Amyoplasia congenita: *A rare form of arthrogryposis*

Sir,

Arthrogryposis multiplex congenita (AMC) is one of the common neuromuscular disorders seen in childhood. A large number of disorders involving brain, spinal cord, peripheral nerves, muscles, myoneurojunction and connective tissue diseases can present with arthrogryposis.[1] An associated common factor is diminished fetal movements in the antenatal period. The prognosis is dependent on the underlying cause and the disease could be fatal early in life. However, some affected children may have a normal life span.[1] Amyoplasia congenita (AC), a rare cause of AMC is being reported.

An 8-month-old boy was evaluated for deformities and contractures of extremities. He was the 5th child of non-consanguineous parents (Iraqi mother). This child was one of a twin pregnancy, the other one having aborted at 6 weeks of gestational age. The pregnancy was otherwise uneventful with normal vaginal delivery. The limb anomalies were noted at birth. The mental and social milestones including head holding were attained normally. The child could not roll supine to prone or vice versa. He could sit at 7 months of age unsupported, when made to sit. On vertical suspension there was no weight bearing on the feet. The head circumference, height and weight were at the 25th centile. There was no facial dysmorphism. The cranial nerves were normal.

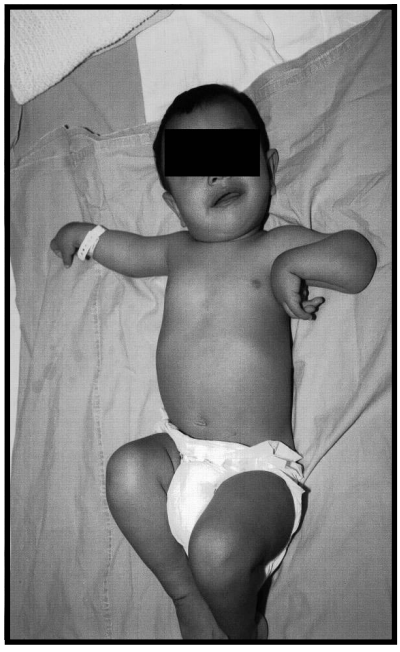

Figure 1 - Contractures of various joints of upper and lower limbs.

The upper extremities were adducted at the shoulders and internally rotated. There were contractures at elbows, wrists and interphalangeal joints. The contracture of the left elbow was more than the right. The muscle mass was grossly reduced with loose skin folds. The lower limbs were extended with mild contractures at knees, small joints of feet and equinovarus deformity (**Figure 1**). All the deep tendon jerks were elicitable. The sensations were intact. Magnetic resonance image (MRI) of spine was normal. Both sensory and motor nerve conductions were normal in upper and lower limbs. Muscle biopsy was not carried out, as the diagnosis was evident.

Amyoplasia congenita has typical features of arms extended with flexion of hands and wrists, shoulders internally rotated with decreased muscle mass, bilateral equinovarus and variable contractures of other joints.[2,3] There is usually symmetrical contractures of all 4 extremities (92%), although some cases involve only lower limbs (7%) or upper limbs (1%).[2] The upper limb involvement is typical of bilateral Erb's paralysis, however contractures at elbow, wrists, metacarpo phalangeal joints and interphalangeal joints is seen to a variable extent. The lower limbs are adducted at hips with usually bilateral dislocation at hips, associated with contractures at knees, ankles and small joints of feet. Other occasional abnormalities associated with AC include facial dysmorphism, stiff spine, abdominal structural abnormalities (bowel atresia, gastrochisis and abdominal wall defects).[3] Amyoplasia congenita is a non-progressive condition and has onset in utero. These children are of normal intelligence and multiple orthopedic procedures are needed for good functional results. The condition is sporadic, with no recurrence risk.[3] Recently, AC has been reported with a balanced de novo insertion of a segment of the long arm of chromosome 5 into chromosome 2.[4] Muscle biopsy from areas affected by amyoplasia show absence of muscle with fibrotic and fatty replacement, whereas other muscle may be entirely normal.[3] Spinal anterior horn cell ischemia, due to intrauterine vascular accident has been suggested as the underlying pathology in AC.[2] This hypothesis has been challenged by some authors who believe that the most likely cause of AC is arrest of myogenesis in different parts of the body at 1-7 weeks of fetal life.[5] This theory is supported by normal MRI of spinal cord and nerve conduction studies as seen in our patient.

Roshan Koul, Hashim Javed, Alexander Chacko
Division of Pediatric Neurology
Sultan Qaboos University Hospital
Baby T. John
Department of Child Health
Ministry of Health
Sultanate of Oman

References

1. Gordon N. Arthrogryposis Multiplex Congenita. ***Brain Dev*** 1998; 20: 507-511.
2. Jones KL, editor. Smith's Recognizable Patterns Of Human Malformation. 5th ed. London (UK): WB Saunders Company; 1997. p. 170-171.
3. Stevenson RE, Meyer LC. Arthrogryposis (Multiplex Congenital Contractures). In: Stevenson RE, Hall JG, Goodman RM, editors. Oxford Monograph On Medical Genetics No. 27. Human Malformations and related Anomalies Vol. II. Oxford (UK): Oxford University Press; 1993. p. 699-804.
4. Eisenhut M, Weindling M, Fryer A. Amyoplasia associated with a balanced de novo insertion of a segment of the long arm of chromosome 5 in chromosome 2. ***Am J Med Genet*** 2002; 107: 337-339.
5. Carpenter S, Karpati G. General aspects of skeletal muscle biopsy. In: Pathology of Skeletal Muscle. 2nd ed. New York (NY): Oxford University Press; 2001. p. 28-62.