



# POSNA

## The Core Curriculum

### **Congenital muscular dystrophy**

#### **Objective**

1. Define the clinical features of congenital muscular dystrophy

#### **Discussion**

Congenital muscular dystrophy is defined as the presence of weakness or hypotonia at birth, contractures before 6 months of age, and dystrophy on muscle biopsy. It can affect both males and females, can be present with and without central nervous system involvement, and is not associated with a disorder of the dystrophin protein. There are two types, Type I is without central nervous system involvement, and has a good prognosis for long-term survival. Control of contractures, hip dislocation, and spinal deformity can require attention. Type II is also known as Fukuyama muscular dystrophy and has been most often described in Japanese infants. It is accompanied by severe developmental delay due to CNS involvement in addition to the peripheral dystrophy. Prognosis is poor.

#### **References**

1. Duggan DJ, Gorospe JR, Fanin M, Hoffman EP, Angelini C. Mutations in the sarcoglycan genes in patients with myopathy [see comments]. *New England Journal of Medicine* 1997;336(9):618-24.
2. Jones R, Khan R, Hughes S, Dubowitz V. Congenital muscular dystrophy: the importance of early diagnosis and orthopaedic management in the long-term prognosis. *Journal of Bone & Joint Surgery - British Volume* 1979;61(1):13-7.
3. Thompson GH. Neuromuscular disorders. In: Morrissy RT, Weinstein SL, editors. *Pediatric Orthopaedics*. Philadelphia: Lippincott-Raven Press; 1996. p. 537-77.