

Arthrogryposis Multiplex Congenita: A Rare Case Report

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1. Case Report

A 27 yr old women (G2 P2 L2) was referred for the first ultrasound scan at 32 wks of gestation. Her past medical history was significant as she took infertility treatment, her first pregnancy was uneventful and a female child delivered by normal vaginal delivery 5 years back. She was married at 19 years of age which was a non consanguinous marriage. Her husband was healthy. No familial h/o any congenital disorders. There was h/o drug use for infertility treatment in both the pregnancies (not known). Ultrasound examination of the present pregnancy at 20wks gestation demonstrated a single, live intrauterine foetus corresponding to 20 weeks. Neck, Chest, Abdomen and Spine was normal. No normal limb movement noted. The patient presented with classic four limb involvement as follow:

- The shoulder – adducted and internally rotated. The right shaft humerus had intrauterine fracture.
- The elbow – extension contracture of the left elbow and flexion contracture of the right elbow
- The wrist – palmar flexion contracture with ulnar deviation and pronation of the hand.
- The hand – finger contractures distally flexion contractures of interphalangeal joints.

Metacarpophalangeal joints present with relative extension contractures. The thumb is adducted. Finger contractures are stiff and have significant deficiency of active finger movements;

- The hip –flexion, abduction, and external rotation contractures of varying degree.
- The knee –flexion contracture of varying severity
- The ankle joint and foot –severe talipes-equino-varus being reported in both side

Based on these findings arthrogryposis Multiplex congenita was made and the patient was advised for termination of pregnancy, but she continued the pregnancy. [Fig-1]. A male live fetus of birth weight 3500 gms was delivered. Baby cry immediately after birth and developed respiratory distress for which kept in NICU on Oxygen support for 2 days and then handed over to mother. Grossly showed the following features: Microcephaly with a space which admits tip of finger in anterior fontanelle, both eye cornea looks normal, ophthalmologic examination suggestive of same, Normal genitals. [Fig- 1,2]



Figure 1: Baby with limb deformities with right arm bandaging for intra uterine fracture shaft humerus



Figure 2: Normal External genitalia



Figure 3: Hand deformity



Figure 4: X-ray hip



Figure 5: Babygram suggestive of multiple bone deformities

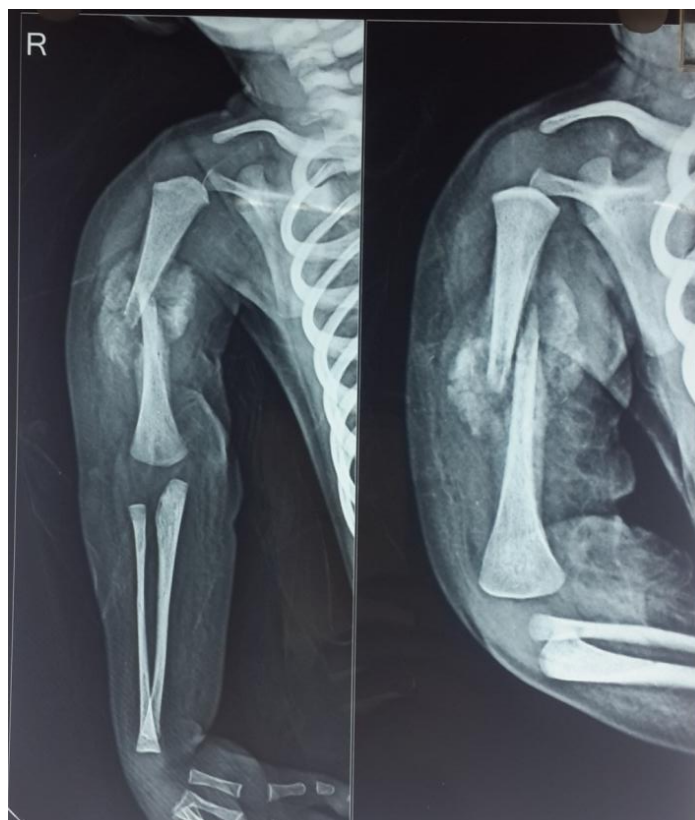


Figure 6: Intrauterine fracture shaft humerus with callous formation

2. Discussion

AMC literally means curving of joints (arthro "joint", gryposis "hooking"). It is characterized by multiple joints contractures seen especially in distal joints^{2,3,4,9}. Overall prevalence is one in 3000 and according to one study

conducted in 2011 in UK it is 11000-12000 among European live births. Males and females are affected equally. Cases are reported in Asian, African, and European descent^{6,7,8}. Exact cause is not known but its complex etiology includes fetal akinesia which can be due to fetal or maternal causes. Fetal causes include neurogenic, muscular

exposure to drugs and vascular compromised. Maternal causes include myasthenia gravis, infections, diabetes mellitus and multiple sclerosis. In 30% of cases a genetic cause can be identified. A research has shown that ZC4XZ point mutations, rearrangement and small deletions are associated with AMC and intellectual disability^{1,5,10}. Our patient was regularly followed in Paediatrics and Orthopedics clinic for management since his birth. Initially patient was evaluated and xrays were advised. Fracture shaft humerus was managed with strapping in arm adducted and internal rotation position. Bilateral congenital talipes-equino-varus were treated with weekly casting by "Ponseti method" for 6 weeks. The final cast is applied with the foot in the same maximally abducted position and dorsiflexed 15 degrees. A percutaneous achilles tenotomy is done to prevent development of a rocker-bottom deformity in bilateral side.

The foot is cast in the final position of approximately 70 degrees of abduction and 15 degrees of dorsiflexion for 3 weeks. Five or six casts usually are necessary to correct the clubfoot deformity.

After the removal of the final cast, the infant is placed in a Pawlik harness brace that maintains the foot in its corrected position (abducted and dorsiflexed). The brace is advised to wear 23 hours each day for the first 3 months after casting and then while sleeping for 3 to 4 years. Mother was trained for gentle exercises and manipulations for the contractures of different joints. Treatment of AMC is still a challenge, despite of multidisciplinary action no successful approach has been found yet¹¹.



Figure 7: Reduction in limb deformities after 7 months of follow up

3. Conclusion

The prevalence of arthrogryposis multiplex congenital is estimated 1/3000 to 1/10000 births. Arthrogryposis is not a specific diagnosis but a syndrome seen in different diseases that have in common the existence of multiple congenital contractures. Etiologies are multiple and can be maternal or fetal, neurogenic or myogenic and the prognosis depends essentially on the etiology. In this case the challenge for obstetricians is to evaluate the fetal and maternal prognosis, in order to insure the most accurate counselling and to be able to formulate a therapeutic action plan. Multidisciplinary care including neonatologist, orthopaedician, obstetrician and geneticist is always recommended.

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