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Case Report

## Three consecutive pregnancies having arthrogryposis multiplex congenita

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### ABSTRACT

Arthrogryposis Multiplex Congenita is a syndrome or disorder that presents with multiple congenital joint contractures at birth. It is X-linked recessive disorder, non-progressive and is of variable severity. Here is a case report with three consecutive pregnancies having fetus affected with Arthrogryposis Multiplex Congenita (AMC) diagnosed antenatally. Arthrogryposis Multiplex Congenita affects 1 in 3000 live births. During embryogenesis, joint development is normal. Motion is essential for the normal development of joints and surrounding structures. Fetal akinesia causes extra connective tissue to develop around the joint. This results in fixation of the joint and a further increase in the contracture. Earlier in pregnancy the contractures form, worse is the prognosis.

**Keywords:** Arthrogryposis multiplex congenita, Contractures, Fetal akinesia

### INTRODUCTION

Arthrogryposis Multiplex Congenita is a syndrome or disorder that presents with multiple congenital joint contractures at birth. It is X-linked recessive disorder, non-progressive and is of variable severity.<sup>1</sup> Arthrogryposis Multiplex Congenita affects 1 in 3000 live births.

A contracture occurs when a joint becomes permanently fixed in a bent or straightened position, which can impact the function and range of motion of the joint. Motion is essential for the normal development of joints and surrounding structures. Fetal akinesia causes extra connective tissue to develop around the joint. This results in fixation of the joint and a further increase in the contracture.

Here is a case report with three consecutive pregnancies having fetus affected with Arthrogryposis Multiplex Congenita (AMC) diagnosed antenatally.

### CASE REPORT

A 26 years old pregnant woman reported to our hospital with amenorrhea of 18 weeks. Her first trimester was uneventful. A routine sonography done at 14 weeks gestation showed a single live fetus with no apparent anomalies and normal amniotic fluid index. After 4 weeks the fundal height was found to be more than period of amenorrhea. Hence a repeat sonography was done which showed a single live foetus in transverse position with fixed flexion deformities at the wrist and ankle joints with acute polyhydramnios.

There was no history suggestive of connective tissue disorders, myasthenia gravis, diabetes mellitus, multiple sclerosis or use of any drugs like alcohol phenytoin and curare in the patient. A diagnosis of AMC in the affected fetus was made and after counseling the patient pregnancy was terminated using vaginal misoprostol. The diagnosis was confirmed after birth of the fetus. Post-abortion period was uneventful. Patient's blood group was

B positive, her hemogram, VDRL, fasting blood sugar, TSH, renal function tests, TORCH profile, urine routine examination was within normal limits. The couple was advised to undergo chromosomal analysis, but they refused. Chromosomal analysis of the foetus and the autopsy was also refused. Patient was advised to take folic acid three months prior to next conception.

### **Pregnancy no. 2**

The same patient reported after 16 months with amenorrhea of 18 weeks and 3 days. At 16 weeks an ultrasound was done which showed a single live fetus having flexion deformities at various joints suggestive of arthrogryposis. She was advised a repeat scan after 2 weeks which showed deformities of the upper and lower limbs, right hand-in fixed, acutely flexed position (cranially angulated). Right foot showed club foot deformity (Figure 1 and 2). Left hand did not exhibit individual finger movements.



**Figure 1: Fetus affected with AMC.**



**Figure 2: Affected right hand and foot.**

Patient was again counseled, and pregnancy was terminated. Chromosomal analysis of both parents was done and found to be normal. However, chromosomal analysis of the fetus and the autopsy was refused. Patient was advised to report early in pregnancy for antenatal care.

### **Pregnancy no. 3**

Patient reported again after 10 months with amenorrhea of 19 weeks with a sonogram showing a live fetus of 18 weeks with left hand-fixed, in acutely flexed position. Right hand did not exhibit index finger movements. Left foot fixed with cranial angulation at ankle joint. The patient was explained about the prognosis, but she wanted to continue with the pregnancy and was lost to follow up.

## **DISCUSSION**

Arthrogryposis Multiplex Congenita was first described in 1905 and affects 1 in 3000 live births. During embryogenesis, joint development is normal. Anything that inhibits normal joint movement before birth can result in joint contractures. Genetic and environmental factors have been implicated in development of contractures resulting from curtailed fetal movements.

Fetal akinesia can be caused by fetal neurogenic disorders, connective tissue disorders. Mother suffering from myotonic dystrophy, myasthenia gravis or multiple sclerosis may have an affected fetus. Maternal hyperthermia, viral infections, chronic oligohydramnios, uterine abnormalities (bicornuate / septate uterus), exposure to teratogens (drugs, alcohol, phenytoin, and curare) and trauma are associated risk factors.<sup>1</sup> Earlier in pregnancy the contractures form, worse is the prognosis.

Males are primarily affected in the X-linked recessive disorders. AMC is divided into three groups: amyoplasia, distal arthrogryposis and syndromic. Various characteristics of amyoplasia are severe joint contractures and muscle weakness. Distal arthrogryposis mainly involves the hands and feet. Types of arthrogryposis with a primary neurological or muscle disease belong to the syndromic group.<sup>2,3</sup>

Ultrasound can detect the syndrome in second or third trimester. The limbs showing decreased movements, presence of persistent breech or transverse lie are a high index of suspicion. Polyhydramnios may be associated due to defect in swallowing and is a poor prognostic sign.<sup>4</sup>

The involved extremities are cylindrical, deformities are symmetrical, and deformity increases distally. Joint rigidity or dislocation may be present. Deep tendon reflexes may be absent. Muscle may be atrophic or absent. Jaw is usually hypoplastic. Craniofacial malformations, respiratory and cardiac problems may also be present. Children born with one or more joint contractures have abnormal fibrosis of the muscle tissue causing muscle shortening, and therefore are unable to perform active extension and flexion in the affected joint or joints.<sup>5</sup>

Management goal include lower limb alignment, establishment of stability for ambulation and upper limb function for self-care. Physical therapy combined with splinting is recommended early to preserve motion.

Surgical correction is usually done for lower limb where hip and knee joint functions are restored. Surgical release of contractures or osteotomy may be necessary to improve joint mobility and function.<sup>6</sup> Other positive prognostic factors for independent walking were active hips and knees, hip flexion contractures of less than 20 degrees and knee flexion contractures less than 15 degrees without severe scoliosis.<sup>7</sup>

There are a number of passive devices for enhancing limb movement, intended to be worn to aid movement and encourage muscular development. For example, the Wilmington Robotic Exoskeleton is a potential assistive device built on a back brace, shadowing the upper arm and forearm. It can be difficult to fit and heavy and awkward to wear.<sup>8-10</sup>

Parsch et al treated 38 affected children successfully over a period of 30 years.<sup>11</sup> Neonates affected intrinsically (genetically) have a poorer prognosis as compared to the ones affected extrinsically. Recurrence risk is higher for the intrinsically derived contractures.

The understanding and early recognition of this syndrome is essential as birth of a child with AMC may be catastrophic event for the child and parents.

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