## **Condition:** Myotonic dystrophy

### Inheritance:
Autosomal dominant.

### Genetic etiology:
Mutation in *DMPK* gene, which encodes dystrophia myotonia protein kinase protein. The mutation consists of a CTG triplet repeat expansion in the 3' untranslated region of the gene. Normal alleles include 5-35 repeats; full penetrance alleles are >50 repeats; intermediate numbers are associated with permutation alleles that are prone to mutation to full penetrance alleles. The pathogenesis is believed to be related to increased binding of an RNA-binding protein to the CUG expansion on the mRNA, interfering with the processing of other mRNAs.

### Frequency:
1/10,000 – 1/20,000.

### Clinical features:
Myotonia is defined as sustained muscle contraction. Aside from this symptom, affected individuals exhibit weakness, particularly of facial and distal muscles. Other features include cataracts, diabetes mellitus, and cardiac conduction defects. Children with extremely large repeat number (>2,000) have congenital onset of severe symptoms.

### Management:
Supportive care; monitoring for cardiac problems and diabetes.

### Genetic counseling:
Affected individuals face a 50% risk of transmission; myotonic dystrophy displays anticipation. Most cases of congenital myotonic dystrophy are inherited from the mother.

*Human Genetics and Genomics,* Fourth Edition. Bruce R. Korf and Mira B. Irons.