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ARTHROGRYPOSIS MULTIPLEX CONGENITA

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ABSTRACT*:*

Arthrogryposis is a congenital disorder characterized by multiple joint contractures which are present at birth. We present the case of newborn with arthrogryposis involving bilateral hip, knee and ankle joints with Osteum secondum Atrial Septal Defect. There is a possibility of genetic counselling for some of the arthrogryphosis conditions which render the very precise diagnosis necessity**.**

KEY WORDS: Arthrogryposis, Contractures

INTRODUCTION

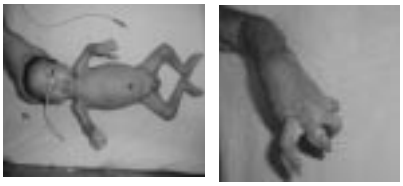
Arthrogryposis Multiplex Congenita also called as amyloplasia is characterized by curved or hooked joints. It is a non-progressive, rare congenital disorder that is characterized by multiple joint contractures and can include muscle weakness and fibrosis, which limits movement[1]. It is a heterogeneous group of disorders. There are about 150 entities[2]. We present a 5 day old baby who presented to us with features of arthrogryposis.

CASE REPORT:

A 5 day old male child born out of a second degree consanguineous marriage to a G2P1L1 mother, delivered by normal vaginal delivery with the birth weight of 1.75 kgs was referred to our hospital in view of congenital anomalies. Mother also gives a history of decreased fetal movements comparing to the previous pregnancy. Antenatal scans were normal. No family history of congenital anomaly.

The clinical examination revealed Term small for gestation age baby with microcephaly, deep set eyes, long eye lashes, low set ears, long philtrum, retrognathia, widely spaced nipple, flexion contracture of all the four limbs. Baby has specific posture-Hip flexed, abducted and externally rotated flexion at the knees and bilateral equinox varus deformity. Extension and pronation at elbow and fingers overlapping each other

Figure :



There was also a left sided inguinal hernia, bilateral crptochrodism, and Osteum secondum Atrial Septal Defect. Based on the above clinical features diagnosis of Arthrogryposis Multiplex Congenita was considered .Karyotyping was 46XY. Serum Creatinine Phospho Kinase, transaminase levels done to rule out muscular dystrophy were normal. Difficulty in initiation of feeds and also maintenance of feeds was there. Child was on Ryle's tube feeding for 14days. Oral stimulation and physiotherapy was given and the gradually changed over to pallada feeds and then discharged on direct breast feeds

DISCUSSION:

Arthrogryposis Multiplex Congenita (AMC) is a rare congenital diseases

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characterised by contractures of all 4 limbs. AMC is seen in 1 out of every 3,000 live births with no sex predominance. Amyoplasia, the most common form (43%) is characterized by fatty and fibrous tissue replacement of the limb muscles .Risk in subsequent pregnancy is about 5%. AMC is typically symmetrical and involves all four extremities with some variation seen .Some of the more common signs and symptoms are associated with the shoulder (internal rotation), elbow (extension and pronation), wrist (volar and ulnar), hand (fingers in fixed flexion and thumb-in-palm), hip (flexed, abducted and externally rotated, often dislocated), knee (flexion) and foot (clubfoot).Complications may include scoliosis, pulmonary hypoplasia, respiratory problems, growth retardation, midfacial hemangioma, facial and jaw variations, and abdominal hernias. Cognition and language are usually normal .Different etiologies, clinical courses, prognosis genetics, and pathologic processes has been put forward[2].

The principal cause of AMC is believed to be decreased fetal movements (akinesia) caused by maternal or fetal abnormalities. Fetal abnormalities (neurogenic, muscle, or connective tissue abnormalities, mechanical limitations to movement like Oligohydramnios .Maternal disorders (infection, drugs, trauma, other maternal illnesses).[1] 30% of the cases genetic in nature and autosommal recessive in inheritance. Life span is usually related to the disease severity and associated malformations. Nearly 20% of patients die in the 1st yr of life. Scoliosis may compromise respiratory function. By age 5 yrs 85% ambulatory with normal mental development[3].

Physical Examination is best for establishing a diagnosis. The limbs are featureless and tubular. Normal skin creases are lacking. Deformities are usually symmetric, and severity increases distally, with hands and feet typically being the most deformed. Associated with joint dislocation, especially the hips and, occasionally, the knees. Atrophy may be present, and muscles or muscle groups may be absent[2].

Sensation is usually intact, although deep tendon reflexes may be diminished or absent. In 2/3 of patients all four limbs are affected equally and in 1/3 of patients lower-limb deformities predominate, and only on rare occasions do the upper extremities predominate. Deformities tend to be more severe and more rigid distally.[4]

Management is mainly conservative and consist of gentle stretching ,light weight splinting, casting, soft tissue relearns, muscle transfer and osteotomy Outcomes better if joint surgery is done early, before adaptive intraarticular changes occur.Osteotomies are usually performed closer to the completion of growth. Early motion, and avoidance of prolonged casting, may increase joint motion, improving function. Many children require long-term bracing or other assistive devices. Many will have feeding problem. As a whole a holistic team approach should be there[5].

No prenatal diagnostic tools are available to test for the condition. Muscle biopsies, blood tests and general clinical findings rule out other disorders like congenital muscular dystrophy, Spinal muscular dystrophy, and Myasthenia gravis and provide evidence for AMC.

Conclusion:

Arthrogryposis is a rare congenital disorder. There is no cure for arthrogryposis, but early vigorous physical therapy can help stretch out the contracted joints and develop the weak muscles. The life span for

an individual with arthrogryposis is usually normal, but may be altered by heart defects or central nervous system problems. Genetic counseling plays an important role in management of Arthrogryposis Multiplex Congenita.

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