Diagnostic Evaluation and Therapeutic Management

The diagnosis is based on the molecular genetic marker for the *SMN* (survival motor neuron) gene, which is located on chromosome 5q13. Prenatal diagnosis may be made by genetic analysis of circulating fetal cells in maternal blood (Béroud, Karliova, Bonnefont, et al, 2003) or circulating fetal cells in amniotic fluid. The risk of subsequent affected offspring in carriers of the mutant gene or in families with known cases of SMA may also be evaluated genetically. Further diagnostic studies include muscle electromyography (EMG), which demonstrates a denervation pattern, and muscle biopsy; however, the genetic analysis has become the gold standard for diagnosis of the condition (Sarnat, 2016a).

There is no cure for the disease, and treatment is symptomatic and preventive, primarily preventing joint contractures and treating orthopedic problems, the most serious of which is scoliosis. Hip subluxation and dislocation may also occur. Many children benefit from powered wheelchairs, lifts, special pressure-adjustable mattresses, and accessible environmental controls. Muscle and joint contractures require careful attention and care to prevent further complications. Nutritional growth failure may occur in infants and toddlers as a result of poor feeding; supplemental gastrostomy feedings may be required to maintain adequate nutritional status and maintain weight gain. The use of lower extremity orthoses may assist with ambulation, but eventually, the child may be confined to a wheelchair as muscle atrophy progresses. Restrictive lung disease is the most serious complication of SMA (Iannaccone, 2007). Upper respiratory tract infections often occur and are treated with antibiotic therapy; they are the cause of death in many children. Rapid eye movement (REM)–related sleep-disordered breathing is common in children with SMA type 1; this progresses to sleep-disordered breathing during REM and non-REM sleep followed by respiratory failure, which often requires nocturnal noninvasive mechanical ventilation (Schroth, 2009). Noninvasive ventilation methods such as bilevel positive airway pressure (BiPAP) have decreased the morbidity and increased the survival rate of children with SMA types 1 and 2. A decreased ability to cough and clear secretions may be managed with airway clearance therapies such as the cough-assist machine and manual cough assistance. Guidelines