

## **Case Report**

# **Multiple Congenital Anomaly with Dysmorphism and Arthrogryposis: Case report**

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**Abstract:** Arthrogryposis multiplex congenita (AMC), or simply arthrogryposis, describes congenital joint contractures in two or more areas of the body found throughout the body at birth. The name, derived from the Greek, means “curved or hooked joints”. This disorder should be considered a symptom complex or a descriptive term rather than a disease and an underlying cause should be sought when possible. Arthrogryposis usually present with well-recognized musculoskeletal abnormalities. We report a case of arthrogryposis with various deformities other than musculoskeletal abnormalities.

**Keywords:** Arthrogryposis, Joint contractures, Musculoskeletal abnormalities

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## **INTRODUCTION**

Since first described by Otto in 1841, the syndrome known as arthrogryposis multiplex congenita has been discussed in the orthopedic, neurologic, and pediatric literature. In 1976, Cohen and Hsaacs reviewed their experience with the otolaryngologic signs and symptoms [1]. Some authors say the overall prevalence is one in 3000 and others say it is one in 11000-12000 among European live births [3]. Congenital clubfoot is the most common single contracture and its prevalence is one in 500 live births [2].

## **CASE REPORT**

The full term male newborn was product of a non-consanguineous parent delivered by emergency cesarean section due to oligohydromnias with unfavorable Cervix to a 25yr old primigravida who was booked, immunized with regular antenatal checkups with normal Anti natal scan at 2, 5 & 7<sup>th</sup> month with last scan suggestive of Oligohydromnias and no congenital anomaly. The birth weight of the child was 2400gms, head circumference 35 cm and approximate length was 44cm.

On physical examination, the infant had characteristic facies with depressed nasal bridged, widely open mouth and low set ear with noted to have cleft palate ridging of few sutures, left ear smaller than right, antimongoloid slant, Long philtrum, Short neck, micrognathia. Temperature was 37.2°C, heart rate 124 beats/minute, and respiratory rate 54/ minute. Anterior fontanel was open. On investigation it was found to have Right diaphragmatic eventration. Lower limbs

were arthrogryptic characterized by the hip was fixed in flexion, knees were extended, ankles were dorsiflexed and the foot had equinovarus deformity. Shortening of the lower limb was present. Bilateral congenital hip dislocation with restriction of the lower limb movement was present. No history of similar illness was found in the family. Based on these skeletal abnormalities, he was diagnosed as arthrogryposis. On investigation, complete blood cell count was: Hb: 15gm%, TC: 10,600/cmm (LY 32%, MO 10%, GR 58%) and platelet count: 3.1 lac/cmm. Neurosonography revealed normal brain parenchyma with a normal ventricular system. USG abdomen and pelvis reported normal. Karyotyping- normal male (46XX) and microarray analysis was normal. We conservatively managed the neonate and discharged after 7 day without any complication.



**Fig. 1: Photograph of the baby**

## **DISCUSSION**

Arthrogryposis multiplex congenita is a congenital anomaly characterized by non-progressive, multiple joint contractures present at birth. In the classic form of AMC all four limb are involved, but the condition can also occur in the upper or lower limb. An autosomal dominant variant called distal arthrogryposis involves the hand and feet with severe deformation [4]. Children born with one or more joint contractures have abnormal fibrosis of the muscle tissue causing muscle shortening, and therefore are unable to do passive extension and flexion in the affected joint or joints[5].AMC has been divided into three groups: amyoplasia, distal arthrogryposis and syndromic. Amyoplasia is characterized by severe joint contractures and muscle weakness [6].

Distal arthrogryposis mainly involves the hands and feet. Types of arthrogryposis with a primary neurological or muscle disease belong to the syndromic group[6]. Arthrogryposis is not a specific diagnosis, but rather a clinical finding, and it is a characteristic of more than 300 different disorders[7]. The overall prevalence of arthrogryposis is one in 3000 live births[8]. The inheritance, natural history, treatment guidelines, and outcomes of arthrogryposis vary among disorders, underscoring the importance of making a specific diagnosis in each child[9-14].

The goals of initial treatment are to mobilize the joints, apply splints for improved position and function, and to provide physical and occupational therapy as well as instructions to the child's caregivers so that they may provide home therapy. Ongoing therapy services are part of most children's lives, with 80% of children with amyoplasia receiving therapy services into their teenage years[9].

## **CONCLUSION**

A considerable clinical and genetic heterogeneity is noted in almost all arthrogryphosis types. Therefore, a comprehensive musculoskeletal evaluation and genetic consultation is necessary. All these cases should be managed conservatively.

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