Manipal Journal of Nursing and Health Sciences

Manuscript 1274

ARTHROGRYPOSIS MULTIPLEX CONGENITA (AMC) WITH AMYOPLASIA: A RARE CASE

Maria Pais Dr.

Leslie Edward Lewis Dr.

Jayashree Purkayastha Dr

Disha Shree B S Dr.

Case Report

Arthrogryposis Multiplex Congenita (AMC) With Amyoplasia: A Rare Case.

Maria Pais*, Leslie Edward Lewis, Jayashree Purkaystha, Disha Shree B S

Email: maria.v@manipal.edu

https://doi.org/10.55889/2582-7979.1274

Abstract

Background: The term "arthrogryposis multiplex congenita" (AMC) refers to nonprogressive contractures that develop in one or more body parts before birth (congenitally). A contracture is a condition where a joint becomes permanently stuck in a bent (flexed) or straightened (extended) position, limiting movement of the affected joint entirely or partially. Isolated congenital contractures, as opposed to arthrogryposis, are used to describe congenital contractures that only affect one body part. The most frequent solitary congenital contracture is clubfoot. Two or more different body parts are impacted by AMC. **Case report:** We report a case of arthrogryposis multiplex congenita (AMC) with bilateral hip, knee, and ankle dislocation in a neonate with facial dysmorphisms in addition to typical facial dysmorphisms, prominent forehead, retrognathia (small mouth), bulbous broad nose and no contraction in upper limbs. To the best of our knowledge, no research has shown arthrogryposis multiplex congenita with bilateral hip, knee, and ankle dislocation in neonates to date. In addition, the neonate was treated with symptomatic and serial manipulation of the clubfoot. **Conclusion:** Arthrogryposis multiplex congenita (AMC) associated with bilateral hip, knee, and ankle dislocation with facial dysmorphisms appears to be a rare association. There is a need to examine this association in the future.

Introduction

Arthrogryposis multiplex congenita (AMC) is an uncommon hereditary condition characterized by bilateral hip, knee, and ankle dislocations. It affects one out of every 3,000 live births, whereas amyoplasia affects one out of every 10,000 live births. X-linked recessive diseases primarily affect male fetuses. The X chromosome is unique to males. The disease is caused by just one recessive gene on the X chromosome. The male XY gene pair is made up of two genes, one on each chromosome of the Y chromosome. However, unlike the X chromosome, the Y chromosome does not have the majority of its genes (Schurz, H et al., 2019). Males are not protected as a result, and the X chromosome contains a recessive gene that causes illnesses such as hemophilia and Duchenne muscular dystrophy (DMD) (Gambineri, E, et al., 2003; RATES, I. & HOUR, P; 2012). Diminished foetal akinesia is characterized by the occurrence of numerous contractures at birth (movement). The specific cause is unknown; however, environmental variables (such as maternal illness and limited space), single-gene mutations (autosomal dominant, autosomal recessive, X-linked), chromosomal abnormalities, and various syndromes have been identified. The authors describe a newborn with AMC as a rare genetic syndrome characterized by bilateral hip, knee, and ankle dislocations and associated morbidity (Chen, C. P. 2012). In addition to other nursing intervention in early AMC diagnosis, it is critical to have appropriate prenatal check-ups and counselling to avoid complications.

Maria Pais[1]*, Leslie Edward Lewis[2], Jayashree Purkaystha[2], Disha Shree B S[3]

1 Assistant Professor - Senior Scale, Obstetrics and Gynecology Nursing, Manipal College of Nursing, Manipal Academy of Higher Education, Manipal, Udupi, Karnataka, India.

2 Professor, Department of Paediatrics, Kasturba Medical College, Manipal Academy of Higher Education, Manipal, Udupi, Karnataka, India.

3 Postgraduate Student (Obstetrics and Gynaecology), Kasturba Medical College,Manipal Academy of Higher Education, Manipal, Udupi, Karnataka, India.

Manuscript received: 10 January 2023
Revision accepted: 21 May 2023

*Corresponding Author

How to cite this article: Pais, M., Lewis, E. L., Purkaystha, J., Disha Shree, B. S. (2023). Arthrogryposis Multiplex Congenita (AMC) with Amyoplasia: A rare case. *Manipal Journal of Nursing and Health Sciences*, *9*(1), 58-61.

Case Report

A male neonate of 21 days old with a weak cry was brought to the neonatal unit of Kasturba Medical College Hospital in the district of Udupi. His upper arms and legs were thin and atrophic, and his joints lacked flexion. Absence of flexion in the lower limbs; normal sensation. At 39 weeks of gestation, he was delivered in the ambulance on the way to Kumta Hospital normally by vaginal birth to a 26-year-old mother weighing 2,500 grams at birth. There was no prior history of radiation exposure, birth trauma, or medication use., A consanguineous marriage or traumatic birth was not documented in the past. On clinical examination, the infant had a reduced activity level, a weak cry, a prominent forehead, retrognathia, a small mouth, and no contracture in the upper limbs. Figure 1 shows that the lower limbs were not flexed or contracted and that normal touch and pain perception were experienced. In addition, he had clubfeet on both sides, a dislocated knee on each side, and respiratory distress. A severe retrognathia was present, Barlow and Ortolani's manoeuvres were negative, and there were no midline lesions found in the spine. To avoid early morbidity, neonates underwent multidisciplinary treatment after receiving a clinical diagnosis of AMC (Figure 2). The initial serum calcium level was 11.6 mg/dl, indicating mild hypercalcemia, and subsequent calcium levels of 8.5 to 10.5 mg/dl were normal, indicating normal blood parameters. The X-ray revealed multiple dislocations.

Bilateral hip and knee joints with spine, and ankle joints were imaged using ultrasound technology, and the right-side haziness revealed a thin rim of fluid with a maximum thickness of 2.4 mm in the suprapatellar recess. Although there was no effusion within the joints, the bilateral femoral head epiphyses, knee, and ankle joints all appeared slightly widened. The nasal cavity, nasopharynx, oesophagus, larynx, hypopharynx, and upper oesophagus were all examined by an ENT specialist without finding any other associated abnormalities. Swallowing was accompanied by many secretions, and flexible laryngoscopy showed a good cough reflex. There were no organomegalies, the abdomen was soft, and it was not swollen.

Clinical features indicated that the infant had AMC with Amyoplasia. Dolichocephaly was detected by a simple MRI of the brain, and no obvious congenital abnormalities were found. The infant's foot, knee, and hip deformities were observed, and the corresponding serial manipulation and slab application were carried out. He was then treated with nasal oxygenation at a rate of 10 liters per minute and required FiO2 20 to 30% because of increased secretion. Because breastfeeding was challenging, expressed breast milk was administered via a feeding tube. For approximately one month, cautious management was used.

During hospitalization, the infant put on weight. The right hip joint was superolaterally dislocated on the infantogram, along with inferomedial and anteromedial dislocations of the left hip joint. The deformities were serially corrected with casts, Pavlik harnesses were suggested for deformity correction, and paediatric ortho consultation was sought. The baby gradually improved as a result of the oromotor stimulatory exercises.

On the 19th day of life, the baby was observed to have episodes of desaturation and tachycardia, raising the possibility of seizures. The baby was started on syrup Levera (Levetiracetam) 100 mg/ml 0.2 ml twice daily, and an EEG was done, and it was normal. The baby was noted to have improved swallowing coordination reflexes. Serial manipulation of the deformity with a cast was continued, and parents were taught about chest physiotherapy. Genetics consultation was sought, and parents were counselled regarding the genetic basis of the disease and the need for preconceptual screening at the next pregnancy. The baby had frequent desaturations, apnoea, and bradycardia requiring intermittent resuscitation. In view of persistent pooled oral secretions, oral Cuvposa (glycopyrrolate) was added (200 mcg/kg/day). Serial manipulation of the deformity with a cast was continued.

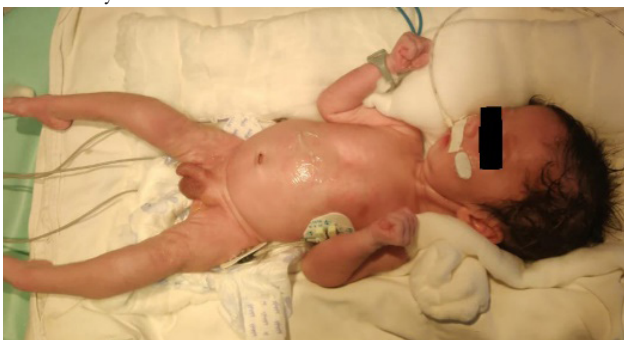

Figure 1: Prominent forehead, retrognathia, and contracture in lower limbs

Figure 2: Arthrogryposis multiplex congenita cast applied to avoid early morbidity.

Multiple rigid joint deformities are present at birth in Arthrogryposis Multiplex Congenita (AMC), and these contractures are typically nonprogressive. Birth defects may impact the central nervous system and are part of a more extensive syndrome. Currently, it is unclear exactly how AMC develops. When congenital contractures affect only one area of the body, they are referred to as isolated congenital contractures rather than AMCs. The most common type of isolated congenital contracture is clubfoot. A condition that affects two or more different body regions can be referred to as AMC. According to Van der Linden et al. (2016), the most common type of AMC is amyoplasia. AMC is occasionally used interchangeably. The five common clinical characteristics are joint rigidity, joint dislocation, particularly in the hips and knees, atrophy, and even the absence of muscles or muscle groups, intact sensation, and occasionally weakened deep tendon reflexes. Due to the need for careful observation of the fetal movements during the ultrasonographic technique, which is not possible until the 16th to 18th week of pregnancy, the severity of joint deformities and related clinical abnormalities varies from patient to patient. According to Lee (H. 2005), 75% of AMC cases are not identified before birth. The lower limbs and hips are typically dislocated, the knees are extended, and the feet are equinovarus (as in the present case). It may affect all four limbs in a symmetrical pattern. Genetic disorders are not typically linked to it. Atrophic muscle tissue replaced by fibrous and fatty tissue is one of its defining features. The case described here is unique because only the lower limbs were affected, which should be noted. Over 350 genes play a role in the emergence of AMC, which has been linked to more than 400 distinct medical conditions. Most patients (70–85%) have associated neurological alterations, but AMC is typically not genetic in origin. These genetic changes are typically autosomal recessive in nature. TPM2 and MYBPC1 gene mutations can cause distal arthrogryposis type 1.

Despite the fact that many cases are sporadic or idiopathic, these genes are active (expressed) in muscle cells, where they collaborate with other muscle proteins to control the force with which the muscle fibres contract (Borensztajn, K., & Spek, 2005; Skaria, P., & Ahmed, A. 2019; Desai, D et al., 2020). Due to the wide range of clinical presentations and the significant number of triggering mutations, it is challenging to pinpoint the underlying cause of a specific type of AMC. Surgery reconstructions, early orthopaedic management, physical therapy, and rehabilitation are the main components of the treatment plan for AMC, which aims to improve the quality of life by providing multidisciplinary treatment modalities. It was difficult to categorize the deformity in this case because it did not affect all four limbs equally. A positive clinical course was also shown by the patient. Remember that the patient's condition could also be explained by a single congenital dislocation of the knee or a dislocation associated with an AMC. The infant also displayed some clinical traits that were consistent with the alternative diagnosis of congenital knee dislocation, such as joint instability, reduction of dislocation with a snap or piston, muscle retraction (hip adductors, quadriceps of the knee), restricted range of motion (hip abduction), and the presence of skin folds or grooves, which became more obvious after full recovery. A series of manipulations using casts were applied to the bilateral hip, knee, and ankle joints. Regular use of oromotor stimulation and physical therapy. To treat both AMC and congenital knee dislocation, the recommended physical therapy outlined above should be continued for 4–8 weeks; if therapy is unsuccessful, a splint or even surgery may be advised. There is little research on these cases' follow-up from birth to skeletal maturity.However, the need for aggressive therapy, physical therapy, occupational therapy, and multiple surgeries may be necessary in extreme cases.

Conclusion

Multiple limb dislocations/deformities can result from either an extrinsic cause (amniotic fluid disorder) or an intrinsic cause (pathological tissues, arthrogryposis). AMC can be identified early during the postnatal period and can be treated by providing proper management and avoiding unnecessary procedures. It is important to differentiate both, as the extrinsic form is much more amenable to conservative management with a good prognosis.

Source of support: None
Conflict of interest: None declared
Source of support in the form of grants: None.

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