verbal, tactile, and auditory stimulation are important aspects of developmental care. Supporting them so that they can see the activities around them and transporting them in appropriate conveyances (e.g., wagon, power wheelchair) for a change of environment provide stimulation and a broader scope of contacts.

Children who are able to sit require proper support and attention to alignment to prevent deformities and other complications. Children who survive beyond infancy need attention to educational needs and opportunities for social interaction with other children. The parents of a child who is chronically ill require much support and encouragement* (see Chapter 17). Parents who have not sought genetic counseling should be encouraged to do so to evaluate further risk potential.

Congenital muscular dystrophies have an onset at birth and clinical manifestations in the first 2 years of life. Although rare disorders, these are divided into three major groups: (1) collagenopathies, (2) merosinopathies, and (3) dystroglycanopathies. In addition to progressive skeletal muscle weakness and hypotonia, some are associated with joint hyperlaxity and eye or brain abnormalities. Genetic studies may help to correlate with specific phenotypes. Evidence-based guidelines for evaluation, diagnosis, and management of congenital muscular dystrophies have been published recently by the American Academy of Neurology (Kang, Morrison, Iannaccone, et al, 2015).

Spinal Muscular Atrophy, Type 3 (Kugelberg-Welander Disease)

SMA type 3 (Kugelberg-Welander disease) is a result of anterior horn cell and motor nerve degeneration. The disease is characterized by a pattern of muscular weakness similar to that of type 1 SMA (see Box 30-7). Several modes of inheritance have been reported for the disease: autosomal recessive, autosomal dominant, and X-linked recessive.

The onset occurs from younger than 1 year old into adulthood, with symptoms resembling type 3 SMA. Proximal muscle weakness (especially of the lower limbs) and muscular atrophy are the predominant features. The disease runs a slowly progressive course. Some children lose the ability to walk 8 to 9 years after the