Concerned parents sought evaluation of their 3-year-old daughter, whose developmental milestones were somewhat delayed. The child sat without support at 10 months of age and walked at 17 months of age.

The mother, a 27-year-old primigravida, had had an uncomplicated pregnancy. Because of fetal distress, the patient was delivered at term by cesarean section. Apgar scores were 7 and 9 at 1 and 5 minutes, respectively. Birth weight was 3.4 kg (7.5 lb); length, 51 cm (20 in).

Mild hip contractures were noted at birth. The infant's respiratory distress in the neonatal period was secondary to transient tachypnea of the newborn and a right phrenic nerve palsy. Supplemental oxygen was given for 24 hours. The neonatal course was otherwise uneventful.

At 17 months of age, the child's face was elongated, and the palate was high-arched. Her mouth was tent-shaped, with an inverted V-shaped upper lip. When the child was crying, her face moved very little. The patient was hypotonic with weakness in the distal muscles; a bilaterally weak hand grasp was particularly noteworthy. No myotonia was demonstrated. Findings of the ophthalmologic examination were normal.

DNA analysis showed 900 repeats of the cytosine-thymine-guanine trinucleotide (normal, 5 to 35 repeats). Both the mother and the maternal uncle had the trinucleotide repeat expansion. The mother was clinically normal; the uncle had a 2-year history of grip myotonia.

Drs Alexander K. C. Leung and C. Pion Kao of Calgary, Alberta, point out that the diagnosis of myotonic dystrophy is based on the typical clinical features, a positive family history, and the characteristic cytosine-thymine-guanine triplet expansion. The disease affects 1 in 30,000 neonates among the general population; it is inherited as an autosomal dominant trait. The gene for myotonic dystrophy is located on the long arm of chromosome 19 at the 19q13 locus and codes for myotonin-protein kinase; the genetic defect consists of an expansion of the gene with numerous repeats of the cytosine-thymine-guanine codon.

The characteristic tent-shaped, or triangular fish-type, mouth is a clue to the diagnosis. In contrast to other primary myopathies, weakness and wasting associated with this disorder are more pronounced in distal rather than proximal muscles. Smooth muscles may also be affected and can lead to slurring of speech and to constipation. Swallowing difficulties occur in some patients. Mental retardation and developmental delay are common. Myotonia usually is not evident until about age 5 years; typically, cataracts do not develop until the second decade of life. Patients with myotonic dystrophy are at risk for hypothyroidism; Addison disease; and diabetes mellitus, most frequently type 1. One third of patients have increased IgG catabolism with resultant low serum IgG concentrations.

Serum levels of muscle enzymes may be normal or mildly elevated. An ECG may reveal conduction defects and, less frequently, arrhythmias. Both myotonic and myopathic features may be evident on an electromyogram. Generally, muscle biopsy is not indicated because it is not definitively diagnostic; however, the presence of characteristic features, such as large numbers of centronuclear fibers and selective atrophy of type I fibers, strongly suggests myotonic dystrophy. To confirm the
diagnosis, a DNA probe can be very helpful.
Treatment is mainly supportive; there is no specific therapy.

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