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Arthrogryposis multiplex congenita

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Introduction:

Arthrogryposis is development of non-progressive contractures affecting one or more areas of the body. It derives its name from Greek, literally meaning "curving of joints" (arthron, "joint"; grȳpōsis, late Latin form of late Greek grūpōsis, "hooking")[1]. A contracture is a condition in which a joint becomes permanently fixed in a bent (flexed) or straightened (extended) position, completely or partially restricting the movement of the affected joint. When arthrogryposis affects two or more different areas of the body, it may be referred to as arthrogryposis multiplex congenita (AMC). Children born with one or more joint contractures are unable to perform active extension and flexion in the affected joint or joints. AMC has been divided into three groups, a) Amyoplasia b) distal arthrogryposis, and c) syndrome. Amyoplasia is characterized by severe joint contractures and muscle weakness. Distal arthrogryposis mainly involves muscle disease belong to the syndrome group[2]. The symptoms of AMC are present at birth (congenital). However, specific symptoms and physical findings can differ greatly in range and severity from one person to another. In most cases, affected infants have contractures of various joints. The joints of the legs and arms are usually affected; the legs are affected more often than the arms. The joints of the shoulders, elbows, knees, wrists, ankles, fingers, toes, and/or hips are also commonly affected. In addition, the jaws and back may also be affected in individuals with AMC. In most cases, AMC occurs randomly, for no apparent reason (sporadic). More than 400 different conditions can cause isolated or multiple contractures and the causes, genetics, specific symptoms, and severity of these disorders vary dramatically. Over 350 genes have been identified as responsible for different types of arthrogryposis[3].

Keywords: Arthrogryposis in a neonate

Case Report:

Full term male baby born to Saudi mother, after lower segment Caesarean section (previous c/s). Apgar score of 7 and 9 at 1 and 5 minutes; needed routine minimal resuscitation. Weight 2.7 kilogram, head circumference 34 centimeter, length 46 centimeter, vital signs were normal. Antenatal history mother is 31 years old has two children both are normal. She had polyhydramnios, gestational age 38 weeks. Baby was shifted to neonatal intensive care unit. On examination micrognathia, high arched palate, contractures of hands and feet, generalized hypotonia, inability to suck. Right upper arm fracture (figure 1, 2).Clinically no abnormality detected in respiratory, cardiovascular, gastro intestinal, genitourinary, nervous system. Sluggish tendon reflexes, decreased muscle tone. Neonate was passing urine stool normally, on full feed through orogastric tube. Investigation of hematological, hepatic, renal profile was normal. Ultrasonography brain, abdomen was normal. Skeletal survey revealed fracture right humerus, contracture hands and feet. Echocardiography normal. Baby developed jaundice (blood group baby B+, mother AB+) subsided with photo therapy. Suspected sepsis treated blood culture no growth. Chromosomal analysis normal. X-ray right arm displaced, maligned with healing fracture of mid shaft of humerus. Evidence of

surrounding callus formation. Baby stayed in the hospital for forty days and was discharged in stable condition on orogastric feeding, baby is on full feed. Mother was taught how to feed the baby. Baby will be followed in pediatric, orthopedic and physiotherapy outpatient clinics.

Figure 1. Arthrogryposis hands and feet.

Figure 2. X-Ray shows Arthrogryposis hand and feet and fracture right humerus.

Discussion:

Arthrogryposis multiplex congenita (AMC) refers to a heterogeneous group of muscular, neurologic, and connective tissue anomalies that present with 2 or more joint contractures at birth as well as muscle weakness. It is associated with abnormal contraction of muscle fibers, causing reduced mobility with a decreased active and passive arc of motion[4]. Arthrogryposis is not a specific diagnosis but a descriptive term with various etiologies and complex clinical features, including multiple congenital contractures of various limb joints. It is associated with 200-300 different disorders encompassing malformations, malfunction, and neurologic deficiencies.

Approximately 1% of all births show some form of contractures of the joints ranging from unilateral clubfoot to pervasive, crippling contractures due to Amyoplasia. The condition has been reported in individuals of Asian, African and European descent.

Overall incidents of arthrogryposis have been reported to be 1 in 5,000-10,000 live births with equal gender ratios. Children with arthrogryposis have many problems such as micrognathia, nutrition issues, and sucking problem as in our patient. In the absence of central nervous system lesions, many children have normal intelligence[5].
AMC is divided into subgroups with different signs, symptoms, and causes as a practical way to make a differential diagnosis. Disorders involving primarily limbs such as Amyoplasia and distal arthrogryposis are the most common as in our case. Disorders with limb involvement and abnormal neurologic function are caused by atypical central nervous system, peripheral nervous system, and damaged or absent anterior horn cells. Amyoplasia, also known as classic arthrogryposis, is a sporadic symmetric disorder that causes fibrotic replacement of the muscles. Symptoms include internally rotated and adducted shoulders, extended elbows, pronated forearms, flexed fingers and wrists, dislocated hips, feet with severe equinovarus contractures and extended knees[6]. Involved muscles are hypo plastic and fibrotic. Intelligence is usually normal. Distal arthrogryposis, an autosomal dominant disorder that primarily affects the distal joints of the limbs. Characteristics of the upper limbs are medially overlapping fingers, clenched fists, ulnar deviation of fingers, camptodactyly, and hypoplasia. Lower limbs show talipes equinovarus, calcaneo valgus, vertical talus, or metatarsus varus[7].
The primary underlying mechanism that causes congenital contractures is believed to be decreased fetal movement during development. The joints begin to develop in a fetus around five or six weeks into pregnancy. Motion is essential for the proper development of fetal joints. A lack of fetal movement allows for excess connective tissue to form around the joints, which can result in the joint becoming fixed and/or limiting the movement of a joint. In theory, any factor that diminishes or restricts fetal movement can cause congenital contractures. Such factors would include fetal crowding (in which there is not enough room for the fetus to move around) such as when there are multiple births or uterine structural abnormalities. Restricted fetal movement can also occur secondary to maternal disorders including viral infections, drug use, trauma or other maternal illness. Low levels of amniotic fluid around the fetus (oligohydramnios) have also been linked to decreased fetal movement[8]. Mother of our patient had polyhydramnios. When a child is born with arthrogryposis a child can have stiff elbows, dislocated hips, dislocated, hyperextended or contracted knees, and clubfeet. The therapeutic and orthopedic goal for the child with arthrogryposis limb deformities is to achieve maximal joint motion and to optimize joint position for function. In the lower extremities, the foot needs to be plantigrade. The knees need to have optimal motion for sitting and standing. Hips need to be stabilized especially if the child has walking potential. In the upper extremities, the goals should include positioning of arm for feeding and the other for toileting in cases where there is extreme stiffness[9]. Clubfoot deformities are the most commonly seen deformities with arthrogryposis as in our case. As would be expected walking is more difficult for children with arthrogryposis due to the muscle weakness and limited joint motion. Children with arthrogryposis who walk have lower activity levels and take fewer steps than their peers.
A diagnosis of AMC is made based upon identification of characteristic symptoms (e.g., multiple congenital contractures), a detailed patient history, and a thorough clinical evaluation. Certain tests may be necessary to determine the underlying cause of AMC including nerve conduction, electromyography and muscle biopsy, which can help diagnose neuropathic or myopathic disorders, and we did not do these tests in our case.
The treatment of AMC is directed toward the specific findings that are apparent in each individual. Standard physical therapy, which can improve joint motion and avoid muscle atrophy in the newborn period is beneficial. Gentle joint manipulation and stretching

exercises may also be beneficial. Removable splints for the knees and feet that permit regular muscle movement and exercise are also recommended. In some cases, surgery may be necessary to achieve better positioning and increase the range of motion in certain joints, especially the ankles, knees, hips, elbows, or wrists[10]. In rare cases, tendon transfers have been performed to improve muscle function. Tendons are the tissue by which muscle is attached to bone. Surgical correction of arthrogryposis upper extremity contractures should be started after 1-3 months and completed by age 12 months so that the child can optimize his or her motor development[11]. Genetic counseling may be of benefit for affected individuals and their families.

Conclusion.

Arthrogryposis multiplex congenita comprises non progressive conditions characterized by multiple joint contractures found throughout the body at birth. The term is currently used in connection with a very heterogeneous group of disorders that all include the common feature of multiple congenital joint contractures. The major cause of arthrogryposis is fetal akinesia (i.e., decreased fetal movements) due to fetal abnormalities (e.g. neurogenic, muscle, or connective tissue abnormalities; mechanical limitations to movement) or maternal disorders (e.g. infection, drugs, trauma, other maternal illnesses[12]. Generalized fetal akinesia can also lead to polyhydramnios, pulmonary hypoplasia, micrognathia, ocular hypertelorism, and short umbilical cord. During early embryogenesis, joint development is almost always normal. 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. This results in fixation of the joint, limiting movement and further aggravating the joint contracture. Contractures secondary to fetal akinesia are more severe in patients in whom the diagnosis is made early in pregnancy and in those who experience akinesia for longer periods of time during gestation[13].

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