



POSNA

The Core Curriculum

Myotonic dystrophy

Objectives

1. Describe the clinical features of myotonic dystrophy
2. Discuss the features of myotonic dystrophy which could necessitate orthopaedic intervention

Discussion

Myotonic dystrophy is an autosomal dominant disorder in which the defect has been localized to an unstable cytosine-thymine-guanine (CTG) trinucleotide repeat in chromosome 19, with manifestations of multiple organ systems in addition to muscle. The child usually has a characteristic elongated expressionless face, delayed developmental milestones, weakness, and hyporeflexia. If the diagnosis is apparent in the newborn period, significant retardation can be expected, with a 25% mortality by age 18. Ambulation is usually delayed. Orthopaedic problems can include clubfoot, hip dislocation, contractures, and spinal deformity. Phenotypes resembling myotonic dystrophy have been reported, and a conference in 1999 designated the autosomal dominant form as DM1. Onset of milder forms can occur later in childhood or even into adult life. Not surprisingly, weaker patients have an increased rate of pulmonary problems associated with anesthesia.

References

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