A 4-MONTH-OLD infant with musculoskeletal anomalies had a history of poor feeding and hypotonia. He was born by breech delivery to a multigravida mother whose pregnancy was complicated by oligohydramnios and decreased fetal movements. At birth, the infant's hips were dislocated and a right femur fracture was detected. On physical examination, the infant had a round face with a midline, glabellar nevus flammeus and an upturned nose. A nasogastric tube was in place for feeding (Figure 1). The shoulders were sloped and internally rotated, and the left elbow was fixed in extension with flexion contractures of the wrist and hand in a “policeman tip” position (Figure 2). The lower extremities had decreased muscle mass with flexion contractures of both knees and talipes equinovarus deformities of the feet. Findings from laboratory studies were normal, including a serum creatine kinase, computed tomography of the brain, and electromyography. The karyotype was 46 XY.

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Denouement and Discussion

Amyoplasia Congenita

Figure 1. The face is round with a midline glabellar nevus flammeus and upturned nose.

Figure 2. The shoulders are rounded, sloped, and internally rotated. The left elbow is extended with flexion contractures of the hand and wrist in a "policeman tip" position.

Arthrogryposis refers to fixed flexion contractures of a joint. The syndrome, arthrogryposis multiplex congenita, consists of a variety of disorders that result in the presence of fixed contractures of joints of the extremities at birth, with an estimated occurrence rate of 1 in 3000 births.1 Amyoplasia congenita is the most common cause of this syndrome, accounting for more than one third of cases of arthrogryposis.1

CLINICAL FEATURES

The limb findings in amyoplasia congenita are usually symmetric, involving all 4 extremities (84%), although some patients have isolated upper (11%) or lower (5%) extremity involvement.1 The arms are extended and the wrists and hands flexed, creating the so-called "policeman tip" position. The shoulders are sloped and rounded and internally rotated. The knees may be flexed or extended and the hips flexed and often dislocated. The feet are almost always in an equinovarus position. The muscle mass of the limbs is diminished, giving the extremities a slender appearance.

Facial features are not pathognomonic, although a round face with micromelia and a small upturned nose are often present. A midline nevus flammeus of the forehead is present in three fourths of patients.1 Other abnormalities described in children with this disorder include cryptorchidism in males and labial abnormalities in girls, congenital hernias, abdominal wall defects, and scoliosis.

PREGNANCY AND BIRTH HISTORY

A history of decreased fetal movement may be present. The amount of amniotic fluid may be increased, decreased, or normal. Intrauterine presentation is breech in almost one third of cases.1 Fractures of long bones are relatively common at birth, related to the immobility of joints and difficulty of delivery.

PATHOPHYSIOLOGY

Based on the finding of decreased numbers of anterior horn cells in the spinal cord in the most common form of arthrogryposis, it has been postulated that hypotension in the developing fetal spinal cord at a time when the anterior horn cells are susceptible to insults may result in this form of the disorder.2,3 Amyoplasia congenita is sporadic in occurrence. No recurrences were reported in families of 135 affected patients.4

LABORATORY FINDINGS

Histopathologic findings of muscle show replacement of muscle with adipose tissue and fibrous tissue. The remaining muscle shows variability in fiber diameter. Myopathic and neuropathic processes have been diagnosed from different muscles of the same patient.4 Results of electromyographic and nerve conduction velocity studies are also inconsistent. Electromyography may show a decreased or absent response to stimulation in some muscle tissues and a normal response in others.4 No structural or microscopic changes in the brain have been reported, but there is a reduction in the number and size of anterior horn cells at various spinal levels.4

MANAGEMENT AND OUTCOME

The prognosis of patients with amyoplasia congenita is relatively optimistic, although patients with markedly small limbs have a less favorable outcome. By age 5 years, 85% of children are ambulatory and completely independent in terms of activities of daily living.1 Primary goals in management are ambulation and maximum upper limb mobilization. Aggressive physical and occupational therapy beginning shortly after birth are key to good outcome. Educationally, almost all children function in regular classroom settings at the appropriate grade level, with only 4% in special programs.1 Amyoplasia congenita should be differentiated from congenital myopathies, myotonia congenita, and congenital muscular dystrophy, which have different clinical and pathologic pictures despite the presence of hypotonia and weakness.

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