic fracture).

Once the cancer begins to spread, clients report fatigue, malaise, fever, nausea, and other symptoms. Therapists working with clients diagnosed with cancer must be vigilant for symptoms or signs suggestive of systemic compromise and be aware of common sites of metastasis for the particular primary tumor.

An awareness of signs and symptoms associated with the potential target organs is important, and any suspicious findings should be reported to the physician. Unfortunately, often the initial presenting symptom associated with the disease is pain from the bone metastasis (back pain), which can result in a delay in the diagnosis. See Chapters 9 and 26 for extensive information related to cancer.

## Infection

As with cancer, infection can originate in the musculoskeletal system or it can spread to the musculoskeletal system from elsewhere in the body. The most common cause of osteomyelitis is direct extension of bacterial organisms by penetrating wounds, fractures, or surgical intervention. *Staphylococci* and *streptococci* are the most common infecting agents (see Chapter 8).

The other common mechanism by which bacterial organisms reach the musculoskeletal system is via the

hematogenous route. The original infection could be of the urinary tract (adults) or skin or teeth (children). In adults, the most common site of osteomyelitis is the vertebral body or intervertebral disc. The sluggish blood flow through the valveless veins facilitates bacterial seeding. Cases have been described illustrating the lengthy delay in diagnosis of vertebral osteomyelitis when back pain is the primary presentation (see the section on Osteomyelitis in Chapter 25).

Back pain can occur as a symptom of many diseases. Anyone with back pain of nontraumatic or unknown origin must be screened for medical disease, especially possible gastrointestinal or abdominal involvement related to infections (e.g., diverticulitis, appendicitis, pelvic inflammatory disease, Crohn's disease).

If infection occurs and penetrates the pelvic floor or retroperitoneal tissues (i.e., those organs outside the peritoneum such as the kidneys, colon, and bladder), abscesses may result in isolated referred hip or thigh pain and antalgic gait.

A variety of objective test procedures may be employed by the therapist to assess for iliopsoas abscess formation, including the pinch-an-inch test (see Fig. 16-24), the iliopsoas muscle test, the obturator test, and palpation of the iliopsoas muscle (see Figs. 16-14 and 16-15).

Approximately 25% of all clients with inflammatory bowel disease (IBD; e.g., Crohn's disease, ulcerative colitis, diverticulitis) may present with migratory arthralgias, monarthritis, polyarthritis, or sacroiliitis. The joint problems and gastrointestinal disorders may appear simultaneously, the joint problems may manifest first (sometimes even years before bowel symptoms), or intestinal symptoms may present along with articular symptoms but be disregarded as part of the whole picture by the client.

Any time a client presents with low back, hip, or sacroiliac pain of unknown origin, the therapist must screen for medical disease by asking a few simple questions about the presence of accompanying intestinal symptoms, known personal or family history for IBD, and possible relief of symptoms after passing stool or gas.<sup>48</sup>

Joint problems usually respond to treatment of the underlying bowel disease but in some cases require separate management. Interventions for the musculoskeletal involvement follows the usual protocols for each area affected.

## References

To enhance this text and add value for the reader, all references are included on the companion Evolve site that accompanies this textbook. The reader can view the reference source and access it on-line whenever possible. There are a total of 102 cited references and other general references for this chapter.

# CHAPTER 23

## Genetic and Developmental Disorders

ALLAN GLANZMAN

Pediatric diseases and disorders comprise a large number of conditions. Entire volumes have been devoted just to pediatric pathologies. Given the format of this book and space limitations, in this chapter we have included as many of the more commonly encountered genetic and developmental disorders as possible. Cerebral palsy is discussed separately as a neurologic condition in Chapter 35.

A brief discussion of several other rare but important diagnoses also is included. Because physical and occupational therapy intervention is not the focus here, the reader is referred to other, more appropriate resources for specific and thorough intervention guidelines for these conditions.<sup>26,94,142,163,171</sup>

### DOWN SYNDROME

#### Definition and Incidence

Down syndrome was the first genetic disorder attributed to a chromosomal aberration and is referred to as trisomy 21 (also Down's syndrome). Down syndrome is characterized by muscle hypotonia, cognitive delay, abnormal facial features, and other distinctive physical abnormalities.

Down syndrome is the most common inherited chromosomal disorder, occurring once in every 700 live births. The incidence of Down syndrome rises with maternal age. Before maternal age 30 the incidence is 1 in 2000 births; it is 1 in 50 for mothers aged 35 to 39, and 1 in 20 for mothers over 40 years. There is a 2% risk of recurrence for a couple who have had a child with Down syndrome.

#### Etiologic Factors and Pathogenesis

The actual cause of Down syndrome is not yet known; however, some hints have begun to emerge as researchers continue to explore gene mapping and develop genetic models for Down syndrome.<sup>9</sup> Evidence from cytogenetic and epidemiologic studies supports multiple causality.

Trisomy 21 produces three copies of chromosome 21 instead of the normal two because of faulty meiosis (cell division by which reproductive cells are formed) of the ovum or, sometimes, the sperm. This results in a karyotype (chromosomal constitution of the cell nucleus) of 47 chromosomes instead of the normal 46.

The faulty cell division can also occur after fertilization, leading to only a portion of cells being affected, with a milder clinical picture. This situation is referred to as mosaicism. Because of the positive correlation between increasing age and Down syndrome, it is hypothesized that deterioration of the oocyte (immature ovum) or environmental factors such as radiation and viruses may cause a predisposition to mistakes in meiosis and the resulting chromosomal abnormality.

In a small number of cases Down syndrome occurs as a result of a translocation of chromosome 15, 21, or 22 (i.e., the long arm of the chromosome breaks off and attaches to another chromosome). Chromosomal translocation can be hereditary or associated with advanced parental age. Although Down syndrome usually is attributed to the aging woman, evidence suggests that in 5% to 10% of cases Down syndrome is correlated with paternal age.<sup>157,195</sup>

The third copy of chromosome 21 is the cause of the phenotypic characteristics that are observed in people with Down syndrome. There are presumably many genes that are present in triplicate in individuals with Down syndrome. Only a few of these have been identified as causative of specific pathology in Down syndrome; most likely there are many more that will be identified in the future.

Alzheimer's disease is also more common in people with Down syndrome, occurring at an earlier age than that of the Alzheimer population in general. By the age of 40 symptoms of Alzheimer can be seen in almost everyone with Down syndrome. The increased rate of Alzheimer's in individuals with Down syndrome is a result of abnormally high production of  $\beta$ -amyloid. This results from the increased presence of a precursor to  $\beta$ -amyloid in Down syndrome and of increased amounts of  $\beta$ -secretase, an enzyme that divides the larger protein into  $\beta$ -amyloid. This is a direct result of their location on the twenty-first chromosome found in triplicate in children with Down syndrome.<sup>75</sup>

The gene for superoxide dismutase is also found on the long arm of chromosome 21 and is thought to have a role in the neuropathology of Down syndrome. It acts to break down superoxide radicals in the brain. Although it shows normal expression in the fetus of individuals with Down syndrome, it is overexpressed in the brains of adults with Down syndrome and declines as symptoms

of dementia appear. This suggests that it may play a role in the developmental brain abnormalities found in people with Down syndrome.<sup>129</sup>

Astrocyte-derived neurotrophic factor is also coded for on the twenty-first chromosome. In Down syndrome this growth factor is overexpressed prior to birth in the brains of those with Down syndrome. This overexpression increases as the person ages, and in the second decade can be correlated with the degree of β-amyloid found in the brain. In addition astrocyte-derived neurotrophic factor may also play a role in neuronal injury via calcium toxicity.<sup>129</sup>

The brains of people with Down syndrome demonstrate increased levels of interleukin-1. Interleukin-1 is a cell signaling cytokine found in both the immune system and the brain. Interleukin-1 is overexpressed in Down syndrome from fetal stages of development through adulthood.

This overexpression is found in microglia and correlates with the distribution of amyloid plaques. Interleukin-1 is not coded for on the twenty-first chromosome but is modulated by both superoxide dismutase and astrocyte-derived neurotrophic factor discussed earlier. Interleukin-1 may also play a role in modulating the expression of these two factors. In addition to these indirect effects, interleukin-1 is neurotoxic in high concentrations and also plays a role in the neurofibrillary pathology described as tangles.<sup>129</sup>

The resulting gross pathology in people with Down syndrome is an overall reduction in brain weight. This especially affects the size of the cerebral and cerebellar hemispheres, the hippocampus, the pons, and the mamillary bodies. Additional abnormal findings may include smaller convolutions within the brain, structural abnormalities in the dendritic spines of the pyramidal neurons of the motor cortex, and abnormalities of the pyramidal system as a whole, including decreased pyramidal neurons in the hippocampus.

This last finding and the decreased size of the amygdala in people with Down syndrome who develop dementia have particular significance as it relates to the increased incidence of Alzheimer's symptoms in older adults with Down syndrome.<sup>4</sup>

#### Clinical Manifestations

Children with Down syndrome are readily identified by their flattened nasal bridges, eye shape, short limbs, and mild to moderate hypotonia. The most frequently observed clinical characteristics are listed in Table 23-1.

Many other associated clinical manifestations may also be present. For example, people with Down syndrome tend to exhibit increased susceptibility to infections such as otitis media (ear infection). They are frequently hepatitis B carriers and have an increased incidence of ophthalmologic disorders (35%), develop thyroid dysfunction with age (8%), experience constipation associated with gastrointestinal tract anomalies (13%), develop acute leukemia (1%), and present with congenital cardiac anomalies (50%) such as atrioventricular and ventricular septal defects. All of these conditions are present at rates that are greater than those found in the population as a whole.<sup>140</sup>

**Table 23-1** Down Syndrome: Clinical Characteristics

Most Frequently Observed Manifestations	Associated Manifestations
Flattened nasal bridge (90%)	Other congenital anomalies
Almond eye shape	Absence of kidney
Flat occiput	Duodenal atresia
Muscle hypotonia and joint hyperextensibility	Tracheoesophageal fistula
Congenital heart disease	Feeding difficulties
Language and cognitive delay	Atlantoaxial instability
Short limbs, short broad hands and feet	Sensory impairment
Epicantal folds	Hearing loss (conductive)
High arched palate; protruding, fissured tongue	Visual impairment
Delayed acquisition of gross motor skills	Strabismus
Simian line (transverse palmar crease)	Myopia
	Nystagmus
	Cataracts
	Conjunctivitis
	Delayed growth and sexual development
	Obesity
	Diabetes mellitus

Children with Down syndrome frequently present with a variety of musculoskeletal or orthopedic problems believed to be acquired secondary to soft tissue laxity and muscle hypotonia. Some of the more common findings include recurrent patellar dislocation, excessive foot pronation, scoliosis, slipped capital femoral epiphyses (secondary to persistent hip abduction associated with hypotonia), and late hip dislocation (after 2 years).

Atlantoaxial instability (AAI) of the cervical spine (subluxation between C1 and C2) is a characteristic in some children with Down syndrome. This instability is thought to be secondary to ligamentous laxity, odontoid maldevelopment, or possibly abnormal syringomyelia in the area of the odontoid process. Syringomyelia is a slowly progressive syndrome in which cavitation (the formation of a cavity from destruction of tissue) occurs in the central segments of the spinal cord.

The majority of cases are asymptomatic; however, clinical changes indicative of symptomatic AAI include hyperreflexia, clonus, positive Babinski's sign, torticollis, increased loss of strength, changes in sensation, loss of established bladder and bowel control, and a decrease in motor skills.

Children with Down syndrome predictably present with feeding difficulties and delayed acquisition of motor skills. These skills, however, improve with age. Because of the hypotonia and decreased strength, midline upper extremity movement is difficult, and gait usually is characterized by smaller step lengths, increased knee flexion at contact and hyperextension in stance, decreased single limb support, and an increased hip flexion posture. These children present with slower reaction times and slower postural reactions.

Secondary disorders often develop after age 30 or 35, including obesity, diabetes mellitus, and cardiovascular disease. Other significant problems can include osteoar-

thritic degeneration of the spine and osteoporosis with vertebral or long bone fractures.

## MEDICAL MANAGEMENT

**DIAGNOSIS.** Measurement of  $\alpha$ -fetoprotein (AFP), human chorionic gonadotropin, and unconjugated estrogen in maternal serum (triple screen) allows detection of an estimated 60% to 70% of fetuses with Down syndrome. Using this screening test prenatal diagnosis may be made during the second trimester (between 15 $\frac{1}{2}$  and 20 weeks' gestation).

Ultrasound identification based on nuchal translucency provides a good way to identify the fetus with Down syndrome at between 10 and 14 weeks' gestation. Ultrasound carries a 6% false-positive rate and will identify only 77% of affected fetuses.<sup>109</sup>

Postnatal diagnosis usually begins with suspected physical findings at birth. Genetic studies showing the chromosomal abnormality can confirm the diagnosis. Specific diagnostic testing for the secondary problems discussed earlier varies depending on the involved organ systems suspected of dysfunction.

**TREATMENT.** Because no known cure exists for Down syndrome, treatment is directed toward specific medical problems (e.g., antibiotics for infection, cardiac surgery, monitoring of thyroid function, and monitoring for development of Alzheimer's disease). Larger medical centers are pursuing plastic surgery to eliminate the hypoplastic facial features based on the premise that this type of surgery has a positive influence on rehabilitation. The overall goal of treatment intervention is to help affected children develop to their full potential. This involves a team of experts, including therapists.

**PROGNOSIS.** The improved life expectancy of people with Down syndrome as a result of the greater availability of surgery and advances in medical care has been documented, but life expectancy still remains lower than that for the general population.<sup>101</sup>

The presence of congenital malformations, especially of the heart and gastrointestinal tract, can result in high mortality rates in the affected population<sup>57</sup>; lack of mobility and poor eating skills are also predictors of early death.<sup>49</sup> Respiratory tract infections are very common secondary to hypotonicity of chest and abdominal muscles and contribute significantly to morbidity and mortality. Immune system dysfunction also may be present associated with a higher incidence of acute myeloid leukemia than in the general pediatric population.<sup>72</sup>

Significant health problems contributing to mortality have been reported in the adult population with Down syndrome, including untreated congenital heart anomalies, acquired cardiac disease, pulmonary hypertension, recurrent respiratory infections and aspiration leading to chronic pulmonary interstitial changes, and complications from presenile dementia and Alzheimer's disease.

Over the last 40 years the lifespan of those with Down syndrome has increased significantly. In 1968 the average age of death was 2 years of age and by 1997 it had increased to 50 years of age. Unfortunately this degree of improvement has not occurred for everyone with Down

syndrome. There is a significant disparity that exists based on race, with Caucasians with Down syndrome enjoying a significantly longer lifespan than nonwhites.<sup>27</sup>

## SPECIAL IMPLICATIONS FOR THE THERAPIST 23-1

### Down Syndrome

#### PREFERRED PRACTICE PATTERNS

*4C: Impaired Muscle Performance*

*5B: Impaired Neuromotor Development (cardiac anomaly)*

*6B: Impaired Aerobic Capacity/Endurance Associated with Deconditioning*

*6D: Impaired Aerobic Capacity/Endurance Associated with Cardiovascular Pump Dysfunction or Failure (cardiac anomaly)*

*6G: Impaired Ventilation, Respiration/Gas Exchange, and Aerobic Capacity/Endurance Associated with Respiratory Failure in the Neonate (congenital cardiac anomaly in the infant less than 4 months old)*

*Other cardiopulmonary patterns may be present depending upon the presence of respiratory disorders or other comorbidities associated with age.*

#### Precautions

Because AAI is a potential problem, radiographs should be considered before any type of event that could result in a direct downward force on the cervical area (e.g., surgery, especially head and neck surgery; manual therapy; tumbling; diving; horseback riding). Transportation in a car or bus or on a bicycle alone or with an adult may be considered a potentially risky activity requiring specific support. Likewise, riding carnival-type rides such as fast-moving carousels, roller coasters, and so on must be discussed with the family.

Some disagreement exists on the degree of screening and restriction that is required. The Committee on Sports Medicine and Fitness of the American Academy of Pediatrics has published a position paper on AAI in children with Down syndrome that disfavors screening of this population; other sources favor radiologic examinations of the cervical spine of children with Down syndrome.<sup>145</sup>

Subluxation of the cervical vertebrae greater than 4.5 mm (occurs in 1% to 2% of cases) with or without neurologic symptoms is considered to be an indicator for intervention (e.g., cervical fusion). The therapist is an important source of information for increasing family and community awareness regarding the potential precautions and contraindications associated with AAI.

Decreased muscle tone compromises respiratory expansion. In addition, the underdeveloped nasal bone causes a chronic problem of inadequate drainage of mucus. The constant stuffy nose forces the child to breathe by mouth, which dries the oropharyngeal membranes, increasing susceptibility to upper respiratory tract infections.

Low oral motor tone and a protruding tongue can interfere with feeding, especially solid foods. Because

the child breathes by mouth, sucking for any length of time is difficult. When eating solids, the child may gag on food because of mucus in the oropharynx.

The parents should be instructed in taking measures to lessen these problems, such as clearing the nose with a bulb-type syringe, especially before each feeding; providing frequent feedings with opportunities for rest; rinsing the mouth with water after feedings; changing the child's position frequently; practicing good handwashing; and performing postural drainage and percussion if necessary.

### Gross and Fine Motor Development

The therapist concerned with motor development as well as other clinical, educational, psychosocial, or vocational issues relevant to people with Down syndrome is referred to other more intervention-oriented resources.<sup>21,73,190</sup>

### Physical Activity and Exercise<sup>102</sup>

Developing an active lifestyle early in childhood is important given the risk factors for the development of obesity, diabetes mellitus, and cardiovascular disease in this group. The presence of cardiac defects may affect the client's overall level of activity, fitness, and endurance training, especially in the school setting.

Some evidence suggests that individuals with Down syndrome physiologically work harder when engaged in physical activity or exercise (e.g., higher heart rate, greater oxygen consumption, and minute ventilation) compared with peers who are without impairment or who are developmentally delayed but do not have Down syndrome.<sup>50</sup>

Anyone with Down syndrome interested in participating in the Special Olympics must work closely with the therapist, support staff, and physician to establish guidelines for safety. These individuals can benefit from aerobic conditioning, but the frequency, intensity, and duration must be modified from the general recommendations of the American College of Sports Medicine (ACSM).

Lower peak heart rate and lower  $\text{Vo}_2$  in this population require a lower level of aerobic conditioning. Vital signs should be monitored throughout the exercise program as a means of determining workload levels and progressing the activity or exercise.

## SCOLIOSIS

### Definition

Scoliosis is an abnormal lateral curvature of the spine. The curvature may be toward the right (more common in thoracic curves) or the left (more common in lumbar curves). Rotation of the vertebral column around its axis occurs and causes the associated rib cage deformity. Scoliosis is often associated with kyphosis and lordosis.

### Overview and Incidence

Scoliosis is classified as idiopathic (unknown cause; 80% of all cases), osteopathic (as a result of spinal disease or

bony abnormality), myopathic (as a result of muscle weakness), or neuropathic (as a result of a central nervous system [CNS] disorder).

Age of onset can vary from birth onward and is referred to as infantile (0 to 3 years), juvenile (ages 3 to 10), adolescent (age 10 until bone maturity at between 18 and 20 years of age), or adult (after skeletal maturation). Between 0.4% and 5.5% of children may present with some type of scoliosis<sup>166</sup> with one in four of those requiring some type of treatment intervention. The incidence is increased with associated neuromuscular impairments such as cerebral palsy, spina bifida, neurofibromatosis, and muscular dystrophy.

As a whole the overwhelming majority of cases of progressive idiopathic scoliosis are found in the adolescent age group when the growth velocity of the spine again increases after relatively slow growth period between the ages of 5 and 11 for girls (13 for boys).<sup>29</sup>

Infantile idiopathic scoliosis (rare in the United States) is characterized by curvatures that are most often thoracic and toward the left and most commonly affects males. Juvenile idiopathic scoliosis is characterized most often by a right thoracic curvature and can be rapidly progressive. Adolescent idiopathic scoliosis of greater than 30 degrees is seen most often in females without any neurologic impairments in a 10:1 ratio.<sup>188</sup> In its milder forms (10-degree curve or less), scoliosis affects boys and girls equally, but girls are more likely to develop more severe curvatures requiring intervention.

Adult scoliosis (curves greater than 30 degrees) affects about 500,000 people in the United States, whereas the prevalence of scoliosis above the age of 50 is reportedly between 6% and 10% (based on routine chest radiographs).<sup>16</sup>

### Etiologic Factors

Scoliosis may be functional or structural. Functional (postural) scoliosis may be caused by factors other than vertebral involvement, such as pain, poor posture, leg length discrepancy, or muscle spasm induced by a herniated disk or spondylolisthesis. These curves disappear when the cause is remedied. Functional scoliosis can become structural if untreated.

Structural scoliosis is a fixed curvature of the spine associated with vertebral rotation and asymmetry of the ligamentous supporting structures. It can be caused by deformity of the vertebral bodies and may be congenital (e.g., wedge vertebrae, fused ribs or vertebrae, hemivertebrae), musculoskeletal (e.g., osteoporosis, spinal tuberculosis, rheumatoid arthritis), neuromuscular (e.g., cerebral palsy, polio, myelomeningocele, muscular dystrophy), or, most commonly, idiopathic.

At the present time, despite extensive study, the cause of idiopathic scoliosis remains unknown. Researchers hypothesize that this type of scoliosis relates to the maturation disturbances of the CNS, including neurohormonal transmitters or neuromodulators secondary to genetic defect.<sup>108</sup>

Multiple areas of research including abnormalities of connective tissue, neuromotor mechanisms, neurohormonal imbalances (e.g., melatonin, calmodulin), and biomechanics (e.g., the importance of the erect posture)

have been explored for a potential relationship to the cause of idiopathic scoliosis. However, no clear evidence supports any one area as an etiologic factor of this disorder; rather, it appears to be multifactorial.<sup>105</sup>

Linkage studies have identified a genetic predisposition to scoliosis<sup>102</sup>; the probability of a person's having idiopathic scoliosis is estimated to be 20% if other family members have scoliosis.<sup>40</sup>

### Pathogenesis

The pathogenesis of scoliosis remains unclear but may be better understood in relation to the underlying cause. Abnormal embryonic formation and segmentation of the spinal column are possible pathologic pathways in congenital scoliosis. Neuromuscular scoliosis is often the result of an imbalance or asymmetry of muscle activity through the trunk and spine.

The earliest pathologic changes associated with idiopathic scoliosis occur in the soft tissues as the muscles, ligaments, and other tissues become shortened on the concave side of the curve. Some hypothesize<sup>106</sup> that scoliosis sets up abnormal forces across the spine due to the differences in length-tension relationships, with the muscles on the convexity being in a lengthened position and those on the concavity positioned in a relatively shortened state, and as a result a muscle imbalance is present.

Evidence establishes the existence of hypertrophy of the muscles on the side of the convexity<sup>99</sup>; however, the muscles on the concavity still are at a mechanical advantage and facilitate the progression of a curve once it is established. In time, bone deformities occur as compression forces on one side of the vertebral bodies apply asymmetric forces to the epiphyseal ossification center, resulting in increased bone density on that side. The compressive force is greatest on the vertebrae in the apex of the concavity, so the apical vertebrae become most deformed (Fig. 23-1).

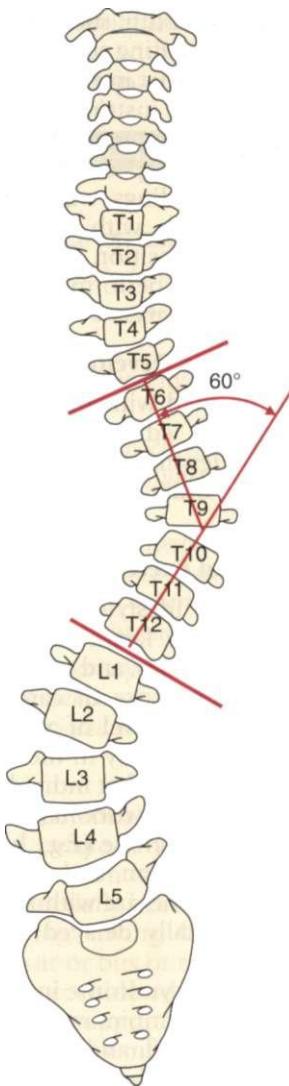
### Clinical Manifestations

Curvatures of less than 20 degrees (mild scoliosis) rarely cause significant problems. Severe untreated scoliosis (curvatures greater than 60 degrees) may produce pulmonary insufficiency and reduced lung capacity, back pain, degenerative spinal arthritis, disk disease, vertebral subluxation, or sciatica.

Back pain is not typical in children or adolescents with mild scoliosis and should be evaluated by a physician who can rule out spondylolisthesis, tumor, infection, or occult trauma. Back pain may be associated with curve progression after institution of brace treatment for idiopathic scoliosis.<sup>146</sup>

The adult with scoliosis often presents with back pain that is considered multifactorial, arising from muscle fatigue, trunk imbalance, facet arthropathy, spinal stenosis, degenerative disk disease, and radiculopathy. Although the incidence of back pain in adults with scoliosis is similar to that in the general population, the pain in the group with scoliosis is greater and more persistent.<sup>16</sup>

Common characteristics of scoliosis are asymmetric shoulder and pelvic position, often identified when clothes do not hang evenly. Curves are designated as right

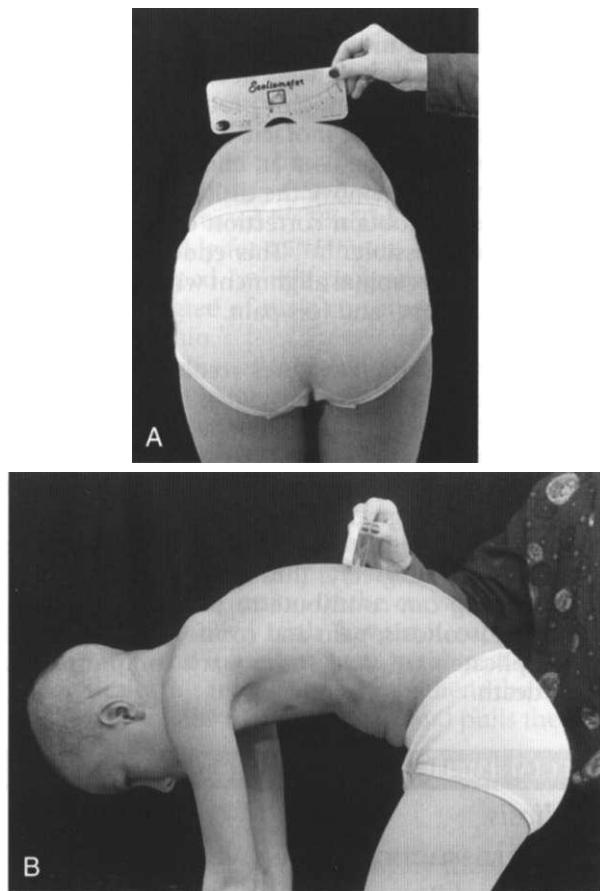


**Figure 23-1**

Cobb's method of measuring scoliosis. This is the method most commonly used because it is readily reproduced. The top vertebra used in the measurement is identified as the uppermost vertebra whose upper surface tilts toward the curvature's concave side. (The superior surface of the vertebra above it usually tilts in the opposite direction or may be parallel to it.) The bottom vertebra is the lowest vertebra whose inferior surface tilts toward the curvature's concave side. (Likewise, the inferior surface of the vertebra below it usually tilts in the opposite direction or may be parallel to it.) A line is drawn parallel to each of these vertebrae. The angle formed by perpendicular lines drawn to each of the parallel lines is the angle of the curvature.

or left depending on the convexity (e.g., right thoracic scoliosis describes a curve in the thoracic spine with convexity to the right). Usually one primary curvature exists with a secondary or compensatory curvature that develops to balance the body. Two primary curvatures may exist (usually right thoracic and left lumbar). If the curvatures of the spine are balanced (compensated), the head is centered over the center of the pelvis; if the spinal alignment is uncompensated, the head is displaced to one side.

Paraspinal muscles become asymmetric as the muscles on the convex side of the curve become rounded, appear-



**Figure 23-2**

The scoliometer. This device can be used by any health care worker trained to screen for scoliosis. Some medical personnel also use this device to monitor curvatures over time, thereby avoiding unnecessary radiographs. Ask the client to bend forward slowly (Adam's position), stopping when the shoulders are level with the hips. View the client from both the front and back, keeping your eyes at the same level as the back. Before measuring with the scoliometer, adjust the height of the person's bending position to the level where the deformity of the spine is most pronounced. This position varies from one person to another depending upon the location of the curvature (e.g., a low lumbar curvature requires further bending than an upper thoracic curvature). Lay the scoliometer across the deformity with the 0 mark over the top of the spinous process. A measurement of 5 degrees or more in the screening test is considered positive and requires medical follow-up. Visually observe for asymmetry of the ribs or paravertebral muscles. In this child, hamstring tightness (greater on the left) accounts for the positional shift to the left. The scoliometer reading was zero. (Courtesy Todd Goodrich, University of Montana, Missoula.)

ing prominent or bulging, while the muscles on the concave side flatten. Rotational deformity on the convex side is observed as a rib hump (*gibbus*) sometimes seen in the upright position but always apparent in the forward bend position.

## MEDICAL MANAGEMENT

**DIAGNOSIS.** Diagnosis by clinical examination requires the client to bend forward 90 degrees with the hands joined in the midline as if taking a dive into a swimming pool. A scoliometer also can be used to measure the angle of trunk rotation (ATR) (Fig. 23-2).

An abnormal finding includes asymmetry of the height of the ribs or paravertebral muscles on one side. The examiner also checks for leg length discrepancy and other asymmetries and for the presence of hair patches, nevi, pits, or areas of abnormal skin pigmentation in the midline indicating possible underlying spinal abnormality.

Differential diagnosis is important in determining whether the scoliosis is structural or functional. Structural curvatures maintain their position irrespective of whether the spine is in an upright or forward bending position. Functional curvatures straighten when placed in a forward bend position. This is especially easy to see when the client is sitting, thereby eliminating weight bearing through the feet. The physician also performs a neurologic examination to rule out an underlying neurologic disorder, especially in the presence of left thoracic curvature.

Full-length radiographs of the spine using techniques to minimize breast radiation dosage are evaluated using the Cobb method (see Fig. 23-1) to measure the degree of curvatures. A curve must be larger than 10 degrees to be considered scoliotic.

The Risser sign is also determined from the film as an indication of maturation and prognostic predictor of progression.<sup>135</sup> Typically, the iliac crest ossifies from anterior to posterior. Risser divided the crest into four quarters according to ossification, grading the ossification from 1 to 4, with a grade 5 indicating that the whole apophysis has ossified and is fused to the iliac crest.

Neuroimaging beyond plain films may be necessary. For example, bone scan may be used to rule out neoplasms, infections, spondylolysis, or compression fractures as the underlying cause. Magnetic resonance imaging (MRI) is used to differentiate cord lesions, disk herniations, neoplasms, infections, spondylolysis, spinal stenosis, and compression fractures.

**TREATMENT.** Prevention of postural or idiopathic structural scoliosis is the key to management of the majority of scoliosis cases. Early detection allows for early treatment without surgical intervention and with good long-term results. Overall goals of management are to prevent severe and progressive deformities that might lead to decreased cardiorespiratory function.

Conservative care in the past has included exercise and electrical stimulation; however, this has not been proven efficacious.<sup>131</sup> Observation and monitoring every 4 to 6 months for curvatures less than 25 degrees, spinal orthoses for curvatures 25 to 40 or 45 degrees (Table 23-2), and surgery for curvatures greater than 45 degrees have been recommended.<sup>123,149</sup> The goal of the use of spinal orthoses is to serve as a passive restraint system to maintain curvatures within 5 degrees of the curvature measurement at the time of initial application. This is accomplished successfully in 85% to 88% of cases.<sup>189</sup> Curvatures with an apex between T8 and L2 and compensated thoracolumbar curves respond the most favorably to bracing,<sup>135</sup> whereas curvatures with an apex at T6 or above have the poorest outcome.

Researchers continue to explore improved dynamic orthotics and holistic treatment approaches. Exercise has

**Table 23-2** Scoliosis: Bracing Options

Brace	Use
Milwaukee (CTLSO)	Best with curvature at T8 or above
Boston (TLSO)	Best with curvature apex lower than T9 or T10
Lyon	For idiopathic scoliosis with thoracic hypokyphosis
Charleston	For idiopathic curves, fabricated in maximum side-bend correction

CTLSO, Cervicothoracolumbosacral orthosis; TLSO, thoracolumbosacral orthosis.

not traditionally been viewed as efficacious for scoliosis; however, there is some renewed interest in its potential effect on the flexibility of an existing scoliotic curve.<sup>74</sup> For many years, exercise has been dismissed as an effective treatment for adolescent scoliosis. More recent findings in support of exercise have been published.<sup>124a</sup>

Looking back at previous studies of exercise for scoliosis, many of the children did not do the exercises. And those who did them only did so occasionally. Unless the exercise program was designed to prepare for a sports activity, compliance was very low. Improved technology and the ability to assess muscle function have changed the picture. We now know that there is asymmetry in muscle function for everyone with idiopathic adolescent scoliosis. More specifically, there is an uneven strength in trunk rotation.<sup>124a</sup>

The former exercise programs of stretching and general strengthening may have been the wrong approach. Progressive resistive exercises specifically aimed at the trunk rotators and extensors are effective for curves less than 45 degrees.<sup>124a</sup>

Orthotic regimens have varied for late-onset idiopathic scoliosis, and the existing research on bracing is promising but does not yet establish conclusively its merit or identify the optimal regimen to be used.<sup>100</sup>

Interventions in the adult with scoliosis should follow a conservative nonoperative course of physical therapy to improve aerobic capacity, strengthen muscles, and improve flexibility and joint motion; nonnarcotic analgesics; nutritional counseling; smoking (or tobacco-use) cessation; and nerve root blocks, facet injections, and epidural steroid injections before surgery is considered. Bracing has never been shown to have an effect on the natural history of adult scoliosis but may be used for certain people who are not operative candidates.<sup>16</sup>

Surgical intervention (e.g., fusion with posterior segmental spinal instrumentation) may be necessary for curvatures greater than 45 degrees, in the presence of chronic pain, or when the curvature appears to be causing neurologic changes. Surgical goals are to halt progression of the curvature, improve alignment, decrease deformity, prevent pulmonary problems, and eliminate pain. The surgical options include a variety of segmental instrumentation systems including Luque, Cotrel-Dubousset, and unit rod instrumentation and Harrington rods. These are combined with a posterior fusion and, in more severe cases, anterior fusion. The use of growth factors to enhance spinal fusion is under investigation.

Minimally invasive surgery can be used in the population to decrease the morbidity associated with open thoracotomy in those people who need an anterior release along with a spinal fusion. This procedure is designed to maximize the stability of thoracic curves with a minimal incision. This technique uses an endoscope to enter the chest anteriorly and remove the disk material to destabilize the spine and obtain correction of the curve to the greatest degree possible.<sup>44,138</sup> This endoscopic approach may result in better spinal alignment with faster recovery, fewer complications, and less pain.<sup>7</sup>

**PROGNOSIS.** Postural curvatures resolve as the primary problem is treated. Structural curvatures are not eliminated but rather increase during periods of rapid skeletal growth. If the curvature is less than 40 degrees at skeletal maturity, the risk of progression is small. In curvatures greater than 50 degrees, the spine is biomechanically unstable, and the curvature will likely continue to progress at a rate of 1 degree/yr throughout life.<sup>16</sup>

Poor seating can contribute to this progression.<sup>93</sup> In severe kyphoscoliosis, pain and comfortable positioning can complicate care, and pulmonary compromise can lead to death.

## SPECIAL IMPLICATIONS FOR THE THERAPIST 23-2

### Scoliosis

#### PREFERRED PRACTICE PATTERNS

##### 4B: Impaired Posture

4I: Impaired Joint Mobility, Motor Function, Muscle Performance, and Range of Motion Associated with Bony or Soft Tissue Surgical Procedures (Neuromuscular practice patterns may apply depending upon the underlying etiology.)

##### 6B: Impaired Aerobic Capacity/Endurance Associated with Deconditioning

6E: Impaired Ventilation and Respiration/Gas Exchange Associated with Ventilatory Pump Dysfunction or Failure (Other cardiopulmonary patterns may apply in the presence of severe scoliosis accompanied by cardiac and/or pulmonary compromise.)

##### 7A (For brace, spinal orthosis, cast): Primary Prevention/Risk Reduction for Integumentary Disorders

### Intervention

Key roles of the therapist are screening for scoliosis and the education of the public about scoliosis. Consumer education must include recommendations for adequate calcium intake and participation in weight-bearing activities. This is especially important for girls; studies show a higher risk of developing osteoporosis in this population.<sup>30</sup>

Traditional exercise programs of stretching and general strengthening continue to be employed but have not been found to halt or improve scoliosis even when used in conjunction with orthoses. When utilized, the focus of strengthening programs is on trunk extensors, abdominals, and gluteal muscles (especially hip extensors).

The client may benefit from a specific exercise program that targets the paraspinal muscles. Progressive resistive exercises for the trunk should be done to both sides using exercise equipment for this purpose. The program should start out with weight equal to one-fourth the child's body weight. When the client can do 20 repetitions of each exercise, then the resistance can be increased by 5%. The exercises should be done twice weekly for 15 minutes. Adults with scoliosis can be helped by this program, too. It may not change the degree of their curvature, but it can help control back pain.<sup>124a</sup>

Stretching activities focus on the iliopsoas and low back extensors and lateral trunk flexors on the concave side of the curvature. The therapist also plays an important role in evaluating and assessing the fit of spinal orthotics while coordinating services with the orthotist and physician.

Electrical stimulation has been used for the management of scoliosis with preliminary success reported in halting the progression of the curvature. However, large-scale controlled trials have not supported this approach,<sup>131</sup> and it has lost favor in the past few years. Theoretically this intervention strengthens the muscles on the convex side of the curvature and pulls the spine into alignment.

### Postoperative

During the hospital stay after surgery the therapy and nursing staff must check sensation, color, and blood supply in all extremities to detect neurovascular deficit, a serious complication following spinal surgery that is monitored for intraoperatively. The person should be log-rolled often and deep-breathing exercises encouraged to avoid pulmonary complications.

For those who are ambulatory, mobilization can begin within 24 hours depending upon the surgeon's protocol. Adults are treated with antiembolic stockings and sequential compression devices until they are ambulatory, and most operative clients (depending on the instrumentation used) are fitted soon after surgery with a custom-molded lightweight plastic orthosis. The brace is to be worn full time when out of bed but, in some cases, may require a progressive tolerance schedule. A vigilant preventive skin care program is instituted.

Activities of daily living (ADLs) (even brushing the hair or teeth can be beneficial) and active ROM exercises of the extremities help maintain circulation and muscle strength. Clients should be instructed in quadriceps setting, calf pumping, and other ROM exercises, and they should be performed frequently on their own throughout the day.

Cast syndrome is a rare but serious complication that can follow spinal surgery and application of a body cast. It is characterized by nausea, abdominal pressure, vomiting, and vague abdominal pain; cast syndrome probably results from hyperextension of the spine.

This hyperextension accentuates lumbar lordosis with compression of a portion of the duodenum

between the superior mesenteric artery and the aorta and vertebral column posteriorly. The therapist encountering anyone in a body jacket, localizer cast, or high hip spica cast must be aware of this condition, because it can develop as late as several weeks to months after application of the cast. Medical treatment is necessary for this condition.

The incidence of other postsurgical complications is low but may include infection at the surgical site, dislodgment of instrumentation, failure of fusion, and urinary tract infection, among other common postoperative complications. Osteophytes and foraminal narrowing in the concavity of the lumbar curvature may develop in older clients, causing nerve root impingement and radicular pain. Recovery in the adult may take 6 to 12 months with improvement continuing for up to 2 years.<sup>16</sup>

### Precautions

Precautions after spinal fusion depend on the type of fusion, segmental stabilization versus Harrington rod, and physician preference. Segmental stabilization provides some advantages over the traditional Harrington rod, including the ability to get out of bed on the first day after surgery and home from the hospital as soon as the fourth or fifth day after surgery. The more rigid segmental stabilization may make osteoporosis more likely to occur; however, the rate of pseudarthrosis is lower.<sup>88</sup>

Segmental instrumentation provides a better correction of the scoliosis, and postoperative casting or bracing often is not required. Precautions generally include avoiding excessive bending, trunk rotation, or hyperextension. Lifting limitations are often imposed depending on type of fusion, and the therapist can provide necessary instructions for safe lifting. These precautions are to help prevent breaking or dislodging of the hardware while promoting bony union in the corrected position.

Functional mobility is severely limited for the first 4 weeks after surgery. After 3 months any type of noncontact sport is acceptable, including aerobic exercise such as walking or stationary bicycling; swimming especially is encouraged and can be started early after some types of fusions; however, diving is contraindicated.

By 1 year, the individual may participate in other noncontact activities such as horseback riding, skating, or skiing. Vigorous activities and contact sports usually are avoided unless the client is directed otherwise by the physician, and restrictions can vary from one physician to another and with various types of fixation devices. These same guidelines usually are followed for the child or adolescent who is wearing a brace but has not had surgery.

Skin care and prevention of breakdown are essential for anyone wearing a cast or spinal orthosis or brace. The client should be taught good skin care and how to recognize signs of irritation that can lead to lesions.

## KYPHOSCOLIOSIS

### Overview and Etiologic Factors

Scheuermann's disease (juvenile kyphosis, vertebral epiphysitis) is a structural deformity classically characterized by anterior wedging of 5 degrees or more of three adjacent thoracic bodies affecting adolescents between the ages of 12 and 16. Scheuermann's disease is the most common cause of structural kyphosis in adolescence. The mode of inheritance is likely autosomal dominant, but the etiologic factors and pathogenesis of this excessive kyphosis remain unknown.

Scheuermann originally proposed that vascular disturbance in the vertebral epiphyses (usually at the thoracic level) during periods of rapid growth was the underlying cause; however, this has not been subsequently supported. Scheuermann's disease also has been associated with increased levels of growth hormone, and individuals with this disease are frequently taller than average.

In the aging population, kyphoscoliosis (adult round back) is more likely to develop as a result of poor posture, aging, degeneration of the intervertebral disks, vertebral compression fractures or osteoporotic collapse of the vertebrae, endocrine disorders (e.g., hyperparathyroidism, Cushing's disease), arthritis, Paget's disease, metastatic tumor, or tuberculosis.

### Clinical Manifestations

Adolescent kyphosis is usually asymptomatic, although some adolescents experience mild pain at the apex of the curvature, fatigue, prominent vertebral spinous processes, and tenderness or stiffness in the involved area or along the entire spine. The pectoral, hamstring, and hip flexor muscles are often tight, producing a crouched posture with anterior pelvic tilt and lumbar lordosis. Signs and symptoms associated with adult kyphosis are similar to those of the adolescent form but rarely produce local tenderness unless caused by vertebral compression fractures.

In both adolescent kyphoscoliosis (Scheuermann's disease) and adult kyphosis, the vertebrae are wedged anteriorly and disk lesions called Schmorl's nodes develop. Schmorl's nodes are localized extrusions of the nuclear material through the cartilage plates and into the spongy bone of the vertebral bodies. The cancellous bone reacts by encapsulating the herniated tissue within a wall of fibrous tissue and bone, producing the Schmorl's node.

### MEDICAL MANAGEMENT

**DIAGNOSIS.** Adolescents may be referred for medical evaluation as a result of school screening, or they may present because of concerns over posture and appearance. Adults more commonly present because of increased pain. Diagnosis is based on clinical examination and confirmed by radiographic findings, including Schmorl's nodes, endplate narrowing, and irregular endplates.

**TREATMENT.** Indications for treatment remain controversial, because the true natural history of this disease has not been clearly defined. Presently, the choice of treat-

ment in Scheuermann's kyphosis is based on the severity and progression of the curve, the age of the individual, and the symptomatology present.

Bracing appears to be very effective if the diagnosis is made early and in adolescents who have not reached skeletal maturity and have curves less than 50 degrees.<sup>104</sup> Surgical management is warranted in those with more severe curves and in adults who continue to show progression of the curve or who have progressive neurologic symptoms or unmanageable pain.

### SPECIAL IMPLICATIONS FOR THE THERAPIST 23-3

#### *Kyphoscoliosis*

##### PREFERRED PRACTICE PATTERNS

*See Special Implications for the Therapist: Scoliosis in this chapter.*

Scheuermann's kyphosis usually responds to physical therapy intervention combined with antiinflammatory medications and behavioral modifications.<sup>175</sup> Exercises (e.g., postural exercises, soft tissue mobilization, stretching, thoracic hyperextension, exercises to strengthen abdominal and gluteal muscles) are helpful to maintain flexibility and improve strength in the thoracic musculature.

Precautions and implications for postoperative care are discussed in the previous section (see Special Implications for the Therapist: Scoliosis). Physical load capacity after extensive surgical correction and spinal fusion for Scheuermann's kyphosis is unknown; an individualized decision must be made in each case.

## SPINA BIFIDA OCCULTA, MENINGOCELE, MYELOMENINGOCELE

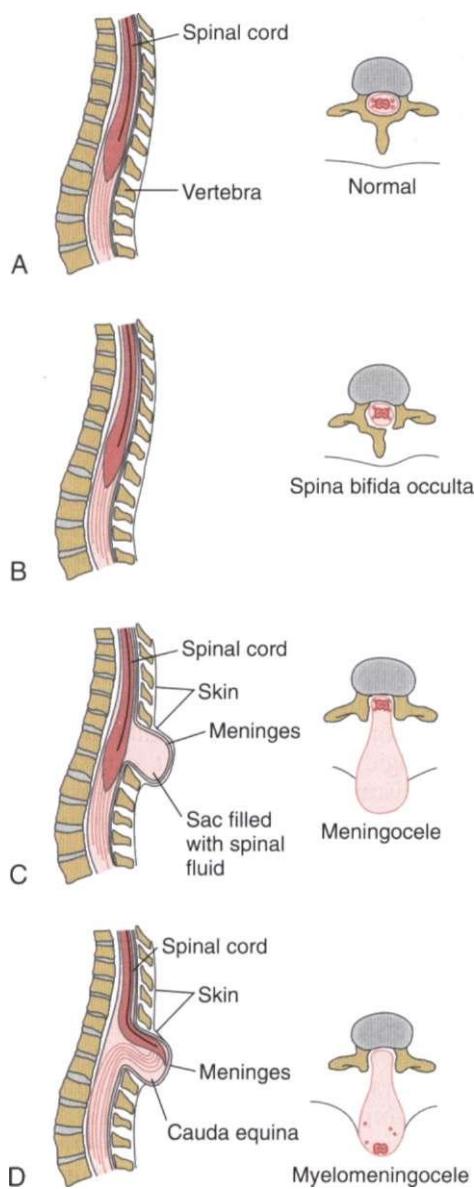
### Definition

Congenital neural tube defects (NTDs) encompass a variety of abnormalities. The term *spina bifida* is the one most often used to describe the more common congenital defects of neural tube closure. Normally, the spinal cord and cauda equina are encased in a protective sheath of bone and meninges (Fig. 23-3). Failure of neural tube closure produces defects that may involve the entire length of the neural tube or may be restricted to a small area.

The three most common NTDs presented here are spina bifida occulta (incomplete fusion of the posterior vertebral arch), meningocele (external protrusion of the meninges), and myelomeningocele (protrusion of the meninges and spinal cord). Generally these defects occur in the lumbosacral area but also may be found in the sacral, thoracic, and cervical areas (Fig. 23-4).

### Incidence and Etiologic Factors

The incidence of NTDs varies by ethnic background, geographic area, and socioeconomic status. Data collected by



**Figure 23-3**

Various degrees of spina bifida. **A**, Normal anatomic structure. **B**, Spina bifida occulta results in only a bony defect, with the spinal cord, meninges, and spinal fluid intact. **C**, Meningocele involves the bifid vertebra, with only a cerebrospinal fluid (CSF)-filled sac protruding; the spinal cord or cauda equina (depending on the level of the lesion) remains intact. **D**, Myelomeningocele is the most severe form because the spine is open and the protruding sac contains CSF, the meninges, and the spinal cord or cauda equina.

the Centers for Disease Control and Prevention place the incidence in Atlanta at 5 per 10,000 live births.<sup>132</sup> Regional variations are significant, however, and in Scandinavia the rate can be as low as 2 per 10,000, whereas in China it can be as high as 100 per 10,000.<sup>159</sup>

The incidence of spina bifida appears to be declining.<sup>194</sup> Termination of pregnancies as a result of the wider availability of maternal serum screening and better nutrition and prenatal vitamins containing folic acid have contributed to this decline.

Evidence supports the hypothesis that the etiology of NTDs is multifactorial and related to the interaction of a



**Figure 23-4**

Myelomeningocele in a newborn. The neural placode is visible at the surface (long arrow) in this lumbosacral myelomeningocele. A placode is an area of thickening in the embryonic epithelial layer where the spinal cord develops later. Abnormal epithelium lines the edges of the cerebrospinal fluid (CSF)-filled cyst (short arrows). (From Burg FD, Ingelfinger JR, Polin RA, et al: *Current pediatric therapy*, ed 18, Philadelphia, Saunders, 2006.)

genetic predisposition, teratogenic exposure, and an essential folic acid deficiency or folic acid metabolic disorder. Folic acid is a B vitamin found chiefly in yeast, orange juice, and green leafy vegetables and bread products, which are now fortified with folic acid.

Multivitamins containing folic acid taken when planning a pregnancy and during the first 6 weeks of pregnancy prevent between 50% and 70% of NTDs.<sup>122,124</sup> Women must be cautioned that half of all pregnancies are not planned and that folic acid must be taken before conception to be effective. Taking supplements containing folic acid is the safest and most effective way of preventing NTDs.<sup>120</sup>

Genetic factors are considered important in the pathogenesis of spina bifida, and several genes have been identified in the folate-homocysteine metabolism pathway. Studies have identified a number of these individually or in combination as being associated with an increased risk of spina bifida. Couples who have had one child with spina bifida have a recurrence rate of between 3% and 8%.<sup>121</sup>

Many genetic disorders are associated with NTDs, either with recessive, dominant, or X-linked inheritance patterns. Increased rates of spina bifida are found in individuals with trisomy 13 and 18,<sup>38</sup> and in chromosome 13q deletion syndrome.<sup>103</sup>

Teratogenic exposure also has been associated with an increased incidence of NTDs. Exposure to vitamin A, valproic acid, solvents, lead, herbicides, glycol ether, clomi-

phenen, carbamazepine, aminopterin, and alcohol has been linked to increased rates of NTDs. A number of occupations have also been linked to NTDs, presumably because of teratogenic exposure. Finally, insulin-dependent diabetes has been associated with increased risk of NTDs as well.<sup>71</sup>

#### Pathogenesis

Normally, about 20 days after conception, the embryo develops a neural groove in the dorsal ectoderm. The neural groove deepens as the two edges fuse to form the neural tube. By about day 23 this tube is completely closed except for an opening at each end. The upper end closes on day 25 and continues to fold and develop, forming the brain, whereas the bottom end closes on day 27 and forms the spinal cord.

The neural groove is formed by both cell proliferation and the production of a hyaluronic acid extracellular matrix. The first opportunity for failure of vertebral architecture to develop and close normally is through abnormalities in the hyaluronic acid matrix or the actin microfilaments that support elevation of the neural crest.

A second opportunity for failed closure is slightly later in development when an abnormal overgrowth at the caudal end may develop, causing closure to fail. Just before closure of the neural tube surface glycoproteins are produced by the ectoderm and act as the glue that holds the cells together.

A third opportunity for failed closure is abnormal production of these glycoproteins, leading to failure of the neural tube to close. A final possible genesis of myelomeningocele is the rupture of the neural tube after its closure as a result of cerebral spinal fluid (CSF) pressure. In this case development of Chiari II malformation occurs, in which the cerebellar tonsils develop below the foramen magnum or are forced through the foramen magnum because of pressure leading to increased CSF pressure and forcing the neural tube open. The defect in myelomeningocele can be identified by the eighth week of gestation and is complete by the twelfth week.<sup>6</sup>

Some animal models support the presence of a defect in homocysteine metabolism in the pathogenesis of NTD, which correlates with an increased risk of NTD in some populations.<sup>112</sup> Plasma homocysteine levels and folic acid levels show an inverse relationship, and current research is focusing on the metabolism of folic acid and its genetic determinants<sup>124</sup> in addition to the importance of these genetic defects in spina bifida. It appears that the genetics of NTDs are multifactorial.

#### Clinical Manifestations

NTDs are typically divided into two groups, occulta (hidden) and aperta (visible). Approximately 75% of vertebral defects are located in the lumbosacral region, most commonly at the L5 to S1 level. Motor dysfunction depends on the level of involvement and sparing of sensory and motor innervation (Fig. 23-5 and Table 23-3).

The loss of motor function is not evenly distributed over the limbs and spine, resulting in muscle imbalance contributing to the development of scoliosis and various

#### Box 23-1

#### SIGNS AND SYMPTOMS OF HYDROCEPHALUS

- Full, bulging, tense soft spot (fontanelle) on top of the child's head
- Large, prominent veins in the scalp
- Setting sun sign (child appears to look only downward; the whites of the eyes are obvious above the colored portion of the eyes)
- Behavioral changes (e.g., irritability, lethargy)
- High-pitched cry
- Seizures
- Vomiting or change in appetite

musculoskeletal deformities that are related to the specific muscles not innervated. Clinical features and other associated characteristics are listed in Table 23-4.

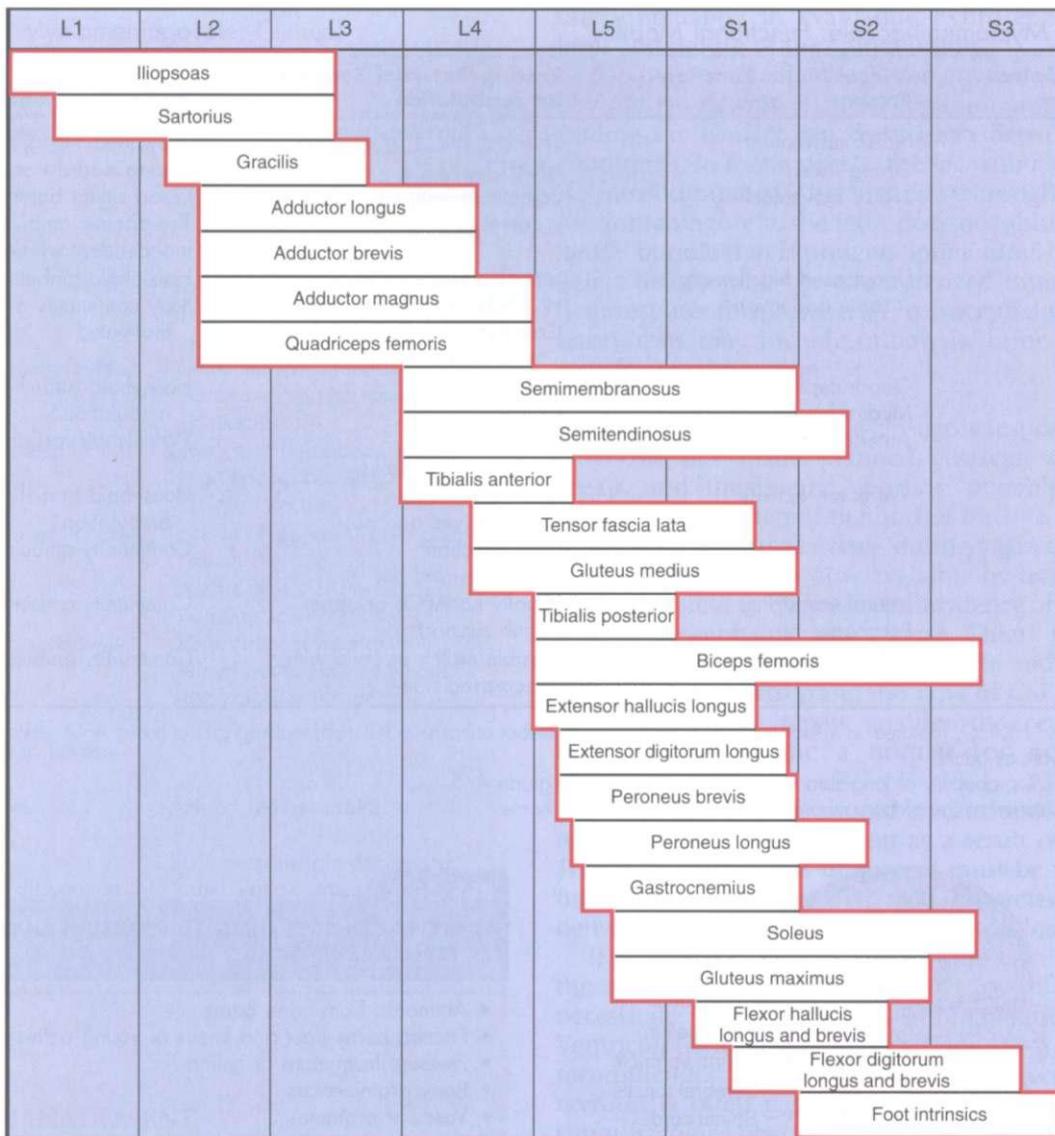
Spina bifida occulta does not protrude visibly but is often accompanied by a depression or dimple in the skin, a tuft of dark hair, soft fatty deposits (subcutaneous lipomas or dermoid cyst), port-wine nevi, or a combination of these abnormalities on the skin at the level of the underlying lesion. Spina bifida occulta usually does not cause neurologic dysfunction, but occasionally bowel and bladder disturbances or foot weakness occurs.

In spina bifida aperta (meningocele and myelomeningocele), a saclike cyst protrudes outside the spine. Like spina bifida occulta, meningocele rarely causes neurologic deficits, whereas myelomeningocele causes permanent neurologic impairment depending on the level of involvement.

Myelomeningocele may be accompanied by flaccid or spastic paralysis, various combinations of bowel and bladder incontinence, musculoskeletal deformities (e.g., scoliosis, hip dysplasia, hip dislocation, clubfoot [talipes equinovarus], hip and knee contractures), hydrocephalus, and sometimes mental retardation. During the first 2 years of life, children with myelomeningocele often present with various degrees of truncal hypotonia and delayed automatic postural reactions, even those children with sacrum-level lesions.

Approximately 90% of children born with this condition have an associated hydrocephalus (Box 23-1). Hydrocephalus accompanying spina bifida usually occurs in the presence of a type I or type II Arnold-Chiari malformation; that is, the cerebellar tonsils are displaced through the foramen magnum (Fig. 23-6), resulting in obstruction of CSF flow and increased CSF pressure and hydrocephalus.

Generally speaking, most children with myelomeningocele have some degree of type II Arnold-Chiari malformation, regardless of the presence of hydrocephalus. This picture has been altered by the advent of fetal repair, and the lower rates of Chiari malformation and hydrocephalus are noted after fetal closure.<sup>20,121,179</sup> Although a Chiari malformation may be present radiographically, it may not be causing any symptoms. Syringomyelia, a cavity or syrinx present within the spinal cord or medulla, also can be present and progress, with pressure impinging on the surrounding tissue.

**Figure 23-5**

Normal lumbar and sacral segmental innervation. For the child with myelomeningocele, once the level of the lesion has been identified, the therapist can begin to assess muscle involvement above and below that level. (From Sharrard WJ: The segmental innervation of the lower limb muscles in man, *Ann R Coll Surg Engl* 35:106-122, 1964.)

Severe Arnold-Chiari malformations and syrinxes are rare but can lead to potentially fatal consequences. Because of the location of the respiratory centers of the brainstem, central apnea can be serious and necessitate the use of mechanical ventilation and can potentially result in death. Sleep problems including hypersomnolence, sleep fragmentation, choking, snoring, and morning headaches are all potential clinical findings.<sup>15</sup>

Cranial nerve involvement with feeding difficulties, choking, pooling of secretions, aspiration, and stridor is also a common finding. Vertigo, ataxia, or spasticity as well as pain, progressive weakness, or diplopia can also be presenting findings in the older child. For a variety of helpful Internet websites related to spina bifida, syringomyelia, and hydrocephalus, see <http://neurosurgery.mgh.harvard.edu/pedi/>.

Tethered cord syndrome is also a common comorbidity following surgical closure of the primary lesion and can occur at any time during growth. As the child grows, the spinal cord can become tethered or bound down, resulting in progressive neurologic compromise. The presenting features are consistent with neurologic compromise and include incontinence, progressive weakness, and back pain. Tethered cord syndrome occurs in between 3% and 5% of children with spina bifida.<sup>85</sup>

Sensory disturbances usually parallel motor dysfunction. Pressure ulcers at the sacrum, ischial tuberosities, knees, and the dorsum of the feet can be a significant comorbidity. Factors contributing to pressure ulcers in this population are listed in Box 23-2. Many of these same risk factors are present in other conditions prone to

**Table 23-3** Myelomeningocele: Functional Mobility

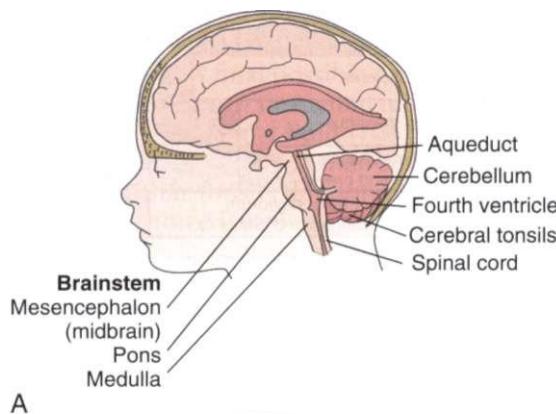
<b>Motor Level Spinal Cord Segment</b>	<b>Critical Motor Function Present</b>	<b>Bracing/External Support for Ambulation</b>	<b>Typical Functional Activity</b>
≤T10	No LE movement	Standing brace or equipment	Supported sitting*
T12	Strong trunk No LE movement	HKAFOs Sometimes with thoracic corset	Sliding board transfers Good sitting balance* Therapeutic ambulation Independent wheelchair mobility Household ambulation*
L1-L2	Unopposed hip flexion, some adduction	Standing brace or equipment HKAFOs, KAFOs, or RGOs Crutches once ambulating with walker	Household ambulation* May community ambulate if motivated
L3-L4	Quadriceps† Medial hamstrings Anterior tibialis	KAFOs Crutches Floor reaction AFOs/twister cables	Household and short community ambulation* Wheelchair for long distances
L5	Weak toe activity	KAFOs Crutches (yes and no) Floor reaction AFOs (yes and no)	Household and short community ambulation* Community ambulation‡
S1	Lateral hamstring Peroneals	Usually no AFOs or upper limb support	Community ambulation
S2-S3	Mild intrinsic foot weakness	Possible crutch or cane with increased age	Community ambulation

LE, Lower extremity; HKAFO, hip-knee-ankle-foot orthosis; KAFO, knee-ankle-foot orthosis; RGO, reciprocating gait orthosis; AFO, ankle-foot orthosis.

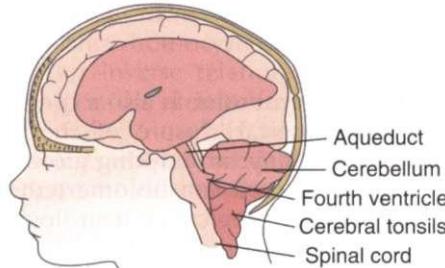
\*Do not usually walk as adults.

†Approximately 50% probability of long-distance ambulation with muscle grade 4/5.

‡Able to use ambulation as the primary means of locomotion outside the home.



A



B

**Figure 23-6**

Arnold-Chiari malformation. **A**, Normal brain with patent cerebrospinal fluid (CSF) circulation. **B**, Arnold-Chiari type II malformation with enlarged ventricles, which predisposes a child with myelomeningocele to hydrocephalus. The brainstem, fourth ventricle, part of the cerebellum, and the cerebral tonsils are displaced downward through the foramen magnum, leading to blockage of CSF flow. Additionally, pressure on the brainstem housing the cranial nerves may result in nerve palsies.

**Box 23-2****FACTORS CONTRIBUTING TO PRESSURE ULCERS IN MYELOMENINGOCELE**

- Ammonia from urine burns
- Friction burns (feet and knees of young active children)
- Pressure from casts or splints
- Bony prominences
- Vascular problems
- Poor transfer skills
- Obesity
- Asymmetric weight bearing or posture (scoliosis, orthopedic deformities)

pressure ulcers (e.g., diabetic neuropathy) (see also Box 10-16 and Table 10-10).

Bowel and bladder problems are present in virtually all children with myelomeningocele because these functions are controlled at the S2 to S4 levels. Even children with sacral lesions and normal leg movement often have bowel and bladder problems.

Problems with urinary incontinence and infection can occur if the bladder is small and spastic (bladder holds little urine) or large and hypotonic (incomplete emptying of the bladder and ureteral reflux). Bladder dyssynergy occurs with either a flaccid or spastic sphincter. Normally, when the bladder contracts, the sphincter relaxes, allowing urine to flow. In a dyssynergistic state, the bladder and sphincter contract together, predisposing the child to urethral reflux.

**Table 23-4** Myelomeningocele: Clinical Features and Associated Characteristics

Clinical Features	Associated Characteristics
Hydrocephalus	90% have intelligence within the normal range (IQ > 80) Increased incidence of learning disabilities 10% to 30% risk of seizures Increased cerebrospinal fluid pressure
Arnold-Chiari malformation	Weakness, pain, sensory changes, vertigo, ataxia, diplopia
Bowel and bladder incontinence	Small spastic bladder: reflux Large flaccid bladder: residual urine, infection
Sensory impairment below the lesion	Lack of response to pain and touch Trophic ulcers of the sacrum and/or lower limbs
Flaccid paralysis below the lesion	0-2 yr: truncal hypotonia Delayed automatic reactions Vasomotor insufficiency Obesity
Absence of deep tendon reflexes	
Clubfoot (talipes equinovarus)	Altered biomechanics
Hip subluxation/dislocation	30% demonstrate decreased ambulation status by age 12 yr
Scoliosis	Late childhood and early adolescence: kyphoscoliosis

## MEDICAL MANAGEMENT

**DIAGNOSIS.** Frequently, NTDs are detected prenatally with ultrasonic scanning and serum AFP testing. Elevated AFP usually occurs by 14 weeks' gestation in the presence of NTDs. This type of screening will not detect skin-covered (closed) neural defects such as spina bifida occulta. The potential for false-positive results with this test may result in unnecessary intervention.

Additionally, as the incidence of this condition continues to decrease, the less reliable the test becomes, because the positive predictive value of the AFP test is dependent on the prevalence of the disease in a population. The less prevalent the disease, the less accurate laboratory results may be. See Chapter 40 for further explanation of the limitations of laboratory tests.

Amniocentesis can detect only open NTDs and is recommended for pregnant women who have previously had children with NTDs or in the case of a large lesion noted with ultrasonic scanning. The need for more accurate, noninvasive imaging of the CNS has been recognized, and fetal MRI has become an effective, noninvasive means of assessing fetal CNS anatomy with superior ability to resolve posterior fossa anatomy over ultrasonography. However, to date fetal MRI has not surpassed

ultrasonography in evaluating hydrocephalus and the level and nature of the spinal lesion.<sup>111</sup>

Postnatally, meningocele and myelomeningocele are obvious on examination. Transillumination of the protruding sac usually can distinguish between these two conditions. In meningocele, the sac with its CSF contents is transilluminated (light shines through the sac); in myelomeningocele, the light does not shine through the neural bundle that is present. Spinal films can be used to detect defects, and the computerized tomographic scan demonstrates the presence of hydrocephalus. Other laboratory tests may include urinalysis, urine cultures, and tests for renal function.

**TREATMENT.** Timing of the closure is important. Prenatal diagnosis has made planned cesarean sections, fetal repair, and therapeutic abortion possible. A cesarean section is the preferred method of birth to avoid trauma to the neural sac that occurs during vaginal delivery.

Prenatal closure is now available by fetal surgery and has been found to decrease the incidence of shunt-dependent hydrocephalus and reverse Chiari malformation from above 90% in each case to 59% and 38%, respectively.<sup>20,179</sup> By interrupting the flow of CSF during gestation, intrauterine repair enables the cerebellum and brainstem to resume a normal (or nearly normal) configuration.

Prospective parents should be cautioned not to expect improvement in leg function as a result of this surgery. The potential benefits of surgery must be weighed carefully against the potential risks of preterm labor and delivery, potential infection, and blood loss.<sup>178</sup>

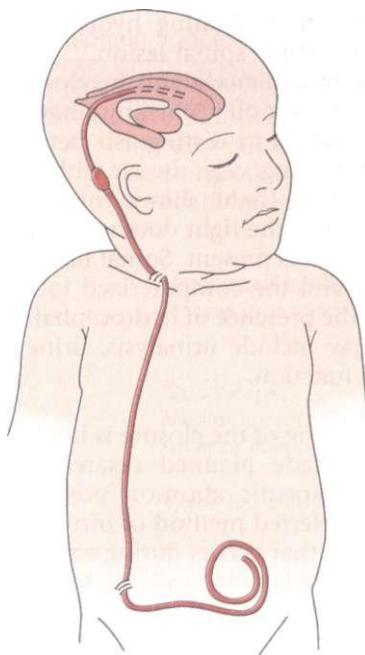
If postnatal closure is chosen, infection and drying of the nerve roots can lead to further loss of function and necessitates surgical closure within 48 hours of birth. Ventriculoperitoneal shunting (Figs. 23-7 and 23-8) is recommended in the presence of hydrocephalus; shunt revision is often required as the child grows or if the shunt becomes obstructed, infected, or separated.

A variety of orthopedic surgical interventions may be required throughout the child's growing years. Surgical correction for hip dislocation rarely is indicated, except in the case of ambulatory clients with unilateral dislocation.<sup>55</sup> Investigation has shown that a level pelvis and good ROM of the hips are more important for ambulation than is reduction of bilateral hip dislocation.<sup>76</sup>

Spinal fusion for kyphotic deformity of the spine has had mixed results and frequent complications. Hip flexion and knee flexion contractures often are addressed with muscle releases, and foot deformity correction often is achieved with soft tissue procedures and in more severe cases with bony procedures (Figs. 23-9 through 23-11).

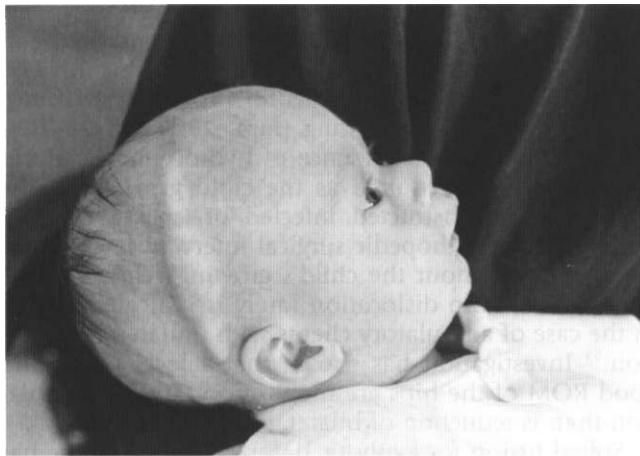
Medical management of the bowel and bladder dysfunction is of critical importance from both a medical and social standpoint. The muscles of the bladder can show either spasticity or flaccidity, leading to either a condition where the bladder is small and under high pressure from urine or large and stretched out and under low pressure.

In children with spastic bladders under high pressure, vesicoureteral reflux and decreased bladder volume and compliance are critical factors that contribute to damage



**Figure 23-7**

Ventriculoperitoneal (VP) shunt. This provides primary drainage of cerebrospinal fluid from the ventricles to an extracranial compartment (usually either the heart [ventriculoatrial] or the abdominal or peritoneal [ventriculoperitoneal] cavity, as shown here). Extra tubing is left in the extracranial site to uncoil as the child grows. A unidirectional valve designed to open at a predetermined intraventricular pressure and close when the pressure falls below that level prevents backflow of fluid.



**Figure 23-8**

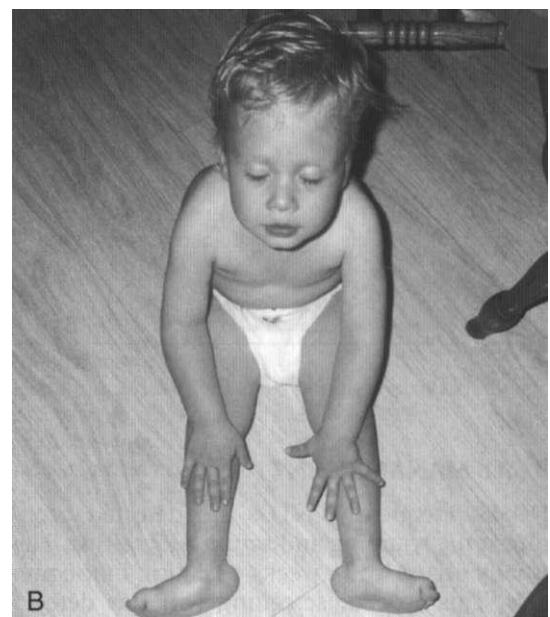
Placement of the shunt. The shunt is placed very superficially, necessitating caution when handling the infant. The therapist must be careful to avoid placing pressure over the shunt, stretching the neck, or placing the child in the head-down position. Parents may be distressed initially by the cosmetic appearance of the shunt, but as the child grows, and with hair growth, the shunt is no longer visible. See Fig. 23-10 of this same child with no obvious signs of a shunt. (Courtesy Todd Goodrich, University of Montana, Missoula.)

to the upper urinary tract and kidneys. Children with hypotonic bladders often have more residual urine and are more prone to infection.

Infection is treated prophylactically in most children with spina bifida, with antibiotics and high fluid intake



**A**



**Figure 23-9**

Orthopedic involvement. **A**, Three-year-old boy with bilateral congenital vertical talus resulting in rocker-bottom foot deformities caused by an L4 to L5 myelomeningocele. Note the compensatory knee flexion and genu valgus along with developing toe flexion contractures (the latter from loss of motor control). **B**, Rocker-bottom foot deformity seen more clearly in the non-weight-bearing position. (Courtesy Zane and Dianna Kuhnhen, Missoula, Montana.)

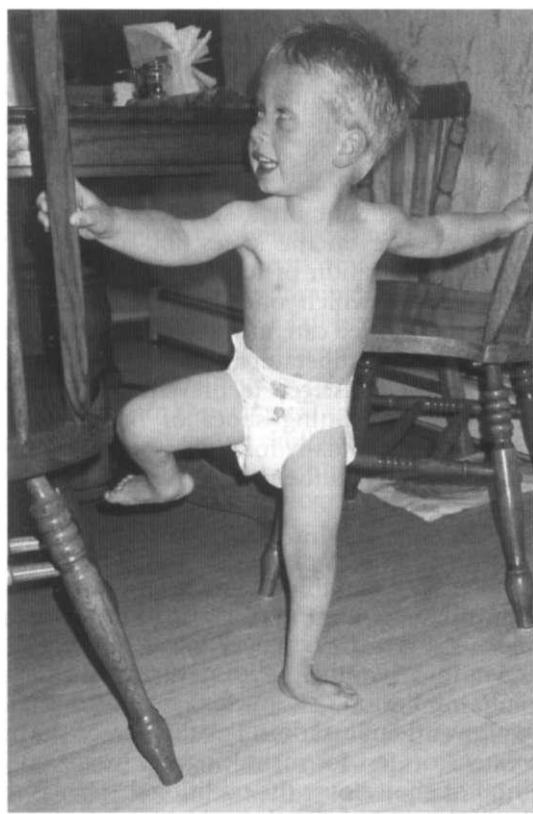
as a critical part of an overall management program. Kidney damage is unusual in children with hypotonic bladders because bladder urine is under low pressure and reflux is less of a problem.

Complete bladder emptying using clean intermittent catheterization provides a means to manage urine flow. Manual pressure on the bladder (Crede's method) is used less often due to its tendency to cause reflux. Implantation of an artificial urinary sphincter has been used in the



**Figure 23-10**

Postoperative inpatient after orthopedic reconstructive surgery for congenital vertical talus deformity. Drainage tubes directly from the incision sites were used for 12 hours. (Courtesy Zane and Dianna Kuhnhen, Missoula, Montana.)



**Figure 23-11**

Postoperative result. Risk for skin breakdown is reduced around the great toe (no longer contracted into flexion), and base of support is improved for ambulation and allows the child to stand on one leg with support (note the more neutral alignment of lower extremity, especially the knee). The child wears ankle-foot orthoses (AFOs) to maintain proper alignment; there may be some regression of alignment in time because of the continued lack of motor control. (Courtesy Zane and Dianna Kuhnhen, Missoula, Montana.)

older child or adolescent,<sup>137</sup> and bladder augmentation or urinary diversion are options for the child with high pressures and insufficient volume.

In most of these devices, the opening and closing of the bladder outlet are accomplished by a cuff placed around the outlet. The cuff can be constricted to close the outlet or relaxed to open the outlet and allow urine to flow. Intravesical electrical stimulation remains controversial; however, it has been employed at some centers, with the benefits of increased bladder compliance and increased bladder volume being the most common positive outcomes.<sup>87</sup>

Stool incontinence is managed most commonly by a program to regulate bowel movements using diet, timed enemas, or suppositories. In some centers the Malone antegrade continence enema procedure is being used to aid in bowel control. This procedure places a cecostomy, bringing the cecum to the abdominal wall in a procedure similar to the placement of a percutaneous endoscopic gastrostomy (PEG) tube. Antegrade enemas are then used to control bowel function.<sup>37</sup>

**PROGNOSIS.** Early, aggressive care of NTDs has now improved the overall prognosis associated with this condition. Prognosis varies with the degree of accompanying neurologic deficit, but researchers are evaluating quality-of-life issues as a possible predictor of prognosis.<sup>91</sup>

At present, prognosis is poorest for those children who have total paralysis below the lesion, kyphoscoliosis, hydrocephalus, and progressive loss of renal function secondary to chronic infection and reflux. At present, survival to adulthood is approximately 85%; most deaths occur before age 4.

Approximately two thirds of children with myelomeningocele and shunted hydrocephalus have intelligence that falls in the normal range. The remaining one third fall into the range for mental retardation, usually mild. Irrespective of IQ, children with spina bifida still have difficulties in perceptual organizational abilities, attention, speed of motor response, memory, and hand function in addition to mental flexibility, efficiency of processing, conceptualization, and problem solving. Overall cognitive delays occur less often as a result of improved medical treatment for these children.

Adult outcome data are incomplete at this time. Most adults over 40 years of age survived the preshunt era of the 1950s and are without hydrocephalus, whereas adults now in their thirties include people with more severe disabilities who benefited from the advances in medical and surgical management.

Adults with myelomeningocele continue to need therapy and medical management secondary to joint and spinal deformities, joint pain, pressure ulcers, neurologic deterioration, depression, and poor social interaction and adjustment.

**Prognosis for Motor Function.** The child's motor abilities vary according to the level of the lesion, but delay in achieving ambulation can be expected in all children with spina bifida, including those with low neurosegment-level lesions.

A child's ability to walk outdoors and use a wheelchair by age 7 usually suggests a good ambulation prognosis.<sup>41</sup>

If functional ambulation is not present by 7 to 9 years of age, it is unlikely to occur subsequently.<sup>3,52</sup> A third of all people with myelomeningocele demonstrate a decline in ambulatory status with increasing age, usually around age 12. These losses in ambulatory status often correlate with a variety of adolescent changes, including increasing body size and composition, loss of upper and lower extremity strength, or immobilization for varied periods of time secondary to musculoskeletal surgery or fracture healing.

Adult ambulatory status in spina bifida is highly determined by two variables, including motor level and sitting balance. Overall ambulation status declines over time.

### SPECIAL IMPLICATIONS FOR THE THERAPIST

23-4

#### *Spina Bifida Occulta, Meningocele, Myelomeningocele*

##### PREFERRED PRACTICE PATTERNS

4A: Primary Prevention/Risk Reduction for Skeletal Demineralization

5B: Impaired Neuromotor Development

5C: Impaired Motor Function and Sensory Integrity Associated with Nonprogressive Disorders of the Central Nervous System—Congenital Origin or Acquired in Infancy or Childhood

6B: Impaired Aerobic Capacity/Endurance Associated with Deconditioning

7A: Primary Prevention/Risk Reduction for Integumentary Disorders

Throughout the lifespan of an individual with any of these conditions, the therapist participates actively in providing direct intervention, preventive care that can reduce complications and morbidity (and the associated costs of these), adaptive equipment, and client and family education. The therapist participates in both preoperative and postoperative care throughout the life of the individual. Functional rehabilitation provided by the therapist facilitates functional outcomes.

A helpful resource to the therapist and family with practical advice regarding the physical, emotional, and cognitive growth of the individual with spina bifida is available.<sup>106</sup> Areas for consideration beyond the scope of this text are also covered in this text (e.g., legal issues, financial planning, vocational assessment).

#### Neonatal Intensive Care Unit

Before surgery to repair the meningocele, pressure of any kind against the sac must be avoided. Whenever holding the (unrepaired) infant, the spine must be maintained in good alignment without tension in the area of the defect. The infant must be kept in the prone position to minimize tension on the sac and to reduce the risk of trauma.

The prone position allows for optimal positioning of the legs, especially in cases of associated hip dysplasia. The infant is placed flat with the hips slightly flexed to reduce tension on the defect. The legs are

maintained in abduction with a pad (a folded diaper or towel) between the knees, and a small diaper roll is placed under the ankles to maintain a neutral foot position.

The prone position is maintained after operative closure, although many neurosurgeons allow a side-lying or partial side-lying position unless it aggravates a coexisting hip dysplasia (see the section on Developmental Dysplasia of the Hip in this chapter) or permits undesirable hip flexion. The side-lying positioning offers an opportunity for position changes, which reduces the risk of pressure sores and facilitates feeding.

In all handling procedures, care must be taken to avoid pressure on the sac preoperatively or on the operative site postoperatively. If permitted, the infant can be held upright against the body. For the infant with hydrocephalus, until the shunt is in place and draining well, activities that position the head above the body tend to decrease intracranial pressure. Activities that position the head below the body increase intracranial pressure; as a result, care should be taken with handling. In the older child positional headaches may be indicative of shunt malfunction.

#### Skin Care

Areas of sensory and motor impairment are subject to skin breakdown and require meticulous care. The loss of skin sensation accompanied by lack of pain can lead to injury and pressure ulcers. Inadequate circulation increases the problem, because the wounds do not heal properly.

Placing the infant on a soft foam or fleece pad reduces pressure on the knees and ankles. Periodic cleansing, application of lotion, and gentle massage aid circulation, which often is compromised. Bath water must be tested carefully because the child cannot feel the water temperature. The family should be advised to use sunscreen to prevent sunburn and to observe for tight-fitting shoes or braces. The skin should be checked daily for red areas that do not disappear readily when the pressure is removed.

#### Early and Ongoing Intervention

##### PRECAUTIONS

Passive ROM exercises must be performed slowly and cautiously given the tendency toward fracture in this population. When the hip joints are unstable, stretching into hip flexion or adduction may aggravate a tendency toward subluxation. For this reason the prone hip extension test for measuring hip extension is the method of choice over the traditional Thomas test.<sup>78</sup>

An early role of the therapist in assessment is to assist in establishing baseline information regarding the level of initially available muscle function and sensation. Information regarding strength assessment specific to this population is available.<sup>78</sup>

Ongoing assessment includes an awareness of signs and symptoms associated with changes resulting from increased CSF pressure in the presence of hydrocephalus with or without a shunt (Box 23-3).

**Box 23-3****SIGNS AND SYMPTOMS OF SHUNT MALFUNCTION**

- Congestion of scalp veins
- Firm or tense soft spot on cranium (fontanelle)
- Listlessness, drowsiness, irritability
- Vomiting, change in appetite
- Marked depression of the anterior fontanelle (overdrainage)
- Disturbance in urinary and bowel patterns
- Increasing head circumference
- Swelling along the shunt
- Seizures
- Nuchal (nape of the neck) rigidity
- Additional symptoms for older children and adults
  - Gradual personality change
  - Headaches
  - Blurring vision
  - Memory loss
  - Progressive coordination problems
  - Declining school or work performance
  - Decrease in sensory or motor functions

A shunting mechanism is used for hydrocephalus associated with a variety of conditions other than myelomeningocele.

Depending on the underlying condition, shunts can become obstructed or stop functioning for many reasons (e.g., occlusion resulting from blood clots or brain fragments, tumor cell aggregates, bacterial colonization, or other debris; the tube itself can become kinked or blocked at the tip; or growth of the infant or child or physical activities can result in disconnection of the shunt components or withdrawal of a distal catheter from its intended drainage site).

Shunt systems also may fail because of mechanical malfunction, including fracture of the catheters, leading to underdrainage or overdrainage.

Infants do not show typical signs of increased intracranial pressure, because skull suture lines are not fully closed. In this age group, a bulging fontanelle is the most obvious sign of pathology. Once the skull bones have fused and the anterior fontanelle is no longer palpable (9 to 16 months of age), pressure can build inside the closed space, resulting in a variety of symptoms, including headache, vomiting, and irritability.

Tethering of the spinal cord may develop with myelodysplasia. The cord becomes caught or tethered from scar tissue and is stretched as the vertebral canal continues to elongate (Box 23-4). Other causes of a tethered cord may include meningocele repair, obstructed CSF shunt, syringomyelia, benign tumor, and spinal cord hypoplasia (i.e., the cord is progressively shorter than the canal and pulled as a result).

Likewise, children with spina bifida have been identified as having the greatest risk of becoming allergic to latex<sup>68</sup> (see the section on Latex Rubber Allergy in Chapter 4). Typical symptoms include watery eyes, wheezing, hives, rash, swelling, and, in severe cases, anaphylaxis, a life-threatening reaction. These responses occur when items containing latex touch the skin,

**Box 23-4****SIGNS AND SYMPTOMS OF TETHERED CORD SYNDROME**

- Changes in bowel and bladder function
- Scoliosis
- Increased spasticity
- Increased asymmetric postures or movement
- Altered gait pattern
- Decreased upper extremity coordination
- Changes in muscle strength (at or below the lesion)
- Back pain

mucous membranes (mouth, genitals, bladder, or rectum), or open areas.

The therapist must avoid using toys, feeding utensils, or other items made of latex that the infant or child might put in the mouth. Parents must be advised to read all labels and avoid products, especially toys and utensils, containing latex. If latex content is not indicated, the manufacturer should be contacted for verification before purchase or use of the item. More information on this topic is available for parents (American Latex Allergy Association, <http://www.latexallergyresources.org>; <http://www.latexallergyhelp.com>).

**CONTROVERSIES**

The timing of the operative repair of the lesion remains under heavy debate. Some centers are repairing the defect before birth and are having good results. In most centers early closure (within the first 24 to 48 hours) after birth is the standard, with the goal of preventing local infection, avoiding trauma to the exposed tissues, and avoiding stretching of other nerve roots, thus preventing further motor impairment.

Other experts contend that surgical repair is best delayed until after further assessment of neurologic function, intellectual potential, and extent of complications. This delay increases the infant's ability to tolerate the surgical procedure, allows for better epithelialization of the sac (thereby reducing the risk of infection), and permits easier mobilization of skin for closure.

A variety of plastic surgical procedures can be used for skin closure. The goal is to place the sac and its contents back in the body with good skin coverage of the lesion and careful closure. Excision of the membranous covering or removal of any portion of the sac may damage functioning neural tissue and is avoided completely. Although this corrective procedure may prevent an infection of the spinal cord or brain, the surgery has little or no effect on the neurologic function of the infant.

Philosophic differences exist regarding the extent and direction of management programs for children with myelomeningocele. Children whose programs emphasize upright activities and ambulation show better outcomes (compared with children whose programs focus primarily on wheelchair mobility) in

*Continued.*

transfer skills (even after they have stopped ambulating), greater bone density, fewer lower extremity fractures, and a smaller incidence of pressure ulcers.<sup>114</sup> High-level lesions do not preclude ambulation; however, this may be a relatively energy-intensive activity as compared with wheeled mobility and may have a negative impact on some aspects of school performance.<sup>54</sup>

Controversy also exists regarding the best choice of lower extremity bracing and ambulation method. One area of contention involves the hip-knee-ankle-foot orthosis (HKAFO) for swing-through gait only versus the reciprocating gait orthosis (RGO), which allows the individual options of a swing-to, swing-through, or reciprocating gait.

Precautions when considering HKAFO or RGO include severe spinal deformity, spasticity, decreased upper extremity strength, moderate obesity, knee flexion or plantar flexion contractures greater than 15 to 20 degrees, and hip flexion contracture greater than 35 degrees.

Another area of bracing controversy is whether to brace high and provide a more normal- appearing pattern and protect against progressive orthopedic deformity or to brace low and allow more freedom of movement. Given the many improvements and number of bracing options available, the key to maintaining ambulatory status is good lower extremity ROM and a level pelvis. The choice of bracing depends on a careful evaluation of the individual's ROM, strength, and gait pattern.

delivery, large neonate, twin or multiple births) and other conditions such as idiopathic scoliosis, myelomeningocele (spina bifida), arthrogryposis, and cerebral palsy.

The presence of other musculoskeletal deformities such as torticollis,<sup>185</sup> metatarsus adductus, and calcaneal valgus deformity should alert the medical practitioner to the need for further evaluation.

Other risk factors include family history, first pregnancies, multiple fetuses, and oligohydramnios (deficient volume of amniotic fluid limiting fetal movement). Certain ethnic groups (Eastern Europeans, Lapps, and Native Americans) also have an increased risk of DDH. One fourth of all cases involve both hips; when only one hip is involved, the left hip is affected three times more often than the right.

#### Etiologic Factors

The cause varies depending on the associated condition but is usually the result of mechanical, physiologic, or environmental factors. Hormonally derived (maternal hormone relaxin may affect the child in utero and during the neonatal period) or hereditary laxity of the ligaments about the joint and positioning are possible etiologic factors.

Infant positioning, both prenatally and postnatally, may affect the formation of the acetabular cup and hip stability because the acetabulum is formed as a result of contact with the femoral head, and this is thought to be one possible cause of DDH.<sup>39</sup>

Cultural customs of how babies are carried affect rates of DDH; those cultures that swaddle infants with the hips in extension and adduction are at greater risk of DDH. Conversely, carrying the infant or young child with hips and lower extremities abducted, flexed, and externally rotated may increase stability of the femoral head in relation to the acetabulum.

#### Pathogenesis

DDH can affect the acetabulum, the femoral head, and the relationship of the femoral head to the acetabulum. The femur, acetabulum, and hip joint capsule usually are well developed by approximately 10 weeks' gestation but continue to enlarge throughout gestation and develop through contact between the femoral head and acetabulum. Most dislocations result in a progressive deformation of the femoral head and acetabulum during gestation.<sup>39</sup>

The subluxated hip maintains contact with the acetabulum but is not well seated within the hip joint. Often this occurs because the acetabulum is shallow, with the roof of the acetabulum sloping at an increased angle in people with DDH rather than showing a normal cup shape. The dislocated hip has no contact between the femoral head and the acetabulum, the femoral head sits on the iliac wing and the ligamentum teres is elongated and taut (Fig. 23-12).

If the dislocation is not diagnosed and treated early, secondary changes in both soft tissues and bony structures occur. The longer the dislocation has been present, the greater the secondary changes that occur. These changes include stretching of the hip capsule, contracture and shortening of the structures of the hip joint, changes

## DEVELOPMENTAL DYSPLASIA OF THE HIP

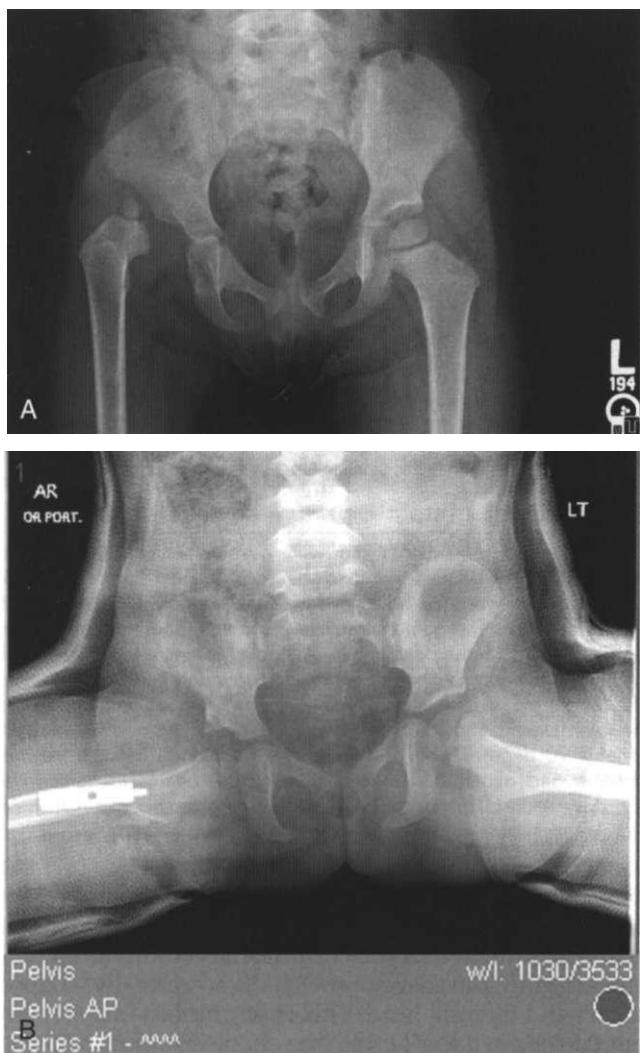
### Overview

Developmental dysplasia of the hip (DDH), previously known as congenital hip dysplasia or dislocation, is a common hip disorder affecting infants and children. The change in name reflects the fact that DDH is a developmental process occurring either in utero or during the first year of life; this condition is not necessarily present at birth as the word *congenital* implies.

DDH can be unilateral or bilateral and occurs in three forms of varying severity: (1) unstable hip dysplasia, in which the hip is positioned normally but can be dislocated by manipulation; (2) subluxation or incomplete dislocation, in which the femoral head remains in contact with the acetabulum but the head of the femur is partially displaced or uncovered; and (3) complete dislocation, in which the femoral head is totally outside the acetabulum.

### Incidence and Risk Factors

The incidence of DDH is between 8.6 and 11.5 per 1000 live births.<sup>98</sup> About 85% of affected infants are females. The risk of hip dysplasia increases dramatically in the presence of certain obstetric conditions (e.g., breech

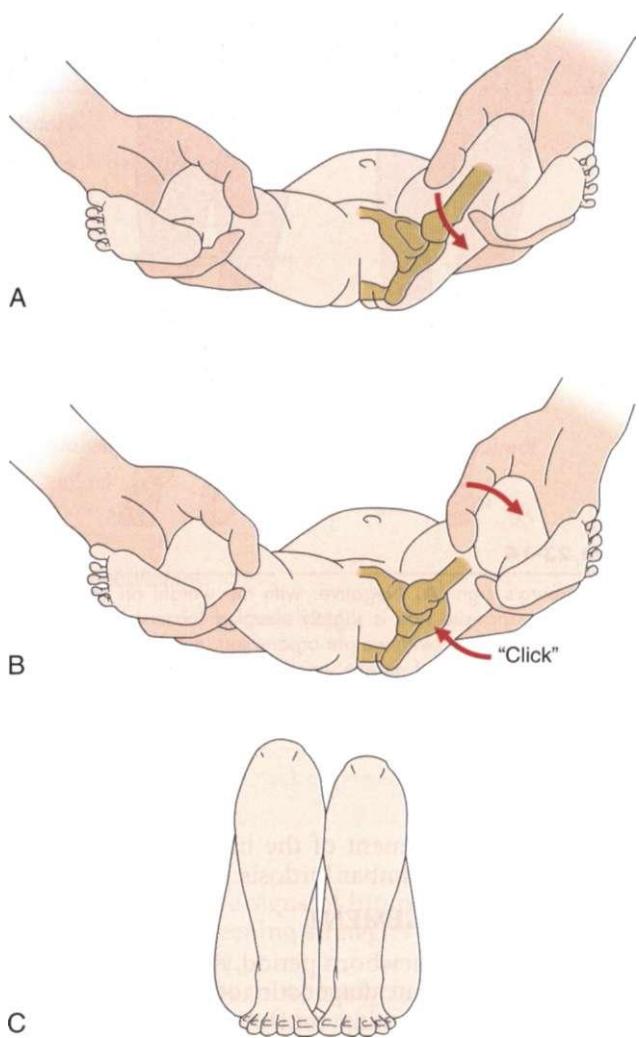
**Figure 23-12**

Developmental dysplasia of the hip (DDH). Three-year-old child with unilateral developmentally dysplastic hip. **A**, Note the head of the femur sitting lateral to the acetabulum. The roof of the acetabulum appears dysplastic and the proximal femur somewhat valgus. **B**, Postoperative: the femur has been relocated in the acetabulum and a varus derotation osteotomy performed. A wedge is cut from the femoral shaft, then internally rotated and positioned in varus to correct the femoral anteversion and valgus. It is also common when there is acetabular insufficiency for portion of the iliac crest to be removed and used as a wedge above the acetabulum to deepen the acetabulum. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

in the blood supply to the hip, flattening of the femoral head, and acetabular dysplasia, sometimes with development of a false acetabulum.

#### Clinical Manifestations

Clinical manifestations of DDH vary with age. In the newborn and nonambulatory period up to 12 months of age, one or more positive signs may be present (Fig. 23-13). Any observed physical asymmetries in ROM (even as little as 10 degrees is considered significant, especially limitation of hip abduction), asymmetry in the buttock or gluteal fold (higher on the affected side), extra

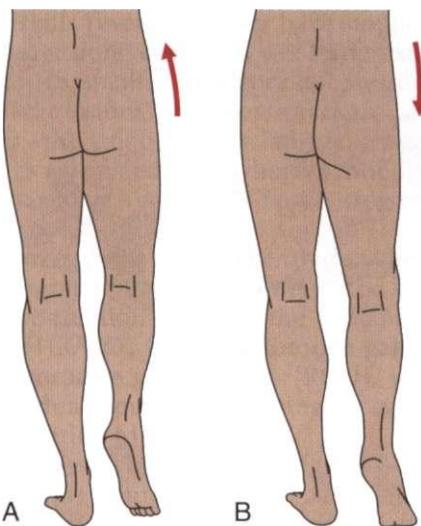
**Figure 23-13**

Signs of hip dislocation. **A**, Ortolani maneuver No. 1 (also the second part of the Barlow test): hip flexion and adduction with downward pressure dislocates the hip. **B**, Ortolani maneuver No. 2: gentle hip flexion, abduction, and slight traction reduce the hip with a discernible click or clunk, and increased hip abduction is possible in a positive test. This test is valid only for the first few weeks after birth. **C**, Galeazzi test (Allis' sign): in the supine position, with hips and knees flexed and feet flat on the floor, the knee is lower on the dislocated side, indicating that the head of the femur is positioned posterior or superior to the rim of the acetabulum. This test is used to assess unilateral hip dislocation and can be used in older children (from 3 months on).

thigh skin folds, or leg length discrepancy requires medical evaluation.

In the ambulating child, uncorrected bilateral dysplasia may cause a characteristic gait pattern known as a compensated Trendelenburg gait. As the child sways the torso from side to side to compensate for an ineffective gluteus medius, the child assumes a waddling gait pattern.

Unilateral dysplasia usually is characterized by a limp with a positive Trendelenburg's sign during the stance phase of gait on the involved side (Fig. 23-14). A flexion contracture on the involved side(s) develops as a result



**Figure 23-14**

Trendelenburg's sign. **A**, Negative: with the weight on one leg, the pelvis on the opposite side is slightly elevated (observed from behind the client). **B**, Positive: with weight on one leg, the pelvis drops on the opposite side because of muscle weakness or pain in the hip joint on the stance side. Trendelenburg's sign measures weakness of the hip abductor muscles, especially the gluteus medius.

of posterior displacement of the hips, which then contributes to marked lumbar lordosis.

## MEDICAL MANAGEMENT

**DIAGNOSIS.** In the newborn period, clinical examination is the most important diagnostic tool and continues to be the standard screening tool. A positive Ortolani or Barlow click confirms DDH in the first month of life (see Fig. 23-13). These tests are considered significantly less diagnostic past 2 to 4 weeks.<sup>65</sup> As ligamentous structures become stronger or if the joints become stretched and worn, it is more difficult to elicit the characteristic popping in and out described.

In some cases, dislocation is not diagnosed by these standard tests, and the disorder may not be apparent at birth. Because a normal neonatal examination cannot guarantee that a hip will not become dysplastic, serial examination throughout infancy is essential. Well-baby checkups should include hip examination until the child begins to walk with a normal gait pattern.

The Galeazzi sign becomes positive in the older infant once shortening of the thigh becomes apparent. Radiographic examination is unreliable in the infant and is used more commonly in older infants and children. Plain radiographs are not able to image the hip adequately until the head is ossified and may not confirm the diagnosis if the unstable hip is in the reduced position at the time the film is taken.

For this reason, ultrasonography is suggested in cases of suspected but unconfirmed DDH. Ultrasonography allows visualization of the cartilaginous structures of the hip and is especially accurate during the first 6 months of life; however, its use as a screening tool remains controversial



**Figure 23-15**

Pavlik Harness. A 7-month old with developmental hip dysplasia wearing a Pavlik harness that holds her legs in flexion and abduction. The harness is worn 23 hours a day, removed only for bathing and diaper changes. The goal of treatment is to keep the femoral head in good contact with the acetabulum. A stable hip encourages the development of a normally shaped socket and rounded head of the femur. The proper hip position must be maintained for enough time to stabilize the joint. The hip should be flexed to 95 degrees and abducted (apart) at least 90 degrees. This position keeps the femoral head in the best position and allows the ligaments and joint capsule to tighten up. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

**TREATMENT.** The goal of treatment for DDH is to ensure stability of the femoral head in the acetabulum, thereby encouraging the development of a normally shaped socket and femoral head. This is accomplished by replacing the head of the femur into the acetabulum with no intervening soft tissue.

The proper position then must be maintained for a period of time sufficient for the bony and cartilaginous structures to develop sufficient stability so that the hip does not subluxate or dislocate with normal movement. Treatment depends on the age of the child and the severity and duration of the dysplasia.

The most common treatment in the infant is placement of the hip in a position of 100 degrees flexion and 90 degrees abduction until the joint capsule tightens and the acetabulum is molded to assume a cup shape. This can be accomplished through the use of a hip harness such as the Pavlik harness (Fig. 23-15). The former standard of treatment with triple diapering is no longer recommended, because proper positioning cannot be ensured and the treatment results in an unacceptable incidence of aseptic necrosis of the femoral head.

The infant must wear the apparatus continuously for 3 to 9 months, eventually weaning its use to nighttime only before its discharge to stabilize the hip in the correct alignment. Criteria for discontinuation of the harness are not standard. Some physicians advocate complete removal of the harness 6 weeks after the hip can no longer be moved in and out of the acetabulum. Others recommend discontinuation when radiographic findings confirm hip stabilization.

Lack of contact of the femoral head in the acetabulum allows the persistence of acetabular dysplasia. This phenomenon is the basis for standing programs for nonambulatory children (i.e., standing provides mechanical forces to assist in the development of the acetabular cup, adding to the stability of the hip).

Treatment in older children who have been walking is usually surgical (e.g. traction, closed reduction [hip spica cast], open reduction, tenotomy of contracted muscles, or osteotomy of either the femur or acetabulum) (see Fig. 23-12, B) depending on the clinical presentation. Traction is used before closed reduction by some in an attempt to aid in reducing the dislocation by applying a distractive force to the joint to loosen the surrounding tissue before the closed reduction.

In the child treated between 6 months and 2 years, a closed reduction is used with an arthrogram to confirm the reduction followed by 3 to 5 months in a hip spica cast. Treatment after the age of 2 years requires surgical reduction, often with both femoral and pelvic osteotomies.

**PROGNOSIS.** Outcome is directly related to the child's age at initiation of treatment. If the dislocation is corrected in the first few weeks of life, the dysplasia is completely reversible and a normal hip can develop, with rates of success as high as 95%.<sup>110</sup> If surgical reduction is required, 86% have a satisfactory outcome.

The two main complications include avascular necrosis (7%) and premature physeal arrest (18%), which presents during the adolescent growth spurt.<sup>197</sup> As the child becomes older and the primary subluxation or dislocation persists, the deformity becomes more difficult to correct conservatively, and increased rates of avascular necrosis and redislocation are seen.

When the condition is untreated, long-term problems can include degenerative joint disease, hip pain, antalgic gait, scoliosis, back pain, or the need for total hip replacement.

## SPECIAL IMPLICATIONS FOR THE THERAPIST 23-5

### *Developmental Dysplasia of the Hip*

#### PREFERRED PRACTICE PATTERNS

**4A:** Primary Prevention/Risk Reduction for Skeletal Demineralization

**4B:** Impaired Posture

**4C:** Impaired Muscle Performance

**4D:** Impaired Joint Mobility, Motor Function, Muscle Performance, and Range of Motion Associated with Connective Tissue Dysfunction

**4I:** Impaired Joint Mobility, Motor Function, Muscle Performance, and Range of Motion Associated with Bony or Soft Tissue Surgery

**7A:** Primary Prevention/Risk Reduction for Integumentary Disorders

Often therapists are involved early and regularly in managing a child's program for some condition other than hip dysplasia and may be the first health care

### Box 23-5

#### HIP DYSPLASIA: QUICK SCREEN

##### *History*

- Breech delivery
- Family history
- Female

##### *Lower Extremity Examination*

- Foot alignment
- Hip range of motion (asymmetry, limited abduction)
- Asymmetric skin folds (thigh, gluteal)
- Buttocks appear flattened
- Leg length discrepancy

##### *Hip Stability (Only One Positive Test Finding Required)*

- Ortolani sign
- Barlow sign
- Galeazzi sign

##### *Gait (If Ambulating)*

- Abnormal (waddling gait, limp)
- Positive Trendelenburg's sign
- Pain

##### *Typical Posture*

- Lower extremity hip flexion, adduction, internal rotation
- Asymmetric head and neck alignment when associated with torticollis

workers to observe signs of hip pathology. An awareness of quick screening strategies for hip dysplasia is critical (Box 23-5).

Physical therapy preoperatively and postoperatively is often a vital part of the rehabilitation process. Preoperative intervention may include lower extremity and trunk strengthening and parent/caregiver education. Positioning and handling techniques are an important aspect of the child's care both before and after surgery. Postoperatively, the therapist reviews cast care (or traction/orthotic care) with the child's family/caregivers.

#### **Precautions**

Positioning and splinting strategies focus on hip abduction and external rotation. Care must be taken not to set the harness in too much flexion (more than 120 degrees) secondary to the potential for impingement on the vascular supply to the femoral head. Force should not be used in flexing and abducting the hip as the excessive pressure can cause avascular necrosis.

Positions to avoid include lower extremity adduction and flexion as occurs in the side-lying position, especially with one lower extremity drifting across the midline.<sup>26</sup> Tests for hip subluxation or dislocation (see Fig. 23-13) should not be repeated too often, because they can result in persistent laxity, articular damage to the head of the femur, and dislocation.

When transferring a child immediately after casting, use the palms to avoid making dents in the cast. Any indentations in the cast can predispose the child to

*Continued.*

pressure ulcers. The cast needs 1 to 2 hours to completely air dry. Check color, sensation, and motion of the child's legs and feet, and notify the physician immediately if dusky, cool, or numb toes. Observe for signs that the cast is too small (e.g., cyanosis, cool extremities, pain).

### **Motor Development**

The widely abducted position of the lower extremities limits opportunities to initiate or continue development of low back and hip extension, especially in the absence of the prone position. Likewise, the widely abducted base of support in sitting decreases opportunities for the use of trunk rotation necessary for transitioning in and out of positions such as sitting and lying prone. The therapist must closely monitor overall progression of motor development during this time and provide as many opportunities as possible for the development of these important skills.

## **NEUROMUSCULAR DISORDERS**

Neuromuscular disorders including the muscular dystrophies, congenital myopathies, and spinal muscular atrophy are presented in this chapter. Other neuromuscular disorders such as Charcot-Marie-Tooth disease, amyotrophic lateral sclerosis (ALS), Guillain-Barre polyneuritis, and chronic inflammatory demyelinating polyneuropathy are discussed in other chapters in this text.

### **The Muscular Dystrophies**

#### **Definition and Overview**

The muscular dystrophies (MDs) comprise the largest and most common group of inherited progressive neuromuscular disorders of childhood. They affect all population types, even animals. Signs of MD can occur at any point in the lifespan.

These disorders, in general, have a genetic origin and are characterized by ongoing, typically symmetric, muscle wasting with increasing deformity and disability. Paradoxically, in some forms (e.g., Duchenne's, Becker's) wasted muscles tend to hypertrophy because of connective tissue and fat deposits, giving the visual appearance of muscle strength.

Six major types of MD are included in this text discussion: (1) Duchenne's muscular dystrophy (DMD), (2) Becker's muscular dystrophy (BMD), (3) facioscapulohumeral (Landouzy-Dejerine) dystrophy (FSHD), (4) limb-girdle dystrophy (LGMD), (5) myotonic dystrophy, and (6) muscular dystrophy congenita (MDC), also known as congenital muscular dystrophy (CMD). These forms of MD involve a primary degeneration of muscle with a gradual loss of strength, but each type differs as to which muscle groups are affected (Fig. 23-16).

#### **Incidence and Etiologic Factors**

The incidence of DMD is approximately 1 in 3500 live births. Rates of occurrence for each type are listed in Table 23-5. All dystrophies are genetically based disorders.

DMD and BMD are X-linked recessive disorders caused by mutations in the dystrophin gene Xp21 that codes for the muscle membrane protein dystrophin. The affected gene on the short arm of the X chromosome (Xp21) is one of the largest genes in the human genome. In these two forms of MD, males are affected clinically and females are usually only carriers.

FSHD is an autosomal dominant disorder with onset in early adolescence. The son or daughter of a person affected with FSHD is at 50% risk of inheriting the defective gene. FSHD occurs with an incidence of 1 in 20,000.

The gene for FSHD has been localized to 4q35 in most people with FSHD; however, the specific gene has not been identified, and there are some people with the FSHD phenotype in whom the defect does not localize to the fourth chromosome.

LGMD may be inherited in several ways depending on the type. LGMD type 2 (A through H) disorders are autosomal recessive disorders of late childhood or adolescence and type 1 (A through G) disorders are autosomal dominant disorders. Dominant disorders have a 50% risk of inheritance if one parent is affected; recessive disorders carry a 25% risk of disease when both parents are carriers and a 50% chance of carrier status.

Myotonic dystrophy has an incidence that varies between 1 in 5000 to 1 in 50,000, with rates in some populations that approach 1 in 550 because of local founder effects.<sup>196</sup> The founder effect occurs when there is a loss of genetic variation such as occurs when a new colony is formed by a very small number of individuals from a larger population. For example, when a small part of a population moves to a new locale, or when the population is reduced to a small size because of some environmental change, the genes of the "founders" of the new society are disproportionately frequent in the resulting population.

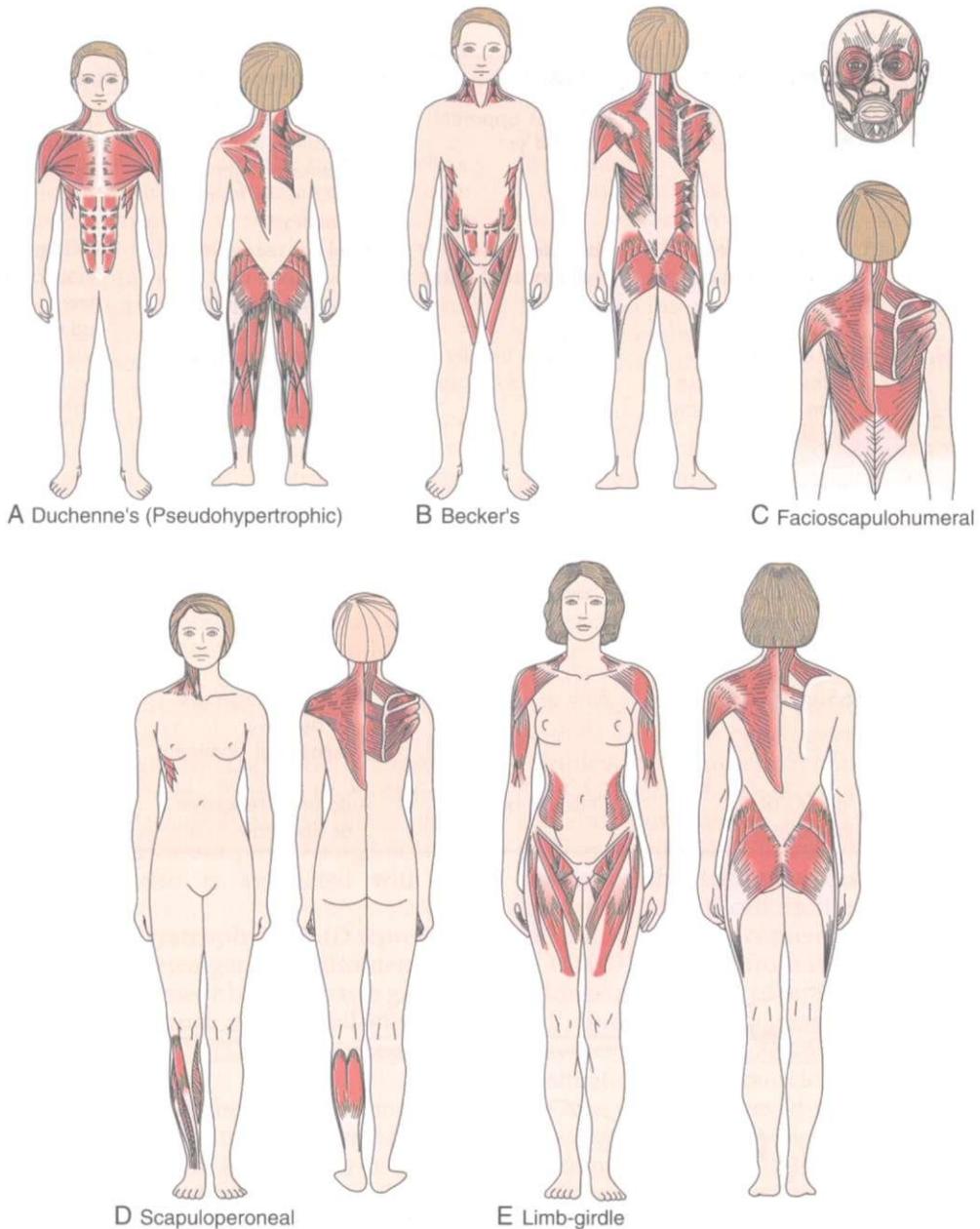
Myotonic dystrophy demonstrates an autosomal dominant inheritance pattern, with each generation being somewhat more severely affected than the last. This increase in the phenotypic severity of the disease state with succeeding generations is referred to as anticipation and can be correlated with an expansion in the size of the triple-repeat genetic enlargement that is the causative factor in this disease.<sup>96</sup>

MDC represents a group of recessively inherited disorders that can be divided into two groups based on the presence of brain involvement. Only the most common forms are included here. The overall incidence has been placed at 4.65 per 100,000 in the Italian population.<sup>127</sup> In Japan, however, Fukuyama MD, one form of MDC, is as common as DMD.

There are a number of classification schemes that have been proposed for the MDCs.<sup>130</sup> For example, MDCs can be divided into those that are the result of extracellular protein deficits, endoplasmic reticulum protein deficits, defects of glycosylation, and defects of integrin.

#### **Pathogenesis**

Knowledge of the MDs and understanding of their increasing complexities escalated dramatically in the late 1980s when the protein dystrophin was identified as the

**Figure 23-16**

Muscle groups involved in muscular dystrophies. These are presented in relative terms; that is, unlike spinal cord injury with definitive muscle involvement, in muscular dystrophy, proximal or distal muscle groups are affected in varying ways with individual differences noted. For example, in the facioscapulohumeral form, the lower erector spinae is featured here but may be spared, and in limb-girdle dystrophy, the lower abdominal muscles may be involved but are not shown in this illustration. **A**, Duchenne's: shoulder girdle (trapezius, levator scapulae, rhomboids, serratus anterior), pectoral muscles, deltoid, rectus abdominis, gluteals, hamstrings, calf muscles. **B**, Becker's: neck, trunk, pelvic and shoulder girdle. **C**, Facioscapulohumeral: muscles of the face and shoulder girdle. **D**, Scapuloperoneal: muscles of the legs below the knees (first), shoulder girdle (later). **E**, Limb-girdle: upper arm (biceps and deltoid) and pelvic girdle.

causative factor in DMD and BMD. Subsequently, other members of the dystrophin glycoprotein transmembrane complex were identified as causative proteins in many other forms of MD.

In addition to transmembrane proteins of the dystrophin glycoprotein complex, there have been proteins in the extracellular matrix, sarcomere, and nucleus identified as causative in MD (Fig. 23-17). Recently it has become apparent that not only can structural proteins

create MD but enzymatic defects and defects in glycosylation (the modification of proteins by the addition of sugars) can also create MD.

These discoveries, along with advances in research and technology, have brought new information on the molecular pathogenesis of these disorders, including the genetic and molecular characterization of many forms of MD.

**Duchenne's and Becker's Muscular Dystrophy.** The affected gene in DMD/BMD encodes messenger RNA

**Table 23-5** Disorders of Muscle

Type	Incidence	Onset	Inheritance	Course
Duchenne's muscular dystrophy (DMD) (pseudohypertrophic)	20-30 in 100,000 live male births; female carrier	Becomes apparent at 2-4 yr	X-linked; recessive; mutation in the dystrophin gene; 30% arise from mutation	Rapidly progressive; loss of walking by 9-10 yr; death in twenties
Becker's muscular dystrophy (BMD)	5 in 100,000 live births; female carrier	Variable, initial diagnosis 5-10 yr	X-linked; recessive; mutation in the dystrophin gene	Slowly progressive; walking maintained past early teens; lifespan until adulthood
Facioscapulohumeral dystrophy (FSHD)	5 in 100,000 live births (males more often affected than females); female carrier	Any age: usually early adolescence	Autosomal dominant; 10%-30% arise from mutation	Slowly progressive; loss of walking in later life; variable life expectancy
Limb-girdle muscular dystrophy (LGMD)	1 in more than 100,000 live births	Late adolescence: early childhood	Autosomal recessive or dominant	Slowly progressive; mild impairment
Myotonic dystrophy	13 in 100,000 live births	Variable onset, classically adolescence	Autosomal dominant	Rate of progression dependent on age of onset; mild involvement, greater functional independence, greater longevity
Congenital muscular dystrophy (MDC; CMD)	4.65 in 100,000	Birth or shortly after	Autosomal recessive or de novo autosomal dominant	Progressive; death for some in first years, in others more slowly progressive and ambulation achieved
Congenital myopathies	2 in 100,000 (nemaline)	Onset at birth	Autosomal recessive or dominant	Initial improvement, static to slowly progressive

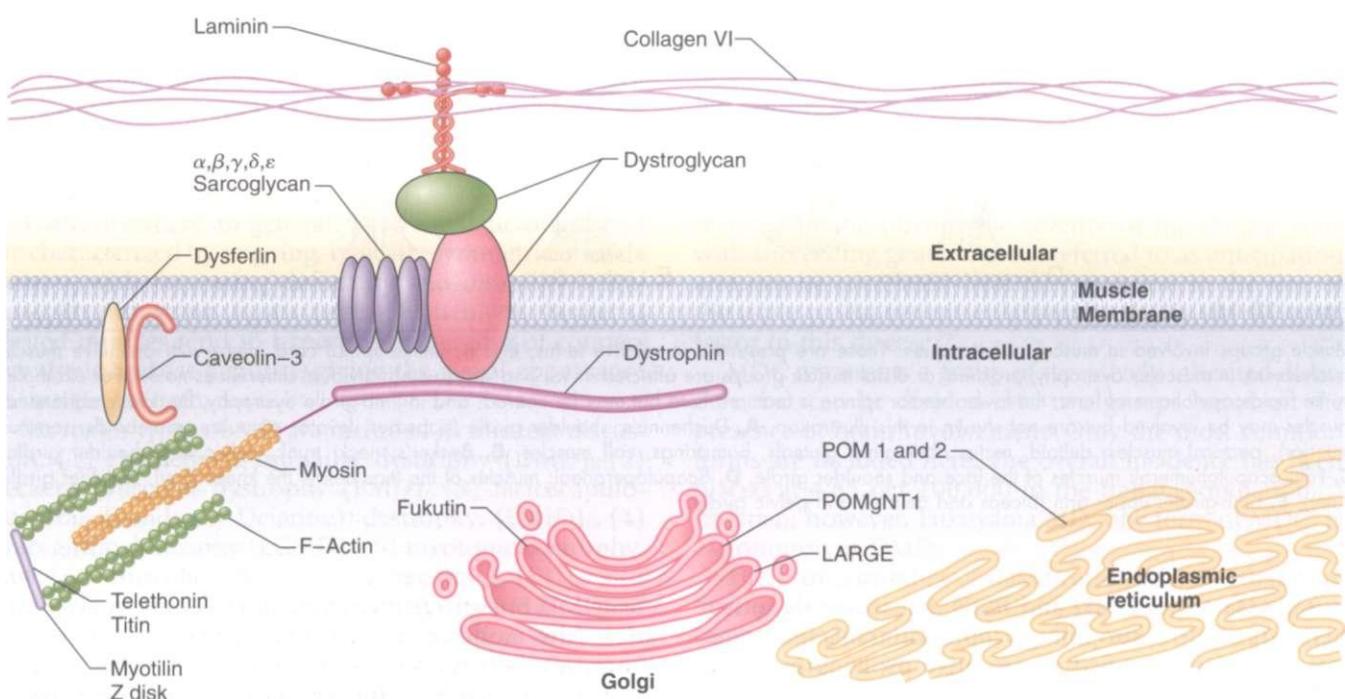
**Figure 23-17**

Diagram showing the most common muscle protein defects that lead to muscle disease. The dystroglycan complex spans the muscle membrane and connects dystrophin and the myosin/F-actin contractile mechanism to the extracellular matrix. The Golgi complex and endoplasmic reticulum are also displayed with the causative factors related to glycosylation defects. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

(mRNA) for the adhesive protein dystrophin that is located in the muscle membrane, the sarcolemma. Muscle membrane lesions play an early role in the pathogenesis of MD, involving skeletal, cardiac, and smooth muscle membranes.

Dystrophin is the protein that links the muscle surface membrane (sarcolemma) with the contractile muscle protein (actin). Lack of normal dystrophin makes the sarcolemma susceptible to damage during contraction-relaxation cycles. Disruption of the muscle membrane and muscle fiber necrosis are initiated by muscle contraction, especially eccentric contraction.<sup>136</sup>

The underlying biochemical defect in all types of MD does not necessarily disrupt the integration of dystrophin in the membrane, and while the absence of some sarcoglycan molecules in LGMD (see later) can lead to susceptibility to mechanical trauma, the absence of others does not.<sup>69</sup>

Males with undetectable levels of dystrophin have Duchenne-type MD, whereas those with nearly normal levels but dystrophin of an abnormal size or low levels have the Becker-type MD. The absence or altered state of dystrophin destabilizes the membrane and allows the influx of  $\text{Ca}^{2+}$ , which triggers the destruction of the cell from the inside through the activation of calpain, a calcium-activated proteinase.<sup>133</sup>

Muscle cells are replaced by fatty and connective tissues, and contractures develop. Fat cells then continue to accumulate between damaged muscle fibers as a response to the ongoing muscle atrophy. Disorganization of tendinous insertions also is associated with fat accumulation.

**Limb-Girdle Muscular Dystrophy.** LGMD represents a collection of genetically heterogeneous disorders that can be broadly divided on a genetic basis into two groups. LGMD type 1 (LGMD1A through 1G) are all inherited dominantly, and LGMD type 2 (LGMD2A through 2K) are inherited recessively.

LGMD type 1A is the result of the absence of myotilin and is very rare. Myotilin is a thin filament protein associated with the Z disk and involved in assembly of the contractile mechanism of the cell.<sup>153</sup> LGMD1B results from the absence of lamin A and lamin C. Lamins A and C are part of a large class of proteins. Lamins A and C are nuclear membrane proteins that are involved in stability of the nuclear membrane and cell differentiation.<sup>67</sup>

LGMD1C results from the absence of caveolin 3. Caveolin 3 is found as part of the muscle membrane and acts in cell signaling. The remaining dominant forms of LGMD (2D through 2F) have genetic characteristics but as yet have no protein identified.<sup>67</sup>

The second group of LGMDs (2A through 2K) are more common than the dominant forms and are inherited recessively. LGMD2A is one of the most common, with an estimated carrier frequency of 1 in 103.<sup>139</sup> The underlying defect in LGMD2A is that of a cellular regulatory enzyme P94, calpain 3, in the calpain family of molecules (calcium-activated protein enzymes). The function of calpain 3 is not well understood, but it appears to play a role in the organization of the muscle cell as it forms.<sup>133</sup>

LGMD2B is caused by an absence of dysferlin (chromosome 2p13). This is also the protein defect in Miyoshi myopathy, which presents with a different dystrophic distal phenotype.<sup>187</sup> Dysferlin acts as a membrane repair molecule and interacts with other molecules at the muscle membrane.<sup>25</sup>

The absent gene in LGMD2C, 2D, 2E, and 2F all code for a specific protein in the sarcoglycan-glycoprotein complex. This glycoprotein complex is found in close association with dystrophin in the muscle membrane. These proteins act to stabilize the membrane. As a group these are known as sarcoglycanopathies, because each is named by the missing sarcoglycan protein ( $\gamma$ ,  $\alpha$ ,  $\beta$ , and  $\delta$ ).

Sarcoglycan-deficient muscle in some cases is sensitive to eccentric contraction-induced disruption of the plasma membrane.<sup>69</sup> In fact, researchers have further identified the specific sarcoglycan deficiency to be that of 8-sarcoglycan. Reduced levels of other forms of sarcoglycan (e.g.,  $\gamma$ ,  $\alpha$ , (3) appear to be involved with the instability at the muscle membrane but do not account for contraction-induced muscle injury.<sup>69</sup>

The pathogenesis of fiber demise in some types of LGMD is presumably similar to that of DMD and BMD. The absence of some of the sarcoglycans affects the integration of other sarcoglycans and the dystroglycan complex in the muscle membrane.<sup>69,176</sup>

LGMD2G is the result of a genetic defect on the seventeenth chromosome (17q11-12), which codes for the protein telethonin. Telethonin is a protein that acts closely together with titin (discussed later) in the formation of the muscle cell. In the adult muscle it is found at the Z disk in the sarcomere.<sup>125</sup>

LGMD2H results from the absence of TRIM 32, which belongs to a family of similar proteins. TRIM 32 is not well understood but acts in conjunction with other enzymes in the muscle and is found to be up-regulated in situations where muscle remodeling is occurring, as is the case when changes in weight bearing occur. In addition, it is found in increased levels when muscle cells are differentiating. TRIM 32 is found associated with the thick myosin filaments of the sarcomere.<sup>95</sup>

LGMD2I is also a common form of LGMD caused by a mutation in the fukutin-related protein (FKRP) gene (19q13.3). FKRP encodes for an enzyme that is involved in glycosylation of  $\alpha$ -dystroglycan.<sup>174</sup>  $\alpha$ -Dystroglycan is a component of the dystrophin glycoprotein complex.

The clinical picture of individuals with abnormalities of this protein is widely distributed, ranging from mild, late-onset disease in the fourth or fifth decade of life to a severe form of MDC with brain abnormalities in addition to severe weakness at birth.<sup>14</sup>

LGMD2J results from a mutation at 2q24.3 in the titin gene. Titin is a large intracellular protein that is responsible for elasticity and stability of the sarcomere. In addition it plays a role in the assembly of the sarcomere in the developing muscle cell. Titin extends from the M line and connects the thick-filament myosin to the Z line, where it forms an elastic connection.<sup>107</sup> Clinically titinopathies can present as the LGMD2J homozygous form, resulting from mutations on both alleles, and as a milder

tibial MD, a heterozygous form with mutations on only one allele.<sup>180</sup>

LGMD2K has been designated to identify the milder phenotypic variants of the POMT1 mutation. This is the same mutation that is found in some of the more severe MDC cases with Walker-Warburg syndrome (WWS). This form of LGMD is one of the only forms that has mental retardation as a component. There are some intermediate forms that fall between LGMD and WWS. Like the other glycosylation defects found in FKRP, there is a spectrum of severity found in people with POMT1 mutations. This is likely the result of still unknown modifying factors.<sup>33</sup>

**Congenital Muscular Dystrophy.** The pathogenesis of the MDCs with brain involvement is related to the glycosylation of  $\alpha$ -dystroglycan. Glycosylation is the addition of sugars or chains of sugars (glycans) to proteins and lipids. This process takes place in the endoplasmic reticulum and Golgi complex and is regulated by a series of enzymes that control the addition of glycans.

O-mannose glycans develop into very complicated structures and are carried by  $\alpha$ -dystroglycan, which is closely associated with  $\beta$ -dystroglycan and is an integral part of the dystrophin-associated protein complex. The  $\beta$ -dystroglycan spans the muscle membrane and binds to both actin and laminin 2.

Five of the MDCs have been linked to mutations in the process of glycosylation. Different mutations in the same gene can produce widely varied phenotypes, which in some cases result from the varied amounts of residual enzyme activity. Glycosylation has been identified as necessary for the binding of laminin 2 to dystroglycan in the extracellular matrix.

WWS is the most severe MDC and is the result of defects in the first step in the glycosylation process where O-mannose is added to  $\alpha$ -dystroglycan caused by the absence of the glycosyltransferase that facilitates this reaction. The gene that is responsible is the POMT1 gene. Because WWS is a phenotypic diagnosis, POMT1 is only responsible for about 20% of the cases. Mutations in POMT2 are also seen in WWS, and the combination of these two genes in the endoplasmic reticulum modulates transferase activity.

Additionally defects in Fukutin and Fukutin-related proteins (FKRPs) can also be seen in WWS and presumably occur in the glycosylation pathway, although FKRP defects can produce a wide range of clinical phenotypes. Muscle-eye-brain disease can be caused by mutations in the POMGNT1 gene that encodes for the transferase in the next step in the pathway.

Clinically muscle-eye-brain disease represents a spectrum of phenotypic expression from mild (those who will live into adulthood) to severe (WWS). Because of this expression variability, other factors are presumed to affect the severity of the presenting phenotype. One such factor might be overexpression of like-glycosyltransferase (LARGE).

Fukuyana muscular dystrophy is caused by a mutation in the fukutin gene; the products are expressed in muscle in the same location as dystroglycan. Fukutin is found in the cis-Golgi, but beyond that its function has not been established.

MDC type 1C has a milder phenotype and is the result of a mutation in LARGE. LARGE is also found in the Golgi complex and rate-limiting step in the glycosylation of  $\alpha$ -dystroglycan. When it is overexpressed,  $\alpha$ -dystroglycan is hyperglycosylated. Despite this knowledge, its exact function still remains unclear. The potent effect of LARGE makes it an ideal target for treatment, since its overexpression can rescue both *in vitro* and *in vivo* models of a variety of glycosylation-based MDs.<sup>56</sup>

Two forms of MDCs without brain involvement include Ullrich-negative and merosin-negative MDC. The Ullrich-negative form is the result of a defect in the extracellular matrix protein collagen VI. Collagen VI is made up of three strands, COL6A1, A2, and A3, which are coded for on 21q22.3 or 2q37.

Dominant mutations traditionally have been thought to result in a more Bethlehem phenotype. Recessive mutations result in the congenital Ullrich presentation. However, there have been recent cases reported where dominant mutations have presented with an Ullrich picture. The underlying defect might relate to the protein interaction with the membrane, but the exact function still remains unclear. Merosin-negative MDC type 1A results in merosin deficiency and is caused by a genetic defect on chromosome band 6q22-23.<sup>92</sup>

**Faciocapulohumeral Dystrophy.** FSHD is less well understood. The genetic defect has been found at 4q35 in 90% to 95% of individuals with FSHD.<sup>24,183</sup> However, the abnormal protein has not been identified, because it is coded for at some distance from the genetic defect and has yet to be cloned.

**Myotonic Dystrophy.** There are three forms of myotonic dystrophy. The major form of myotonic dystrophy, also known as Steinert type or MD1, has been linked to chromosome band 19q13.3 and represents 98% of the cases. Two other types of myotonic dystrophy have been identified. Myotonic dystrophy type 2 (MD2) has been linked to chromosome band 3q21.3 (MD2)<sup>118</sup> and a third type has been linked to chromosome band 15q21-24.

In MD1 the underlying defect of the gene is a trinucleotide repeat in which cytosine, thymine, and guanine (CTG) are repeated an abnormally large number of times (see Fig. 1-4). With succeeding generations this defect expands, a condition known as anticipation, also seen in other triple-repeat disorders, where each succeeding generation is more affected than the last.<sup>96</sup>

This results in the absence of myotonin protein kinase through a very complex and poorly understood pathway involving the creation of abnormal RNA transcript processing and alterations in splicing of other genes, some of which code for the chloride channel.<sup>12</sup> In myotonic dystrophy muscle fibers demonstrate altered resting muscle membrane potentials, possibly resulting from a dysregulation of ion channel function.<sup>10</sup>

### Clinical Manifestations

People with myotonic dystrophy have muscular weakness, wasting, and hypotonia. The degree of severity and age of onset vary with the type of myotonic dystrophy present (see Table 23-5).

**Duchenne's Muscular Dystrophy.** DMD is usually identified when the child has difficulty getting up off the



**Figure 23-18**

Gowers' sign. This boy adopts the typical movement seen with proximal weakness, such as myopathies, when arising from the floor, a chair, or even when climbing stairs. During Gowers' maneuver the client places the hands on the thighs and walks up the legs with the hands until the weight of the trunk can be placed posterior to the hip joint. This sign is characteristic of weakness of the lumbar and gluteal muscles. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

floor (Gowers' sign; Fig. 23-18), falls frequently, has difficulty climbing stairs, and starts to walk with a waddling gait (proximal muscle weakness) and an increased lumbar lordosis (compensation for abdominal and hip extensor weakness). At the same time, the child begins to walk on the toes because of contracture of the posterior calf musculature and weakness of the anterior tibial, peroneal, and proximal muscles.

Hip abductor weakness produces a positive Trendelenburg's sign (see Fig. 23-14), which eventually changes to a compensated gluteus medius gait as hip abductor weakness progresses. Classically ambulation continues to deteriorate up to the age of 10 to 12 years, at which time the majority of people with DMD are no longer able to walk.<sup>17,164</sup> However, with more recent medical treatment with prednisone or deflazacort children may walk beyond their twelfth birthday, which previously was not seen.

The shoulder girdle becomes involved, with excessive scapular winging and muscle hypertrophy (especially the upper arms but also the calves and thighs) (Fig. 23-19). One of the major problems in gait occurs when the



**Figure 23-19**

Duchenne's muscular dystrophy with pseudohypertrophy of calves and lordotic posture that places the weight of the trunk behind the hip joint. Even though weakness occurs symmetrically, habitual standing postures may create asymmetries in flexibility in some cases. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

affected person requires upper extremity support. Shoulder girdle weakness and the need to maintain the weight line posterior to the hips and anterior to the knees often prevents the use of crutches to support the body weight.

Weakness of the shoulder girdle also causes difficulty in performing overhead activities related to hygiene and work. Bicipital tendinitis or other impingement disorders at the shoulder occur as the children get older as the result of weakness and the repeated manual lifts that become necessary for transfers. Muscle imbalances create biomechanical dysfunction, and weakness impairs the ability to stabilize the shoulder girdle, contributing to shoulder problems.

With the progression of weakness, scoliosis occurs at a rate of 80% to 90% and usually progresses more rapidly after the person is wheelchair bound. Spinal fusion is usually considered when the spinal curve approaches 40 degrees. Early fusion, when a good correction and level pelvis can be obtained, and when the individual's respiratory status is relatively more intact, produces the best result.

Common comorbidities associated with DMD include cognitive, respiratory, cardiac, and gastrointestinal dysfunction. The average IQ of individuals with DMD is 1

standard deviation below the mean, with specific reading disorders noted irrespective of IQ. Even so, many children with DMD have normal or above-normal intelligence.

Children with DMD develop a progressive restrictive respiratory impairment secondary to weakness and contracture of the respiratory muscles. Respiratory problems become more of a problem after the children become wheelchair bound. Nocturnal hypoventilation is one of the earlier manifestations of respiratory involvement and is usually accompanied by headaches, sleep disturbance, or nightmares. Chest muscle deterioration combined with joint contractures and involvement of the spine results in diminished ventilation and ability to produce pressure to cough up secretions, leading to pneumonia, other respiratory infections, and even death.

Disruption of sarcoglycan complexes in vascular smooth muscle also can result in vascular irregularities of the heart, diaphragm, kidneys, and gastrointestinal tract. Dilated cardiac myopathy and, less frequently, conduction abnormalities can be life-threatening. Gastrointestinal problems are common, and constipation and pseudo-obstruction can result from the smooth muscle deterioration and gastric dilatation that can occur.

**Becker's Muscular Dystrophy.** Signs and symptoms of BMD resemble those of DMD but with a slower progression and longer life expectancy. Ambulation is preserved into midadolescence or later but often is marked by the toe walking with bilateral calf muscle hypertrophy.

Proximal muscles tend to be affected to a greater degree and before the involvement of the distal musculature, with primary effects observed in the neck (relatively preserved in BMD versus DMD), trunk, pelvis, and shoulder girdle. Muscle cramps are a common complaint in late childhood and early adolescence.

Scoliosis and contractures (elbow flexors, forearm pronators, and wrist flexors in the upper extremity and plantar flexors, knee flexors, and hip abductors in the lower extremity) and other comorbidities also found in DMD are common; however, these occur less frequently and with less severity in the BMD type.

**Limb-Girdle Muscular Dystrophy.** LGMD affects both proximal and distal muscles and follows a slow course, often with only mild impairment, although the course can vary widely even in a given family. Early symptoms develop as a result of muscle weakness in the upper arm (biceps and deltoid muscles) and pelvic muscles, usually noticed in late adolescence or early adulthood but as late as the person's forties.

Winging of the scapulae, lumbar lordosis, abdominal protrusion, waddling gait, poor balance, and inability to raise the arms may also develop. The lack of consistent clinical features makes this type of MD more difficult to diagnose. Identification of the specific type of LGMD should be pursued to provide the best anticipatory guidance for the client and to allow the therapist and physician to treat the person proactively with full knowledge of the natural history of the specific disorder rather than approaching the case reactively.

**Congenital Muscular Dystrophy.** MDC represents a spectrum of disease states that most commonly present

at the more severe end of the spectrum in infancy with rapidly progressive muscle strength loss and progressive respiratory symptoms.

Clients demonstrate a mixed central and peripheral picture with involvement of both the brain and muscle in addition to involvement of the visual system. WWS is the most severe of the MDCs and presents at birth with a rapidly progressive course, with death most commonly prior to a year of age. Ocular impairments include retinal abnormalities, microphthalmia glaucoma, cataracts, and anterior chamber abnormalities.

The CNS complications include a cobblestone lissencephaly with agyria as well as areas of macrogryria and polymicrogyria. Cerebellar abnormalities are also present and include hypoplasia in addition to fourth ventricular dilatation. Muscle-eye-brain disease has similar clinical findings as WWS but a wider range of phenotypic presentations. Fukuyama MD can also present as an MDC but is more often found in its milder LGMD form.

Common phenotypic presentation includes onset at or shortly after birth with weakness and delayed gross motor skills with progressive contractures and weakness, with very few achieving ambulation and those typically requiring a wheelchair by the age of 10.

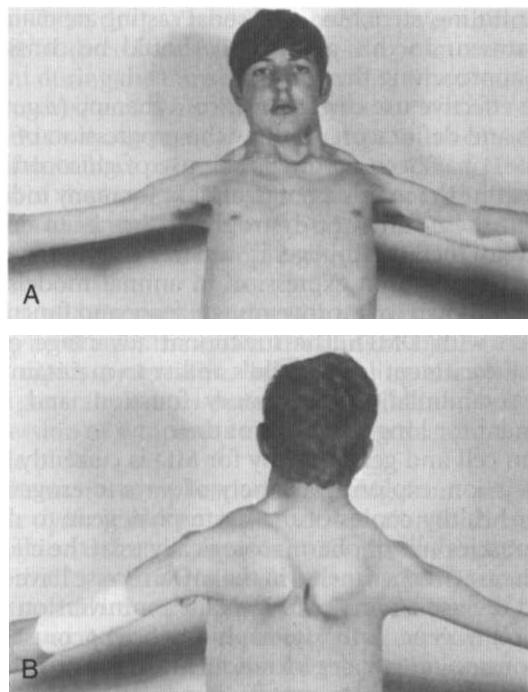
Cognitive impairment is common and often severe, with MRI findings somewhat similar to those found in WWS. Ullrich MDC can present along a spectrum of severity, with its milder form, Bethlem myopathy, representing the mildest presentation. In its more severe forms children have significant weakness that either prevents ambulation or children only walk for a short time prior to adolescence.<sup>119</sup>

The most prominent feature of the phenotype is the prominent distal laxity mixed with proximal contractures, particularly of the knee. People with merison-negative MDC typically present with weakness at birth, with respiratory insufficiency developing typically in the first decade; however, a spectrum including an LGMD phenotype has been reported.<sup>92</sup>

**Facioscapulohumeral Dystrophy.** FSHD is a mild form of MD beginning with weakness and atrophy of the facial muscles and shoulder girdle, usually presenting in the second decade of life. Phenotypic expression is more common in males than in females (95% versus 69%), with more females being carriers. Inability to close the eyes may be the earliest sign; the face is expressionless even when laughing or crying, forward shoulders and scapular winging develop, and the person has difficulty raising the arms overhead (Fig. 23-20).

Other changes in the face include diffuse facial flattening, a pouting lower lip, and inability to pucker the mouth to whistle or, for the infant, an inability to suckle. Progression is descending, with subsequent involvement of either the distal anterior leg or hip girdle muscles.<sup>170</sup> Weakness of the lower extremities may be delayed for many years. Contractures, skeletal deformities, and hypertrophy of the muscles are uncommon.

There is wide variability in age at onset, disease severity, and side-to-side symmetry, even within affected members of the same family. Associated non-skeletal muscle manifestations include high-frequency hearing loss and retinal telangiectasias, both of which are usually



**Figure 23-20**

**Faciocapulohumeral dystrophy.** Weakness of subscapular musculature makes it difficult to perform overhead activities; wasting also causes the clavicles to jut forward and the shoulders to have a drooping appearance. During humeral movement, the scapulae wing and ride up over the thorax. (From Morgan-Hughes JA: Diseases of striated muscle. In Asbury AK, McKhann GM, McDonald WI, eds: *Diseases of the nervous system: clinical neurobiology*, ed 2, Philadelphia, 1992, Saunders, p 170.)

asymptomatic. The natural history and progression are well documented.<sup>170,183</sup>

A variation of FSHD is scapuloperoneal MD, involving the proximal muscles of the shoulder girdle with sparing of the face. The process slowly spreads to the distal part of the lower extremities in several years or decades. Early symptoms may include shoulder weakness characterized by scapular winging; foot drop develops later.

**Myotonic Dystrophy.** The clinical presentation of myotonic dystrophy represents a spectrum of disease severity that is based on the size of the genetic triple repeat. Three phenotypes have been identified. The most severe is congenital myotonic dystrophy with weakness and myotonia at birth. The classic form is characterized by weakness and some degree of disability, with mild myotonia and cataracts.

The clinical symptomatology of myotonic dystrophy includes muscle weakness and wasting with a delayed relaxation of the muscle and increased excitability. Ocular cataracts are also a defining factor; cardiac conduction defects represent a serious comorbidity. A wide variety of other symptoms, including sensorineural hearing loss, hypersomnia, testicular atrophy (and sterility), and endocrine dysfunction, are also found in myotonic dystrophy.<sup>10</sup>

## MEDICAL MANAGEMENT

**DIAGNOSIS.** Researchers continue to develop noninvasive imaging procedures for evaluating the localization,

extent, subtype, and mechanisms of skeletal muscle damage in MD. Diagnosis is currently based on clinical presentation, family history, and diagnostic testing such as muscle ultrasound, genetic testing, electromyography (EMG), muscle biopsy, and serum enzymes. The use of these five diagnostic tests with each of the major types of MD is presented briefly in this section.

**Duchenne's and Becker's Dystrophy.** Chorionic villi sampling and amniocentesis are prenatal diagnostic techniques in which deoxyribonucleic acid (DNA) is removed during gestation to determine the presence or absence of the defective gene. Currently, standard laboratory genetic testing can detect large deletions of the dystrophin gene in approximately 65% and large duplications in about 5% of fetuses and aid in identification of carriers; the accuracy partially depends on the genetic heterogeneity of the particular disease.

Most of the remaining 35% of cases represent point mutations and are more difficult to identify but can be found by sequencing the gene. New mutations cause 30% of cases of DMD. Mothers who are carriers of a dystrophin gene mutation will pass on the mutated gene to 50% of their daughters (making them carriers) and 50% of the sons who will be affected by DMD.

EMG studies in DMD/BMD demonstrate the presence of fibrillation potentials, positive sharp waves (more in DMD), and long-duration polyphasic motor unit action potentials (MUAPs) (more in BMD) with full recruitment at low force. Nerve conduction velocities are normal in both DMD and BMD.<sup>12</sup> A muscle biopsy specimen shows variation in the size of muscle fibers; central nuclei, inflammatory cells, and fat and connective tissue deposits are prominent characteristics of the biopsy specimen. In DMD the muscle stains negative for dystrophin antibodies, whereas in BMD levels of dystrophin vary.

Serum enzyme levels are a final diagnostic test used to identify the presence of active muscle damage. Approximately 50% to 60% of female carriers of DMD/BMD have elevated creatinine kinase (CK). Males affected by DMD/BMD present with CK levels that are approximately 2 to 10 times normal, reflecting active muscle damage (see the section on Serum Enzymes in Chapter 40; see Table 40-15).

Levels are extremely high in the first years of life before the onset of clinical weakness and persist as symptoms develop. Eventually, after replacement of muscle substance has become chronic and extensive, the CK level may be either normal or only mildly elevated (less than five times normal). The CK isoenzyme may be increased in DMD, especially in the earlier phase of the illness.

**Limb-Girdle Muscular Dystrophy.** LGMD presents with markedly increased levels of CK, however, often not to the same magnitude seen in DMD. EMG and muscle biopsy results demonstrate myopathic changes. EMG findings reveal positive sharp waves, and fibrillation potentials are absent in some individuals and increased in others. Short-duration, small-amplitude MUAPs and an increased number of MUAPs are characteristic of LGMD.<sup>42</sup>

Muscle biopsy specimens present with variable fiber size and atrophy alternating with hypertrophy; in the later stages connective tissue is increased. Muscle in

LGMD can be stained for a variety of components of the sarcoglycan complex as well as many other known protein defects. However, often this does not provide a specific diagnosis, because defects in one sarcoglycan can affect incorporation of the others and some protein stains are not available; clues can lead to appropriate genetic testing to confirm the diagnosis.<sup>176</sup>

**Congenital Muscular Dystrophy.** A good clinical examination will provide insight into the basis of an infant's hypotonia and determine if it is thought to be central in origin (based on soft neurologic signs) or peripheral. Initial CK will be increased, and a muscle biopsy specimen will present as an active dystrophic process. MUAPs will be diminished, and in merison-deficient MDC, a mixed picture may be noted, including demyelination.

**Facioscapulohumeral Dystrophy.** In FSHD serum CK is elevated in 75% of affected individuals. Electrodiagnostic testing demonstrates a myopathic pattern, with positive sharp waves and fibrillation potentials often noted; however, these are less prominent than in DMD. The most striking characteristic in FSHD is short-duration, small-amplitude, polyphasic MUAPs.<sup>42</sup>

Muscle biopsy findings are somewhat dependent on which muscle is biopsied with variable fiber size and necrotic and regenerating fibers being common; central nuclei inflammatory infiltrates can also be noted. Analysis for the underlying genetic defect 4q35 can be used to confirm the diagnosis.

**Myotonic Dystrophy.** In myotonic dystrophy microscopic evaluation of muscle and nerve demonstrates alterations. In the muscle selective atrophy of the type I fibers is noted, with central nuclei and hypertrophic fibers and increased connective tissue present. Nerve biopsy results show a variable degree of demyelination, particularly in large fibers, in addition to regenerating fibers characteristic of axonal neuropathy.<sup>186</sup> EMG is extremely important in documenting myotonia, with the unmistakable "dive bomber" sound produced by a myotonic discharge.

**TREATMENT.** At present no known treatment halts the progression of MD. Despite recent advances in our understanding of the MDs, current therapy for these disorders remains primarily supportive. Research in the area of molecular biology that has brought specific information about the molecular pathogenesis involved may one day lead to effective treatment.

Presently, treatment intervention is directed toward maintaining function in unaffected muscle groups for as long as possible, utilizing supportive measures such as physical and occupational therapy, orthopedic appliances, orthopedic surgery, and pharmaceuticals. Children who remain active as long as possible avoid complications (e.g., contractures, pressure ulcers, infections) and deconditioning that are common once they are wheelchair bound.

It is important to remember that there is an active muscle degeneration underlying the MDs. Strengthening, especially eccentric exercise, is not helpful and may cause increased weakness, particularly in DMD. Contracture management is the focus of treatment for the therapist and is important in maintaining function in clients with

MD. Splinting, stretching, and serial casting are mainstays of treatment in this group and should be considered when approaching these clients.

The effective use of glucocorticoid therapy (e.g., prednisone and deflazacort) to slow the progression of DMD and BMD has been reported. The use of glucocorticoids has become the mainstay of treatment for many individuals with these forms of dystrophy. It has been demonstrated to increase myogenic differentiation, myoblast fusion, and laminin expression in animal models<sup>2</sup> and has been shown to improve muscle force and function in children with DMD. The functional advantage of this medical treatment is the child's ability to maintain independent ambulation, respiratory function, and spinal alignment for longer periods of time.

Stem cell and gene therapy for MD is currently under investigation, exploring a variety of ways to exogenously deliver healthy copies of the dystrophin gene to dystrophic muscles<sup>97,117</sup> or pharmacologically treat the effects of this disease.<sup>28</sup> Experiments in the MDX mouse have investigated the use of viruses to implant a miniversion of the dystrophin gene into dystrophin-deficient muscles to delay or stop muscle degeneration. The main obstacle has been immunologic; however, human trials are on the horizon.

In other research models, attempts have been made to inject skeletal muscles with donor cells, a gene transfer method referred to as myoblast transfer therapy (MTT). These myoblasts fuse with diseased muscle fibers and provide the missing gene to replace dystrophin. To date, there have been no reports of improved strength in people with DMD with this procedure.<sup>128,165,184</sup>

There are also treatments on the horizon for specific genetic defects. DMD is the end result of a variety of different genetic mutations. Most people with DMD have large mutations; others have point mutations, duplications, or early stop codon mutations. There is the potential that some of these may be amenable to drug treatments, either to reestablish the reading frame or to facilitate read through in the case of a premature stop codon.

In a small portion of people (up to 15%) with DMD and other genetic disorders, the underlying genetic defect is a premature stop codon or nonsense mutation (a stop codon normally directs the mRNA sequences for coding polypeptide chains to stop at the appropriate time).

The inability to read the genetic material and produce dystrophin in these individuals potentially can be suppressed by the use of the antibiotic gentamicin. This has been investigated in animal models and in cell culture with the production and incorporation of dystrophin noted in the muscle membrane.<sup>8</sup> There are also drugs under development that bind to the ribosome and can induce the read through of the premature stop codon by the ribosome in individuals with premature stop codon mutations.

People with point mutations might be amenable to a treatment with antisense oligonucleotides designed to induce exon skipping. Genes are read in sets of three base pairs. When someone has a point mutation, if it is an out-of-frame mutation, everything after the mutation is shifted and the groups of three base pairs do not make

sense. These drugs cause RNA to skip over the exon (a full set of three base pairs) where the point mutation is present during the transcription process when the introns are removed and the mRNA is formed.

In this way, the gene is allowed to continue reading in sets of three base pairs. If these drugs are effective in clinical trials, the hope is that this strategy could induce production of increased levels of dystrophin and result in an effective treatment for people with DMD.

**PROGNOSIS.** Prognosis varies with the type of MD present. As a general rule, the earlier the clinical signs appear the more rapid, progressive, and disabling the dystrophy. DMD generally occurs during early childhood with rapid progression of symptoms and results in death in the third decade of life.

Pulmonary complications resulting from respiratory muscle dysfunction or cardiac dysfunction in the form of conduction defects or myopathy are the common sources of morbidity and mortality. People with BMD usually live into the fifth decade (their forties) or beyond; death occurs secondary to respiratory dysfunction or heart failure. Those with FSHD involvement may appear almost stable over a period of years; variable progression occurs among those with LGMD. People with both FSHD and LGMD have a relatively normal lifespan.

## SPECIAL IMPLICATIONS FOR THE THERAPIST 23-6

### Muscular Dystrophy

#### PREFERRED PRACTICE PATTERNS

4C: Impaired Muscle Performance

5A: Primary Prevention/Risk Reduction for Loss of Balance and Falling

5B: Impaired Neuromotor Development

6B: Impaired Aerobic Capacity/Endurance Associated with Deconditioning

6E: Impaired Ventilation and Respiration/Gas Exchange Associated with Ventilatory Pump Dysfunction or Failure

#### Precautions

When people with MD become ill or injured and are on bed rest (at home or in the hospital) even for a few days, they may lose many of their functional abilities. For example, a child who falls and breaks a leg and is on bed rest or otherwise immobilized may never regain the ability to ambulate. These children should be encouraged to be as mobile as possible, and if possible, ambulate for even a few minutes during the course of any illness.

Although activity helps the client maintain functional abilities, strenuous exercise may facilitate the breakdown of muscle fibers, so that exercise must be approached cautiously. Low-repetition maximum weightlifting, especially eccentric strengthening, is not recommended. Exercise is best done in the pool, where exercise is concentric. Any exercise program should produce only minimal fatigue with no postexercise soreness, because the amount of damage to the muscle membrane with exercise is related directly to the mag-

nitude of the stress placed on it during contraction.<sup>136</sup>

Respiratory involvement requires careful monitoring of breathing techniques, respiratory movements, and oxygen saturation levels. Monitoring oxygen during exercise and activity is recommended. See Appendix B. The client should be instructed in diaphragmatic, deep-breathing exercises. Airway clearance techniques, including the use of percussion and postural drainage and mechanical insufflator-exsufflator for assisted cough, are especially useful during illness.<sup>5</sup>

Investigators have shown that the inspiratory muscles can be trained for both force and endurance in this population. These training-related improvements in inspiratory muscle performance are more likely to occur in those who are less severely affected by the disease. In those clients who have disease to the extent that they are already retaining carbon dioxide, little change occurs in respiratory muscle force or endurance with training.<sup>116</sup>

In the later stages of respiratory compromise nighttime mechanical ventilation (e.g., continuous positive airway pressure [CPAP] or bilevel positive airway pressure [BiPAP] delivered by face mask) is an intervention used to rest the respiratory muscles. A major priority for these children is to avoid or delay the need for intubation and full-time mechanical ventilation; these noninvasive methods can aid in this goal.<sup>35</sup>

#### Therapy Interventions

For individuals with the more disabling forms of MD such as DMD, the therapist can provide anticipatory guidance about the course of the disease and valuable information regarding the use of various types of adaptive equipment. Initially, grab bars provide for safety, but eventually a rolling commode or combination commode and bath chair is needed. As DMD progresses, a power wheelchair provides functional mobility once ambulation is no longer possible.

Eventually, adapted controls (minijoystick, touch pad, or fiberoptic switches) may be required for the power chair to accommodate the severe weakness and contracture that develop in the later stages of DMD. Power tilt-in-space wheelchair systems allow for pressure relief where air or gel cushions are no longer sufficient.

In the individual who no longer has access to a computer for school or work secondary to severe weakness, environmental control systems allow computer access by inferred link and mouse emulation to allow control of the mouse from the wheelchair control or completely hands-free control through the use of voice recognition software.

Overhead slings and mobile arm supports are helpful with feeding and other upper extremity activities, especially after spinal surgery, when axial flexibility is removed and greater active ROM is required for these functional tasks.

Splinting and night positioning in addition to active and passive ROM exercises will aid in delaying the

*Continued.*

onset of contractures and reducing the associated morbidity. Home environmental assessment and careful family and client interviews are important in planning out the appropriate adaptive equipment and home modifications.

Both children and adults can benefit from ambulation and pool therapy programs aimed at improving endurance. For a more in-depth discussion of the direct intervention protocols for this condition, the reader is referred to other resources.<sup>26,36,143</sup>

## Congenital Myopathy

### Definition and Overview

Congenital myopathy describes a group of disorders with somewhat similar phenotypic course, including central core disease, nemaline myopathy, multicore-minicore disease, and myotubular myopathy/centronuclear myopathy. A full appreciation of the specifics of each disease is necessary for treatment planning. As a group they are characterized by weakness at birth or shortly thereafter, with a course that is relatively stable or slowly progressive.

Often there are developmental gains made early in the course of the disease while the natural developmental progression of the child is in full force. Later the child might lose skills, as muscle strength does not keep up with the gain in body size or contractures interfere with function.

### Incidence

Nemaline myopathy is the most common of these diseases and occurs at a rate of 2 per 100,000.<sup>22</sup> The remaining types each account for a smaller incidence but greater total number of cases (prevalence).

### Pathogenesis

Nemaline myopathy is a heterogeneous disease with a number of genetic loci identified. The genes, loci, and protein products responsible for the resulting pathology include  $\alpha$ -tropomyosin slow (1q22-23, TPM3),  $\beta$ -tropomyosin (p13.2, TPM2), troponin 1 (19q13.4, TNNT1), nebulin (2q21-22, NEB), and  $\alpha$ -actin (1q42.1, ACTA1). These genes can have either autosomal dominant or recessive inheritance.

Central core disease is the result of a mutation in the RYR1 gene on chromosome band 19q13.1 with a defect in the ryanodine receptor and can be inherited both as a recessive or dominant gene. The RYR1 encodes the ryanodine receptor protein, which functions as part of a calcium release channel. It sits between the T-tubule and the sarcoplasmic reticulum and is integral to the process of excitation-contraction coupling through its regulation of cytosolic calcium homeostasis.

Multicore-minicore disease can be the result of a mutation in SEPN1 gene on chromosome band 1p36, which is inherited recessively, or it can have the same mutation as central core disease (RYR1). Both central core and

multicore-minicore disease are named because of the characteristics of the muscle biopsy findings.

In multicore-minicore disease there are several intracellular collections within the muscle cell. In central core there is one larger collection. These cores occur in type I fibers and lack oxidative enzyme activity. Early in the course of a ryanodine receptor defect, the muscle biopsy specimen can present with multiple intracellular cores. As the disease progresses these cores coalesce and form one central core, which accounts for the overlap in the genetics of these two diseases.

Myotubular (centronuclear) myopathy is X-linked and caused by a mutation in the myotubularin gene (MTM1) located on Xq28.<sup>22</sup>

### Clinical Manifestations

**Nemaline Myopathy.** Nemaline myopathy presents in a phenotypically heterogeneous way with more than five different types identified. Type 1 is the severe congenital form, type 2 the intermediate form, type 3 the most typical, and types 4 and 5 present in childhood or adulthood, respectively.

The typical form presents in infancy and is characterized by hypotonia throughout the body, including the face. Feeding difficulties, including aspiration and respiratory insufficiency, initially present at night and are common comorbidities. Contractures and spinal rigidity are also common; there is both weakness of the extremities and a lack of flexibility of the trunk, especially in flexion. Rigid spine presentation is typical in individuals who have selenoprotein defects.

**Central Core Disease.** Central core disease typically presents in infancy but can also present later. CK levels are usually normal or only mildly elevated. Common comorbidities include congenital hip dislocation, scoliosis, and talipes equinovarus. Because there is often variable penetrance, members of the same family can have varied phenotypic presentations. Anyone with central core disease is at risk for malignant hyperthermia, a severe and life-threatening reaction to certain anesthetics.

**Multicore-Minicore Disease.** Four groups of multicore-minicore disease have been identified. The classic form is characterized by proximal weakness and scoliosis as well as pulmonary insufficiency. Distal joint laxity is a common finding. Myopia is a common visual finding. Individuals with group II have ophthalmoplegia and severe facial weakness in addition to more global weakness. Individuals classified as having group III disease also have arthrogryposis and early onset.

**Myotubular (Centronuclear) Myopathy.** Myotubular myopathy is very phenotypically variable, with a range of presentations possible. The severe neonatal form is the most common type and can lead to death in the first year of life. Despite the fact that many have life-threatening pulmonary involvement, those who receive intensive ventilatory support can survive past the first year, gain strength, and show improvement in their respiratory status as they progress past the first year. Even so, upper respiratory infections remain significant challenges for many affected individuals. A less common form presents with a milder course and survival into adulthood.<sup>22,169</sup>

## MEDICAL MANAGEMENT

**DIAGNOSIS.** Diagnosis of congenital myopathy is made first by clinical examination. The differentiation of central versus peripheral causes of the hypotonia will help guide the physician's workup. These factors include deep tendon reflexes, upper motor neuron signs, and cognitive status.

If a peripheral process is suspected, an EMG can differentiate a neurogenic versus myogenic process and then a muscle biopsy can be performed to evaluate the pathologic characteristics. Special stains or electron microscopy can be ordered to further narrow the possible diagnoses. Finally, genetic testing can be ordered to confirm the diagnosis.

**TREATMENT.** Treatment is primarily symptomatic. Management of contractures is important in maintaining function, and supportive pulmonary care is important, especially in those clients who develop nocturnal hypoventilation. Cardiac monitoring is important for those with a propensity toward cardiac symptoms.

## Spinal Muscular Atrophy

### Overview and Incidence

Spinal muscular atrophy (SMA) is a neuromuscular disease characterized by progressive weakness and wasting of skeletal muscles resulting from anterior horn cell degeneration. SMA is the second most common fatal autosomal recessive disorder after cystic fibrosis. The overall prevalence is 1 in 20,000 live births, and 1 in 50 individuals carry the genetic defect (Table 23-6).

Childhood SMA is divided into severe (type I), intermediate (type II), and mild (type III). Type I, the more severe or acute form, is referred to as Werdnig-Hoffmann disease and causes respiratory failure and early death in the first few years of life if respiratory support is not provided.

Kugelberg-Welander disease, or type III SMA, is the mildest form. These individuals learn to walk without assistance; a relatively slow progression is noted in type III. Type II represents an intermediate form; affected individuals demonstrate the ability to sit independently at some point, but significant functional impairment and reliance on power mobility is typical (see Table 23-6). Despite the classification system into three types, SMA really represents a continuous spectrum of severity.

### Etiologic Factors and Pathogenesis

The basis of this inherited pathologic condition (autosomal recessive trait) is gene deletions in the SMA critical region of the long arm of chromosome 5 (5q13.1).<sup>60</sup> The SMN1 gene is defective in 99% of all cases of SMA and is the cause of SMA.

The NAIP gene is defective in 45% of the more severely involved type I individuals; however, no direct evidence has been presented that an NAIP deletion can cause SMA or modulate its severity.<sup>126</sup> The SMN1 gene has a homologous gene, SMN2, that can compensate for the absence of SMN1 by producing some survival motor neuron (SMN) protein. SMN2 can be present in multiple

copies; the more copies of SMN2 present, the more SMN protein is produced, and the milder the phenotype becomes.

Progressive degeneration of anterior horn cells of the spinal cord is noted in SMA, with selected motor nuclei of the brainstem being variably affected. In the remaining axons, sprouting occurs, resulting in enlarged motor units. The underlying pathogenesis of anterior horn cell loss appears to be the persistence of programmed cell death in the anterior horn cells.<sup>161</sup>

The SMN1 gene mutation decreases intracellular levels of SMN protein<sup>60</sup> present in the cytoplasm and nucleus of all cells. The SMN protein is not fully understood but it is involved in prevention of neuronal cell death.<sup>90</sup> The variability of phenotype in SMA is related to the presence of multiple copies of the SMN2 gene, which is less effective at producing the gene product SMN, resulting in various intracellular levels of SMN.

Other modifying genes (discussed earlier) and the presence of a protein called bcl-2 also may affect the modulation of SMN's control of neuronal cell death.<sup>156</sup>

### Clinical Manifestations

Progressive atrophy of skeletal muscles is noted, with a variable degree of hypotonia, weakness, and fatigue reported. Often fatal restrictive lung disease is present. Studies indicate initial weakness but no progressive loss in muscle strength. There is a slowly progressive loss of motor function. Explanations for this loss of function remain undetermined, but decrease in motor function could be caused by factors such as increased body size.<sup>82</sup>

Other factors that may contribute to weakness and fatigue include chronic respiratory insufficiency with hypoventilation and carbon dioxide retention and chronic malnutrition.<sup>83</sup>

Children with SMA type I present features of this disorder within the first 3 to 4 months of life. The child has marked hypotonia, severe generalized weakness, and is unable to sit unsupported. Children with type II present before 18 months with chronic weakness and attain sitting but never walk without assistance.

By definition, individuals with type III SMA are able to ambulate at some point in their lives, although they often require the use of a wheelchair by midadulthood. Clinical problems associated with the muscle weakness seen in SMA include feeding and nutrition, respiratory, cardiac, and orthopedic problems.

## MEDICAL MANAGEMENT

**DIAGNOSIS.** The diagnosis is suspected on the basis of clinical manifestations but is established from muscle biopsy and EMG in which a neuropathic pattern is found. Nerve conduction velocities can be normal (slowing may be noted later in the course); motor action potentials are decreased in magnitude.

On needle EMG fibrillations and sharp waves are usually present and action potentials are high amplitude, long duration, and show polyphasic morphology with an increased firing rate.<sup>167</sup> Muscle biopsy specimens show groups of small atrophic fibers with large hypertrophic fibers, representing those without anterior horn cells and

**Table 23-6** Spinal Muscular Atrophy

Type	Incidence	Onset	Inheritance	Features	Course
SMA (type I) (Werdnig-Hoffmann) Acute or severe form	1 in 15,000-25,000 live births	0-3 mo	Autosomal recessive	LEs flexed, abducted, and externally rotated (frog position) UEs abducted, externally rotated, unable to move to midline against gravity Poor head control Significantly decreased muscle tone/weakness Decreased newborn movements, decreased diaphragmatic movements High risk of scoliosis Proximal muscle weakness greater than distal weakness Weak cry and cough Normal sensation and intellect	Rapidly progressive Severe hypotonia Death within first 3 yr 32% survive second year 8% survive 10 yr <sup>124</sup>
SMA type II Intermediate form	Same as type I	Before 18 mo	Autosomal recessive	LEs flexed, abducted, and externally rotated Limited trunk control Weakness Increased risk of scoliosis Normal sensation and intellect	Progressive but stabilizes Moderate to severe hypotonia Shortened life span <sup>124</sup> Attain the ability to sit at some point Reliance on power mobility
SMA type III Kugelberg-Welander Mild form	6 in 100,000 live births	Present after 18 mo	Autosomal recessive	Proximal LE weakness (greatest with trunk, hip, knee extension) Trendelenburg's gait, especially with running Slow continued developmental progression Sits independently Walks independently (lumbar lordosis, waddling gait, genu recurvatum, protuberant abdomen) Wheelchair bound by early adulthood (dependent on age of onset) Good UE strength	Slowly progressive Mild impairment Attain the ability to ambulate at some point Wheelchair dependent in adulthood

LE, Lower extremity; UE, upper extremity.

those with anterior horn cells, respectively. Genetic testing for SMA also confirms the diagnosis.

**TREATMENT.** Treatment is symptomatic and preventive, primarily preventing pulmonary infection and treating or preventing orthopedic problems, the most serious of which is scoliosis. Feeding problems are common, especially in cases with bulbar muscle weakness; gastrostomy tube feedings are often necessary to optimally manage nutrition.

Respiratory problems (involvement of the intercostals) are common, and percussion and postural drainage and treatments with an in-exsufflator (also known as coughalator, a machine that helps in the removal of bronchial secretions from the respiratory tract) can aid in airway clearance, especially during intercurrent illness. Positive pressure ventilatory support, typically by BiPAP

(initially at night), can extend the lifespan in these clients. Cardiac involvement is often secondary to the chronic respiratory insufficiency typical of this disease.

The majority of people with SMA type I or II develop some type of scoliosis; individuals with type III who become nonambulatory are also likely to develop scoliosis. Bracing has not been found to delay the progression of scoliosis but might help with sitting balance (Fig. 23-21). Care should be taken to allow good diaphragmatic movement and not create increased respiratory effort if a soft spinal orthosis is chosen to manage sitting posture.

Spinal fusion is the primary means of management for scoliosis. Although fusion is often necessary, there is some consequence to function. Many will not return completely to their prior functional level.<sup>58</sup> Individuals with type II SMA can develop hip subluxation or disloca-



**Figure 23-21**

Spinal muscular atrophy (SMA). This 4-year-old child with SMA type II is fitted with a one-piece body jacket or thoracolumbosacral orthosis (TLSO). The TLSO offers support and control of the trunk and lower spine for improved sitting posture, balance, and greater stability. Full body jackets of this type may increase the work of breathing; an abdominal cutout to allow diaphragmatic excursion is typically provided for individuals with SMA who rely on diaphragmatic respiration due to the pattern of muscular weakness. The chair is a titanium ultralight wheelchair, which this child can propel for independent mobility. (Courtesy Tamara Kittelson-Aldred, Access Therapy Services, Missoula, MT. Used with permission.)

tion, but this is not typically painful and the literature on surgical correction is not supportive.

Individuals with type II SMA should participate in a standing program (Fig. 23-22). Knee-ankle-foot-orthoses (KAFOs) with ischial weight bearing are ideal for this in the younger age group. However, as contractures develop, standing will become more difficult despite the most aggressive splinting, ROM, and serial casting program.

SMA type III clients are most likely to ambulate, although about half of this group loses ambulatory skills in childhood or adolescence. Fractures are common and a significant source of morbidity and loss of functional skills.

**PROGNOSIS.** Prognosis varies according to age of onset or type of SMA (see Table 23-6). The earlier the disease occurs, the faster the progression of muscle weakness and the poorer the prognosis. The presence of respiratory distress also contributes to a poorer prognosis.



**Figure 23-22**

Static vertical standing frame provides support and stability in the upright position for the child with spinal muscular atrophy. Ankle-foot orthoses provide support for weight bearing through the lower extremities. (Courtesy Tamara Kittelson-Aldred, Access Therapy Services, Missoula, MT. Used with permission.)

SMA type I is the most severe and has a very poor prognosis, with death likely in the first 2 years of life as a result of respiratory failure or respiratory infection. Most children with this form of SMA do not survive past 3 years without the aid of mechanical ventilation. Clients with type III (with onset after 2 years of age) remain independently ambulatory throughout adult life; onset before 2 years results in loss of ambulatory ability at an average age of 12 years.<sup>151</sup>

#### SPECIAL IMPLICATIONS FOR THE THERAPIST 23-7

##### **Spinal Muscular Atrophy**

###### PREFERRED PRACTICE PATTERNS

4A: Primary Prevention/Risk Reduction for Skeletal Demineralization

4B: Impaired Posture (scoliosis)

5E: Impaired Motor Function and Sensory Integrity Associated with Progressive Disorders of the Central Nervous System

6C: Impaired Ventilation, Respiration/Gas Exchange, and Aerobic Capacity/Endurance Associated with Airway Clearance Dysfunction

*Continued.*

### 7A: Primary Prevention/Risk Reduction for Integumentary Disorders

A variety of opinions exist regarding the usefulness or effectiveness of an active developmental program for children with SMA. However, often children with SMA type I or II outlive predictions, and therapy intervention is helpful in improving function and preventing musculoskeletal problems.

#### Precautions

The infant or child with SMA who is immobile requires frequent changes of position to prevent skin problems and other complications, especially pneumonia. The pharynx may require suctioning to remove secretions in the more severe cases, and feeding must be carried out slowly and carefully with good positioning to prevent aspiration in those individuals with oral motor involvement.

The involvement of a therapist with specialization in feeding (usually an occupational therapist or speech-language pathologist) is essential for these children. These children are intellectually normal and require verbal, tactile, and auditory stimulation and various types of assistive technology.

Respiratory weakness or diminished head control may prevent the child from benefiting from prone positioning. This is especially problematic when the child cannot lift the head to clear the airway. The use of prone positioning must be evaluated and monitored carefully by the therapist; vertical positions (sitting and standing) tend to be the most functional.

Monitoring oxygen saturation levels may be necessary in evaluating programming effectiveness. Observe how much work is required to breathe, and whenever possible use a pulse oximeter (see Fig. B-1, Appendix B) to measure oxygen saturation noninvasively. Pulse oximetry can provide an outcome measure for documentation (see Appendix B).

#### Therapy Intervention

Specific treatment protocols for this condition are beyond the scope of this book. The therapist is referred to a more appropriate resource.<sup>26,171</sup> An overall management program should include positioning to encourage head and trunk control and to promote functional strengthening, in addition to splinting to maintain ROM. Assistive technology can provide the maximum possible independence for children with SMA.

Power mobility for the child with SMA who has no independent mobility is essential and should be considered in the child as young as 2 years of age<sup>172</sup> (Fig. 23-23). Low-technology solutions such as "slings and springs" also may be very liberating for the child who has limited antigravity upper extremity movement by providing a wide variety of exploratory opportunities.

Facilitation and active assistive work toward standing and ambulation have been found to be effective in



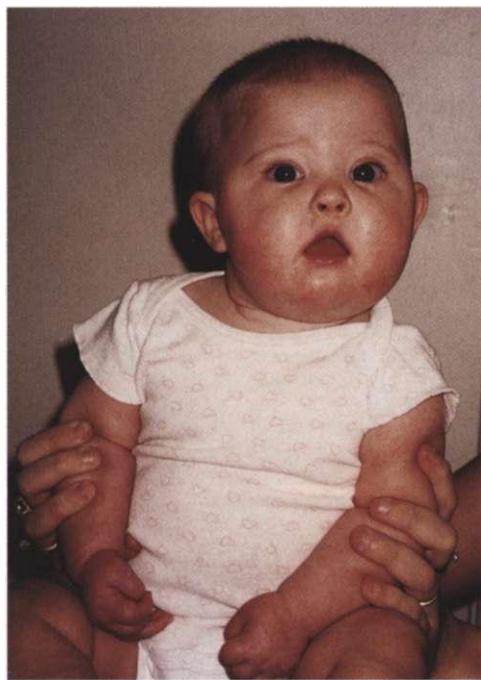
**Figure 23-23**

Spinal muscular atrophy (SMA). Three-year-old with SMA in her power wheelchair, which allows her to adjust the seat height so that she can be on the floor to aid with transfers or at eye level with her peers. The adjustable seat allows the child to participate in activities at elevated surfaces (e.g., counter or table heights), retrieve objects from a shelf, or help decorate the tree at Christmas. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

increasing forced vital capacity and in reducing the incidence of hip dislocation and contracture. More severely involved clients may benefit from positioning in a standing frame or instruction in standing to assist with or perform transfers independently.<sup>168</sup>

Elastic abdominal binders similar to those used in spinal cord injury can be used to provide increased trunk, abdominal, and diaphragmatic stability, especially if there is evidence of decreased oxygen saturation in sitting.<sup>168</sup> Inspiratory muscle training also has been found to be effective in neuromuscular disorders in improving maximal voluntary ventilation, maximal inspiratory mouth pressure, as well as respiratory load perception<sup>63,191</sup> and should be considered in this population.

Aquatic therapy can be a valuable adjunct to traditional intervention strategies for people at all levels of the SMA continuum. By using the physical properties of water such as buoyancy, hydrostatic pressure, viscosity, and turbulence, the therapist provides additional tools for intervention, especially in the case of extreme weakness characteristic of this disorder.<sup>51</sup>



**Figure 23-24**

Torticollis. Five-month-old with torticollis (head tilt toward the involved side and rotation away from the involved side). (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

## TORTICOLLIS

### Definition and Overview

*Torticollis* (congenital muscular torticollis or CMT; wry neck) means twisted neck and is a contracted state of the sternocleidomastoid muscle (SCM), producing head tilt to the affected side with rotation of the chin to the opposite side (Fig. 23-24). Four types of muscle abnormalities have been identified on ultrasonography: 15% had a fibrotic mass in the SCM (type I), 77% had diffuse fibrosis mixed with normal muscle (type II), 5% had fibrotic tissue without normal muscle (type III), and the last group (type IV) presented with a fibrotic cord and represented only 3% of the population.

Torticollis often is confused with a separate disorder known as cervical dystonia (also referred to as acquired torticollis or spasmotic torticollis). These are two separate entities and are presented separately in this text. CMT as it is presented in this section is a musculoskeletal phenomenon, whereas cervical dystonia is a movement disorder with an underlying CNS pathology (see the section on Dystonia in Chapter 31).

### Incidence and Etiologic Factors

Reports of the overall incidence of CMT vary significantly from 0.6 to 400 per 100,000 live births, but this condition is not considered uncommon.<sup>79</sup> A variety of possible causes of CMT exist, but the etiology remains unknown.

Initially it was thought that the fibrosis was related to birth trauma, because incidence is increased in breech

(19%) and forceps (6%) delivery, vacuum extraction (30.5%), and cesarean section (17.9%).<sup>31</sup> Predisposing factors can include restrictive intrauterine environment, poor muscle tone, or cervical-vertebral abnormalities. A genetic contribution also has been proposed in a portion of cases of CMT.<sup>48</sup>

### Pathogenesis

The possible pathogenesis of the muscular fibrosis seen in CMT has been explored experimentally in animal models and has been produced through venous occlusion, and this, in addition to arterial occlusion, has been proposed as the possible pathogenesis.<sup>79</sup>

One theory postulates that the malposition of the head potentially leads to a compartment syndrome. In this scenario, the SCM is not stretched or torn but rather kinked or compressed. With the head and neck in a position of forward flexion, lateral bend, and rotation, the ipsilateral SCM kinks, causing an ischemic injury and subsequent edema at the site of the kink.<sup>34</sup>

### Clinical Manifestations

The first sign of CMT identified in a portion of affected children is a firm, nontender, palpable enlargement of the SCM often referred to as a sternocleidomastoid tumor of infancy. A portion of cases demonstrate bulbous fibrotic tissue at the base or midportion of the involved SCM. This local lesion usually reaches its maximal size by 1 month and then slowly regresses within 4 to 8 months and does not always result in torticollis.<sup>177</sup>

The typical position observed of lateral head tilt and rotation to the opposite side predominates regardless of whether a fibrotic mass is present (estimated in 15% to 66% of cases<sup>79,81</sup>) or no mass is palpated and the muscle is uniformly fibrotic and shortened.

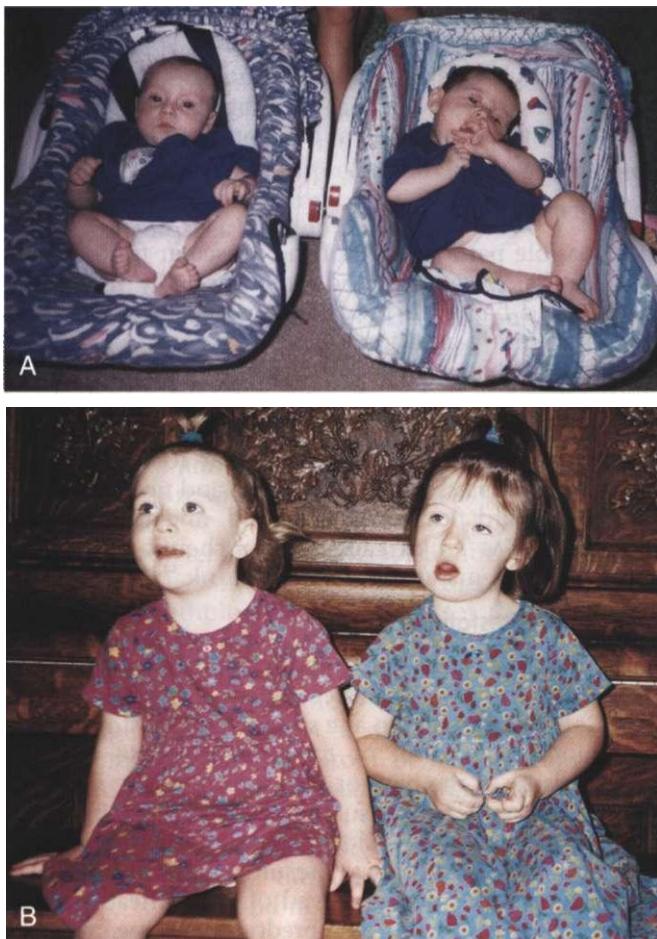
If the deformity is severe, the infant's face, ear, and head flatten from resting on the affected side, a condition referred to as plagioccephaly ("oblique head"); this cranial asymmetry gradually worsens. The infant's chin turns away from the side of the shortened muscle, the head tilts to the shortened side, and the shoulder is elevated on the affected side, further limiting cervical movement.

The side of the plagioccephaly (best observed by looking down on the head from above) is usually defined by the side of the flattened forehead. When torticollis and plagioccephaly occur together, this condition is referred to as plagioccephaly-torticollis deformation sequence (Fig. 23-25).

The incidence of other deformities such as hip dislocation and positional clubfoot is elevated in cases of CMT.<sup>31,64</sup> Subluxation of the cervical spine can also be associated with CMT and should be ruled out by cervical spine radiographs.<sup>79,162</sup>

### MEDICAL MANAGEMENT

**DIAGNOSIS.** Clinical observation combined with the history forms the basis of the initial diagnostic process. Medical evaluation including radiographic studies of the spine is always indicated to rule out congenital deformities of the cervical spine, ocular anomalies, and less frequently tumors or other CNS pathology in children with presumed torticollis.



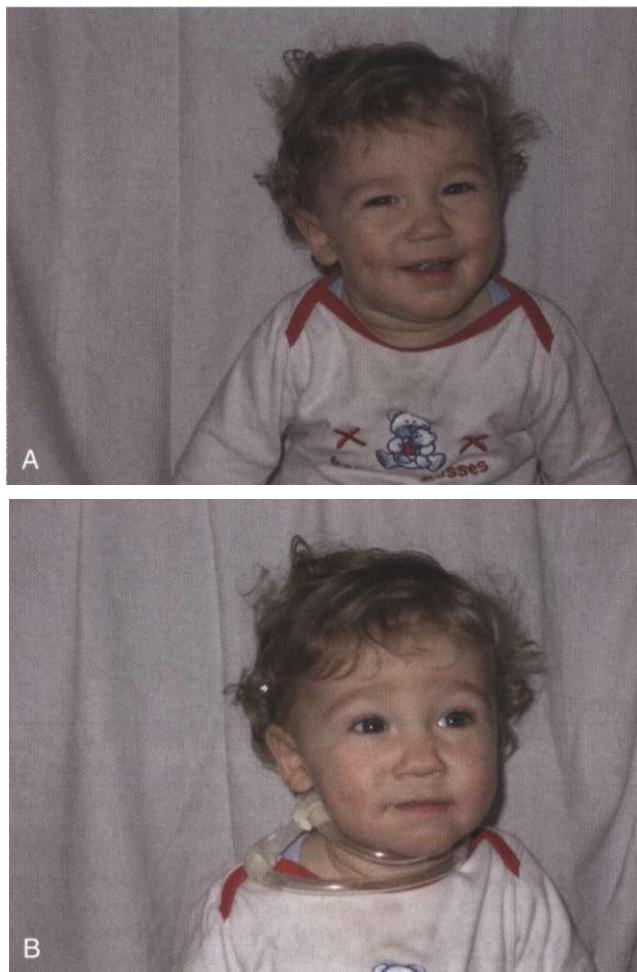
**Figure 23-25**

Plagiocephaly-torticollis deformation. **A**, Four-month-old fraternal twins; the child on the right has marked untreated congenital muscular torticollis (CMT) with plagiocephaly. Note the positional pelvic asymmetry from placement in a car seat. **B**, The same twins (2 years old) after physical therapy (PT) intervention at age 6 months for the child on the right. PT intervention over a 3-month period of time included passive range of motion, facilitated active range of motion, positioning, and a cervical collar. A home exercise program was prescribed with periodic rechecks. Eventually the use of a helmet was instituted to remodel craniofacial asymmetry (see Fig. 23-27). Some craniofacial asymmetry persists, although full active and passive range of motion are present. (Courtesy Laurie Matteson, Great Falls, MT. Used with permission.)

**TREATMENT.** Initial management involves a period of active observation for spontaneous resolution. During this time physical therapy to correct the positional/deformational effects is the mainstay of treatment for CMT. Interventions include twice daily passive ROM to stretch the shortened muscle preceded by warm compresses, massage, and slight traction to relax the muscle before stretching; stabilization of the proximal attachment of the SCM and trapezius is important during ROM.

Positioning is also important to encourage erect and midline head posture. Strengthening activities should include both active and active assistive exercises in addition to the incorporation of postural reactions in treatment when these reactions begin to develop.<sup>88</sup>

Splinting has been advocated by some for older children (older than 4 months) who continue to demon-



**Figure 23-26**

Congenital torticollis. **A**, Note the head tilt in this toddler with right-sided congenital torticollis. Despite his full range of motion, he has an occasional residual head tilt to the right and turn to the left. **B**, The same child wearing a TOT Collar (tubular orthosis for torticollis) to encourage a more vertical head position. (Courtesy Allan Glanzman, Children's Seashore House of the Children's Hospital of Philadelphia, PA.)

strate head tilt.<sup>84</sup> A cervical collar or tubular orthosis for torticollis (TOT) can be helpful in providing tactile cueing for movement in the direction opposite the lateral tilt. Usually these collars are the most effective at a time that is compatible with active head control (Fig. 23-26).

In cases of delayed treatment or where craniofacial asymmetry persists, nonsurgical remodeling of the skull using externally applied pressure can be used (Fig. 23-27). With advances in computer software and technology (e.g., pressure scanners), researchers are determining the pressure per square inch (PSI) that applies the appropriate force needed to achieve the remodeling process for each individual head diameter, volume, and topography.<sup>181,182</sup>

Surgical intervention is rare (e.g., SCM tenotomy, plastic surgery for craniofacial asymmetry) and is considered only if the individual continues to demonstrate significant motion restrictions of 30 degrees after 6 months of age or the deformity persists past 12 months of age.



**Figure 23-27**

Fourteen-month-old girl wearing a polypropylene helmet lined with durometer foam. This helmet placed remodeling pressure on the cranium to reshape unresolved craniofacial asymmetry that persisted as a result of delayed medical intervention and inconsistent use of a cervical collar. The helmet was accepted readily by the child and worn at all times (except for bathing) for approximately 4 months. Pressure was applied to the right posterior occiput to bring the head and neck into midline alignment while space was created where the skull was flattened in the left posterior occipital area to allow for bony growth in that area. See Fig. 23-25, B for intervention outcome. (Courtesy Laurie Matteson, Great Falls, MT. Used with permission.)

Increased thickening of the SCM or increased deformity are also indications to consider surgical intervention.<sup>88</sup>

**PROGNOSIS.** CMT usually resolves with conservative treatment. Complete recovery, including full passive ROM, can be expected to take approximately 3 to 12 months, with fewer than 16% of children presenting in the first year requiring surgery.<sup>45</sup> Left untreated or poorly managed, chronic, unresolved torticollis can result in persistent deformity and asymmetry of the head shape and position.

#### SPECIAL IMPLICATIONS FOR THE THERAPIST 23-8

##### **Torticollis**

###### PREFERRED PRACTICE PATTERNS

*4B: Impaired Posture*

*4C: Impaired Muscle Performance*

*4D: Impaired joint Mobility, Motor Function, Muscle Performance, and Range of Motion Associated with Connective Tissue Dysfunction (cervical subluxation)*

*7A: Primary Prevention/Risk Reduction for Integumentary Disorders (orthosis, bracing)*

The prognostic information provided makes it possible for therapists to better predict treatment duration at the time of initial assessment. When parents are provided with more precise information about the length of treatment, parents may be more willing to adhere to the exercise program.<sup>46</sup>

The therapist must remain alert to recognize cervical subluxation in cases of CMT. This may be observed as residual head-neck posturing problems, even after successful neck muscle therapy; usually no neurologic deficits are present.<sup>162</sup>

Likewise, torticollis that does not respond to physical therapy may have a nonorthopedic cause such as ocular torticollis, requiring further medical evaluation. Do not hesitate to ask for reevaluation if and when a child does not respond to therapy.

##### **Postoperative**

After surgery for this condition postoperative bracing to position the lengthened muscle on stretch has been advocated to allow the muscle to heal in the lengthened position.<sup>84</sup> Standard postoperative monitoring of vital signs and skin condition is recommended.

#### ERB'S PALSY

##### Definition and Overview

Erb's palsy is a paralysis of the upper limb typically resulting from a traction injury to the brachial plexus at birth. Erb's palsy actually comprises three distinct types of brachial plexus palsies: (1) Erb-Duchenne palsy affecting the C5 to C6 nerve roots (95% to 99% of all cases), (2) whole-arm palsy affecting C5 to T1, and (3) Klumpke's palsy affecting the C8 and T1 (lower plexus) nerve roots.

##### Incidence

The incidence of brachial plexus injuries has decreased secondary to improved obstetric management of difficult labors. Traction injuries are most common in newborns, occurring in 0.1% of spontaneous, 1.2% of breech, and 1.3% of forceps deliveries. Overall, the incidence of birth-related traction injuries is between 0.5 and 2 per 1000 births.

##### Etiologic and Risk Factors

The major contributing factor to these injuries has been attributed to forced stretching of the brachial plexus, that is, a pulling away of the shoulder from the head secondary to a traction maneuver during the birth process.

The lower plexus injury resulting in Klumpke's palsy usually is caused by manipulation during delivery resulting from hyperabduction of the arm at the shoulder, that is, the head and trunk remain relatively immobile in the pelvis while the upper extremity is stretched severely. However, some question remains about the role of the obstetrician as compared with the position of the infant and the forces encountered in the canal before birth.

Evidence suggests that the propulsive nature of the birth process when stretching of the involved nerves occurs is something over which the birth attendant has no control.<sup>155</sup>

Obstetric history associated with Erb's palsy is characterized by high birth weight or vertex delivery with shoulder dystocia (i.e., during delivery the baby's shoulder impinges on the mother's symphysis pubis). Klumpke's palsy more commonly is associated with heavy sedation, difficult breech delivery, and brow or face presentation. Brow or face presentation makes vaginal delivery impossible. Brow presentation rarely persists; in face presentation, the head is hyperextended and the chin presents.

Rarely, neoplasm present at birth results in brachial plexus palsy. The absence of signs of a traumatic injury accompanied by the onset of weakness and progressive course in the first few days of life must be investigated by MRI.<sup>1</sup>

#### Pathogenesis

Plexus injury during birth is usually the result of a stretch or avulsion of the plexus. A mild lesion is characterized by stretching of the nerve fibers, whereas a moderate injury involves some nerve fibers being stretched and others actually torn.

A more severe injury is characterized by a complete rupture of the plexus trunks with avulsion of the roots from the spinal cord. The degree of disability depends on the site and severity of injury. Diaphragmatic and serratus anterior paralysis suggests an avulsion injury as indicated by the location of the nerves with respect to the plexus.

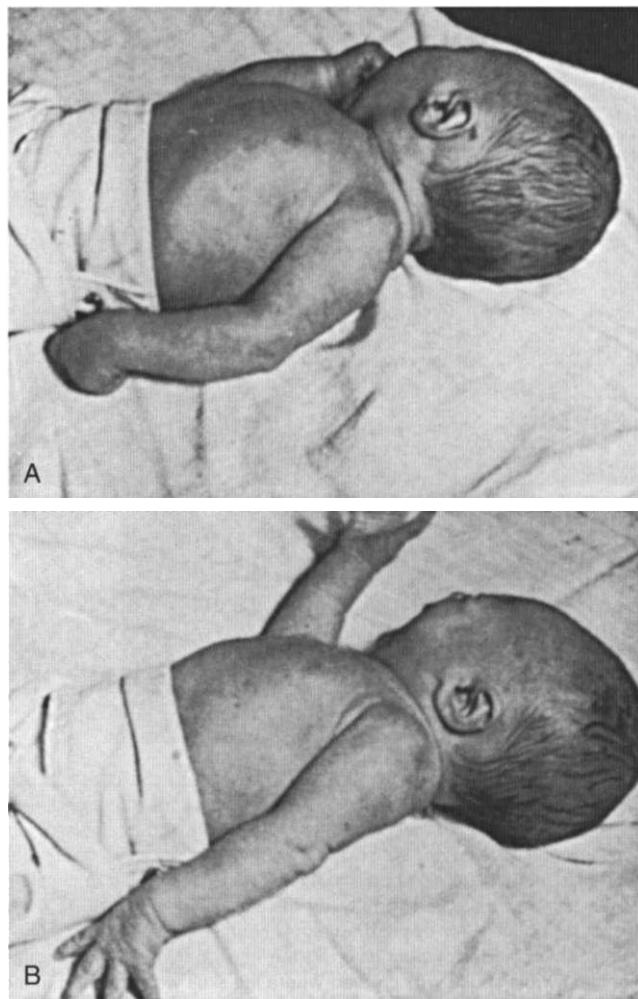
Persisting disability in neonatal brachial plexus palsy is due in part to impaired motor unit activation. This impairment may be a form of developmental apraxia caused by defective motor programming in early infancy.<sup>18</sup>

#### Clinical Manifestations

Children with brachial plexus injuries are unlikely to demonstrate postural or placing responses with the involved upper extremity when tested. In Erb's palsy the arm is maintained in adduction and internal rotation at the shoulder with the lower arm pronated and fingers flexed, assuming the waiter's tip position (Fig. 23-28). Children with this type have difficulty with activities such as hand-to-mouth, hand-to-head, and hand-to-back of neck movements but usually have control of the wrist and fingers.

In Klumpke's palsy, paralysis of the small muscles of the hand and wrist flexors causes a claw hand appearance. Proximal shoulder control is good, but voluntary wrist and hand control is difficult. In severe forms of brachial palsy (whole-arm palsy), the whole plexus can be affected but to a varying degree (Fig. 23-29). In this case careful examination is necessary to identify affected muscles. In all three cases (Erb's, Klumpke's, and whole-arm palsy) normal sensation is diminished; however, gross pain sensation may not be decreased to the same degree as movement.

The clinical characteristics of brachial plexus injury are summarized in Table 23-7.



**Figure 23-28**

Erb's palsy. **A**, In this infant with Erb's palsy, the arm is maintained in a position of adduction and internal rotation at the shoulder with the lower arm pronated and fingers flexed. **B**, Same infant demonstrating an asymmetric Moro reflex with opening of the left hand but still in the "waiter's tip" position. (From Behrman RE, Kliegman RM, Jenson HB: Nelson textbook of pediatrics, ed 17, Philadelphia, 2004, Saunders.)

#### MEDICAL MANAGEMENT

**DIAGNOSIS.** Some injuries are recognizable readily at or soon after birth. Radiographs may be taken to rule out associated fractures of the clavicle. Imaging of the brachial plexus using MRI is not invasive and can demonstrate proximal and distal lesions.

MRI can be used to detect nerve root avulsions, nerve ruptures, brachial plexus scarring, posttraumatic neuroma, brachial plexus edema, spinal cord damage, abnormalities of the shoulder joint, trauma, neoplasms, and infection. This type of imaging allows diagnosis and careful preoperative evaluation of children with brachial plexus injuries.<sup>11</sup>

EMG can be used to delineate the extent of injury and aid in the prognosis and assist the surgeon in identifying appropriate surgical procedures. EMG usually is delayed until 4 to 6 weeks after birth and may be followed serially



**Figure 23-29**

Brachial plexus palsy. Child with limited shoulder external rotation and abduction of the left arm associated with whole-arm palsy. Full motion of the upper extremity is demonstrated on the right side. (From Green DP, Hotchkiss RN, Pederson WC: *Green's operative hand surgery*, ed 5, London, 2005, Churchill Livingstone.)

over time to track recovery. Conduction studies can aid in separating actual axonal loss from conduction block. Needle EMG can help determine the portion of the plexus damaged as well as the severity of the damage.<sup>42</sup>

**TREATMENT.** Although medical intervention may include the use of botulinum toxin (Botox) to address contractures that may develop over time<sup>150</sup> or surgery, treatment is primarily with a therapist following the strategies outlined in Table 23-7. Surgery has found renewed favor as evidence is increasing that microneurosurgical intervention at an early stage can improve the outcome in some cases. For example, some children have no chance of recovery unless they undergo early aggressive surgical reconstruction of the injured brachial plexus. In children with global or total paralysis, surgery is performed by 3 to 4 months to maximize ultimate extremity function and minimize disability.<sup>66,173</sup>

Options for surgical care include tendon transfers considered after a plateau in recovery has occurred or micro-neurosurgery (e.g., nerve decompression, neurolysis, nerve repair, and nerve reconstruction with grafts or tubes). The latter procedure is best considered between

the ages of 6 and 12 months for optimal functional results.<sup>152</sup> Unfortunately, skepticism exists about the role of surgery, and many cases are referred too late for primary nerve surgery. Secondary reconstructive procedures at a later date can still improve the outcome in many cases.<sup>89</sup>

**PROGNOSIS.** In most instances full recovery can be expected; however, some children do have long-term disability as an outcome and require careful follow-up to prevent the development of contractures and facilitate active motor control.

The first muscles to return are the elbow, wrist, and finger extensors followed by the deltoid and biceps and later the external rotators. The timely recovery of these muscles (beginning at 6 weeks and continuing through 3 months) is prognostic of good functional recovery.<sup>42</sup>

The long-term prognosis for recovery of motor control is poor beyond 18 months (Table 23-8), and probably 15% of infants experience significant disability, with reports showing a wide range of long-term impairments.<sup>152</sup> Recovery of shoulder external rotation is highly indicative of a good long-term outcome and is a key movement for performing a variety of functional tasks. Almost half of those with the Erb-Duchenne type of injuries do not recover shoulder external rotation, and contractures of the shoulder and elbow joint with atrophy of the affected muscles can occur.

#### SPECIAL IMPLICATIONS FOR THE THERAPIST

23-9

##### *Erb's Palsy*

###### PREFERRED PRACTICE PATTERN

*5F: Impaired Peripheral Nerve Integrity and Muscle Performance Associated with Peripheral Nerve Injury*

An integrated team approach to congenital brachial plexus injuries is imperative. Each child must be carefully evaluated, therapy interventions maximized, and the surgical approach (when required) individualized to obtain the best outcome.

An aggressive and integrated physical and occupational therapy program is essential in the treatment of these injuries. The therapist uses a problem-solving approach and continually adjusts the interventions based on each child's unique needs. The maintenance of full passive mobility during the period of neurologic recovery is essential for normal joint development.

Early surgical correction of shoulder contractures and subluxations reduces permanent disability. Post-operative rehabilitative therapy can preserve and build on gains made possible by medical or surgical interventions.<sup>144-147</sup>

Treatment should focus on activities that encourage active and active assistive movement and that maintain the normal joint kinematics. The shoulder requires particular attention to maintain the normal scapulo-humeral and scapulothoracic relationships in addition to maintaining the normal "roll and glide" of the glenohumeral joint and preventing subluxation.

*Continued.*

**Table 23-7** Brachial Plexus Injury: Clinical Characteristics

Type	Typical Posture	Strength Losses	Sensory Losses	Skeletal Changes	Treatment Strategies
Erb's palsy (C5-C6)	Shoulder IR, adduction, finger flexion (difficulty with hand to mouth, hand to head, and hand to back of neck)	Deltoid, supraspinatus, infraspinatus, teres minor, biceps, brachialis, brachioradialis, supinator	C5-C6 deficits	Flattening of glenoid fossa/humeral head Elongating deformity of coracoid process hooking down and lateral Scapular winging potential Posterior shoulder dislocation	Active/active assistive exercise: shoulder abduction; elbow flexion; forearm supination and shoulder ER
Klumpke's palsy (C8-T1)	Pronation, elbow flexion contractures; no grasp reflex	Wrist flexors, long finger flexors; hand intrinsics	Diminished sensation	Hypertrophy of olecranon and coronoid process; elbow flexion contracture; posterior dislocation of radial head (25%)	Active/active assistive exercise: forearm supination, elbow extension, finger flexion Positional splint to assist with elbow extension; combined with UE weight bearing
Total plexus injury (whole arm)	Combinations of above	Combination of Erb's and Klumpke's type	Moderate losses	Posterior glenohumeral dislocation	Combination of above

IR, Internal rotation; ER, external rotation; UE, upper extremity.

**Table 23-8** Key Indicators of Recovery of Motor Control\*

Muscle	Time Since Birth (Mo)
Elbow, wrist, and finger extensors	1½
Deltoid, biceps	2
Shoulder external rotators	3

\*Good functional recovery is expected if the child achieves a strength grade of 3 or better within the listed time frames.

Some strategies used to maintain functional upper extremity ROM, prevent subluxation, and improve active movement include neuromuscular electrical stimulation, biofeedback, myofascial release techniques (sometimes referred to as soft tissue mobilization), joint mobilization, and positioning using splints. Carefully applied neurodynamic techniques to physically challenge the nervous system in Erb's palsy may contribute to the physical health of the nervous system leading to optimum physiology. Scapulothoracic stabilization for winging of the scapula using taping may be helpful, but no reported outcomes have been published for these last two interventions.

Passive ROM exercises should be performed three times a day in the direction of limited movement to help prevent the development of contractures, and a well-thought-out home program is an integral part of the therapy program. When splints are used, careful follow-up and family education by the therapist is necessary, especially if sensory impairment is present.

## OSTEOGENESIS IMPERFECTA

### Overview and Incidence

Osteogenesis imperfecta (OI), sometimes referred to as brittle bones, is a rare congenital disorder of collagen synthesis affecting bones and connective tissue. Four primary types of OI exist, with varying degrees of severity and clinical presentation (Table 23-9). Clinical features vary widely between types, within types, and even within the same family. Experts estimate that between 30,000 and 50,000 people have OI in the United States (prevalence), or about 1 in 20,000 (incidence).<sup>134</sup>

### Etiologic Factors and Pathogenesis

Most children with OI inherit the disorder from a parent (autosomal dominant inheritance). Genetic counseling requires recognition that the parent can be a carrier of the dominant gene by parental mosaicism (i.e., the parent carries the mutation in a portion of his or her germ cells).

Approximately 25% of children with OI, however, are born into a family with no history of the disorder. In these cases, the genetic defect occurred as a spontaneous mutation. Because the genetic defect is usually dominant (whether inherited from a parent or resulting from a spontaneous mutation), affected people have a 50% chance of passing on the disorder to each of their children.<sup>134</sup>

More than 150 mutations have been identified as causative in OI, all affecting the genes (COL1A1 and COL1A2) that code for type I collagen. Type I collagen (see the section on Collagen in Chapter 6 and Fig. 6-7) is found