CLINICAL QUIZ (p190) ANSWER

Figure 1 showed multiple joint contractures, or arthrogryposis. The typical pattern of deformity with reduced muscle bulk points to amyoplasia congenita as the most likely diagnosis. Water soluble contrast study (Figure 2) revealed loops of dilated bowels with the most distended loop extending from the right side to the central abdomen and rectal gas was absent, compatible with lower gastrointestinal tract obstruction. The boy underwent laparotomy where single segment atresia at the distal ascending colon with complete separation of bowel ends was identified (Type III), partial colectomy with primary anastomosis was performed. Unfortunately, the patient deteriorated post-operatively with intestinal obstruction and perforation, further complicated by severe pulmonary hemorrhage. He succumbed on Day 33 of life; postmortem examination was declined.

Arthrogryposis multiplex congenita (AMC) comprises of a variety of underlying disorders that result in the presence of fixed contractures of joints of the extremities at birth, and has a prevalence of 1 in 3000 live births. Intrinsic fetal causes of arthrogryposis include neurological abnormalities, primary myopathy and abnormalities in connective tissue or cartilage, while extrinsic factors consist of intrauterine compression, maternal infection or exposure to teratogens as well as in-utero vascular compromise (Table 1). The pattern of contractures in our patient is consistent with that of amyoplasia congenita, the most common cause of AMC. Amyoplasia is a sporadic disorder with characteristic positioning of the limbs: internal rotation at shoulders, extension at the elbows and flexion and ulnar deviation of wrists ('policeman tip' appearance); hip dislocation, knee extension and severe equinovarus are usually present.[1] The involvement of upper and lower limbs is usually symmetrical. During pregnancy, mothers may report decreased fetal movement. Since motion is essential for the normal development of joints and their contiguous structures, lack of fetal movement causes extra connective tissue to develop around the joint. The amount of amniotic fluid may be increased, decreased or normal. Breech or transverse presentation may be present. Fractures of long bones at birth are relatively common due to immobility of joints. A midline nevus flammeus of the forehead is typically present in three fourths of patients. Other common features include facial hemangioma, round face and micrognathia. Intelligence is normal in most patients.

Several case series suggest an association between amyoplasia and bowel atresia or gastroschisis.[2] Reid et al hypothesised that the limb anomalies seen in amyoplasia may be due to maternal and/or fetal vascular compromise leading to hypotension in the developing spinal cord,[3] explaining the unusually symmetrical limb involvement; whereas a similar vascular event may also be responsible for associated structural anomaly such as intestinal atresia.

In establishing the differential diagnosis, a throughout neurological examination is important. A normal neurological examination suggests amyoplasia, distal arthrogryposis, connective tissue disorder, or fetal crowding as the cause of arthrogryposis. On the other hand, an abnormal examination indicates a central nervous system etiology or a progressive neurological etiology. In amyoplasia, histopathology of muscles of may show features of myopathy, but the results are inconsistent, as are electromyography and nerve conduction velocity study. Genetic evaluation is expected to be normal as in our patient, compatible with the sporadic occurrence of the disease.

The goals of treatments are to mobilise joints, increase muscle strength and improve functional ability for activities of daily living. Aggressive physical and occupational therapy shortly after birth are essential. Splints are applied for improved position and function. Persistent limb deformities are often treated surgically. The prognosis is relatively optimistic, with the ability to walk and perform activities of daily living reported to be as high as 85% with proper interventions.[4]

Table 1 Common causes of arthrogryposis

Disease	Genetic influence	Additional factors/findings
Primary Myopathy, Structural and/or Functional		
Amyoplasia	Sporadic	Usually quadrimelic involvement
Steinert's myotonic dystrophy	AD	Myotonia, typical facies
Congenital muscular dystrophy	AR	Heterogeneous group of diseases, some with CNS involvement
Distal arthrogryposis syndromes	AD	Hand, foot involvement
Freeman-Sheldon syndrome	AD	Whistling appearance to face, ulnar deviation of hands, clubfoot, and congenital vertical talus
Multiple pterygium syndromes	AR	Pterygium of upper and lower extremities, neck
Neurological abnormalities		
Spinal Muscular Atrophy	AR	Anterior horn cell degeneration
Abnormalities in Connective Tissue (Hyperextensibility) or Cartilage		
Diastrophic dysplasia	AR	Clubfeet, hitchhiker's thumb, short stature, scoliosis, hypertrophic pinnae
Larsen syndrome	AD	Joint dislocations, spatulate thumbs, flattened nasal bridge
Congenital contractural achonodactyly (Beals syndrome)	AD	Slender limbs with knee, elbow, and hand contractures
Intrauterine Compression		
Multiple gestation		
Uterine anomaly		
Oligohydramnios		
Maternal Exposure Including Infection or Teratogens		
Maternal myasthenia gravis		
Vascular Compromise to the Developing Fetus		

AD=autosomal dominant, AR=autosomal recessive, CNS=central nervous system

Modified from Bernstein RM. Arthrogryposis and amyoplasia. J Am Acad Orthop Surg 2002;10:417-24.[5] Rink BD. Arthrogryposis: a review and approach to prenatal diagnosis. Obstet Gynecol Surv 2011;66:369-77.[6]

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