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Arthrogryposis Multiplex Congenita In A Preterm Female Neonate: A Case Report Of Laryngomalacia, Multiple Limb And Craniofacial Anomalies, Spinal Dysraphism, And Teratologic Hip Dislocation

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Abstract

Background: Arthrogryposis Multiplex Congenita (AMC) is a rare, heterogeneous group of congenital disorders characterized by multiple joint contractures present at birth due to decreased fetal movement during intrauterine development. The condition presents with a broad clinical spectrum ranging from isolated limb abnormalities to complex multisystem involvement affecting neurological, respiratory, and craniofacial structures.

Case Presentation: We report a 35-week preterm newborn girl born via spontaneous vaginal delivery with a birth weight of 1.9 kg presenting with multiple congenital anomalies consistent with AMC. Clinical findings included bilateral limb contractures with cortical thumb deformities, dorsiflexed feet, and calcaneovalgus deformities. Craniofacial anomalies comprised bilaterally low-set ears and micrognathia. Orthopedic evaluation revealed left teratologic hip dislocation and right femur shaft fracture. A lumbosacral hair tuft prompted spinal imaging, which confirmed spinal dysraphism with cord tethering. Respiratory assessment identified laryngomalacia contributing to inspiratory stridor and mild respiratory distress. Neurological examination demonstrated generalized hypotonia and markedly reduced spontaneous movements. Multidisciplinary management involving neonatology, orthopedics, neurology, pulmonology, and genetics was initiated with respiratory support, early physiotherapy, and staged surgical planning. Written informed consent was obtained from the patient's legal guardians for publication of this case report.

Conclusions: This case emphasizes the multisystemic complexity of AMC and the significance of comprehensive clinical and radiological evaluation for accurate diagnosis. Early multidisciplinary intervention is crucial for optimizing functional outcomes and managing associated complications including spinal dysraphism and respiratory anomalies. Genetic counseling and long-term specialized follow-up remain integral components of individualized care for AMC patients.

Keywords: Arthrogryposis; Contracture; Laryngomalacia; Spina Bifida Occulta; Congenital Hip Dislocation; Patient Care Team

Introduction

Arthrogryposis Multiplex Congenita (AMC) represents a heterogeneous group of congenital disorders characterized by multiple joint contractures present at birth, affecting approximately 1 in 3,000 live births.[1] The term "arthrogryposis" derives from the Greek words "arthro" (joint) and "grypos" (curved), reflecting the characteristic joint deformities that define this condition.[2] AMC results from

decreased fetal movement (fetal akinesia) during critical periods of intrauterine development, leading to impaired joint mobility and subsequent contracture formation.[3]

The pathogenesis involves diverse etiologies affecting neuromuscular function, including disorders of the central nervous system, peripheral nervous system, muscles, connective tissue, or environmental factors that limit fetal movement.[4,5] The condition presents with a broad clinical spectrum ranging from isolated distal limb involvement to complex multisystem syndromes with associated neurological, craniofacial, cardiac, and respiratory abnormalities.[6,7]

Classification systems distinguish between amyoplasia (the most common form affecting limbs symmetrically) and distal arthrogryposis (primarily affecting hands and feet), with over 400 distinct conditions now recognized within the AMC spectrum.[8,9] Early diagnosis relies on comprehensive clinical evaluation supported by imaging studies and genetic testing when indicated.[10]

Management requires a multidisciplinary approach involving orthopedic surgery, physical therapy, occupational therapy, and specialized care for associated anomalies.[11] While prognosis varies significantly depending on the underlying etiology and extent of involvement, early intervention with passive range-of-motion exercises and appropriate surgical procedures can substantially improve functional outcomes and quality of life.[12,13]

We present a case of a preterm neonate with AMC accompanied by multiple limb contractures, craniofacial anomalies, laryngomalacia, and teratologic hip dislocation to illustrate the complex multisystem nature of this condition and highlight important diagnostic and therapeutic considerations.

Case Presentation

A female neonate was born at 35 weeks gestation via spontaneous vaginal delivery to a 28-year-old gravida 2, para 1, living 1, dead 1 mother. The pregnancy was unremarkable with no history of maternal illness, teratogenic exposures, or consanguinity. Routine prenatal ultrasounds were normal until the third trimester when reduced fetal movements were noted. Birth weight was 1.9 kg (small for gestational age, <10th percentile), length 45 cm, and head circumference 32 cm. APGAR scores were 6 and 8 at one and five minutes, respectively. The infant failed to cry immediately at birth and exhibited mild respiratory distress characterized by inspiratory stridor and intermittent oxygen desaturation requiring supplemental oxygen.

Physical examination revealed multiple congenital anomalies consistent with arthrogryposis multiplex congenita (Figure 1). Craniofacial features included bilaterally low-set ears and micrognathia. The upper extremities showed fixed flexion and adduction deformities of both thumbs (cortical thumb deformities) with limited wrist extension and notably small, narrow wrists. Lower limb examination demonstrated flexion deformity of the right knee with dorsiflexion of the right foot, mildly elevated left second toe, bilateral flat feet, and calcaneovalgus deformities. Tenderness was elicited on palpation over the right thigh, raising suspicion for fracture.

Neurological assessment revealed generalized hypotonia, severely diminished spontaneous limb movements, weak but present primitive reflexes (Moro and grasp reflexes), and hypoactive deep tendon reflexes throughout. Cranial nerve examination was intact. A distinct tuft of hair overlying the lumbosacral region was noted, raising suspicion for underlying spinal dysraphism.

Baseline laboratory investigations including complete blood count, comprehensive metabolic panel, liver function tests, renal function tests, and sepsis screening were within normal limits for gestational age. Radiographic evaluation confirmed a right femoral shaft fracture and demonstrated left teratologic hip dislocation with the femoral head displaced superiorly and laterally (Figure 2). Clear lung fields were seen on chest radiography with no structural abnormalities.

Spinal ultrasonography confirmed spinal dysraphism, demonstrating tethering of the spinal cord at the lumbosacral level. Flexible fiberoptic laryngoscopy revealed laryngomalacia with inspiratory collapse of supraglottic structures, explaining the observed stridor and respiratory symptoms.

The constellation of clinical findings led to the diagnosis of arthrogryposis multiplex congenita with associated complications including multiple joint contractures affecting upper and lower extremities, right femoral shaft fracture, left teratologic hip

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dislocation, spinal dysraphism with cord tethering, and laryngomalacia with mild respiratory compromise.

A multidisciplinary management approach was immediately instituted involving neonatology, orthopedic surgery, neurology, pulmonology, and genetics teams. The patient received respiratory assistance with supplemental oxygen and careful airway monitoring. The fractured femur was managed conservatively with immobilization and pain control. Early passive range-of-motion physiotherapy was initiated to maintain joint mobility and prevent progression of contractures. Orthopedic planning included staged surgical interventions for the hip dislocation and long-term management of limb deformities. Neurological monitoring was established for potential complications related to spinal dysraphism, with neurosurgical consultation obtained for future management planning.

The patient remained clinically stable during the initial hospitalization period with gradual improvement in respiratory status. Genetic counseling was provided to the family regarding the heterogeneous etiology of AMC, recurrence risks, and the potential benefits of genetic testing. Written informed consent was obtained from the patient's legal guardians for publication of this case report.

Discussion

This case illustrates the complex, multisystem nature of arthrogryposis multiplex congenita and highlights important clinical considerations in this heterogeneous disorder. The combination of severe musculoskeletal deformities, spinal dysraphism, and respiratory involvement in a preterm neonate represents a challenging presentation requiring comprehensive evaluation and coordinated multidisciplinary care.

The presence of spinal dysraphism with cord tethering provides insights into AMC pathogenesis. While decreased fetal movement is the final common pathway leading to joint contractures, the underlying neurological abnormality likely contributed significantly to the severe phenotype observed.[14] Spinal cord tethering can impair motor neuron function and reduce fetal movement during critical periods of musculoskeletal development, potentially explaining the severity and distribution of contractures.[15] This emphasizes that AMC should be conceptualized as a multisystem disorder requiring integrated diagnostic approaches.

The cutaneous marker of a lumbosacral hair tuft proved valuable in prompting appropriate imaging studies. This highlights the importance of careful dermatological examination in neonates with AMC, as occult spinal dysraphism may be present in a significant proportion of cases.[16] Early identification of spinal abnormalities influences both prognosis and management strategies, particularly regarding timing of neurosurgical interventions.

The presence of laryngomalacia represents an important but often underemphasized complication of AMC. While joint contractures dominate the clinical presentation, respiratory involvement can significantly impact morbidity and mortality, particularly in preterm infants.[17] Previous studies have reported respiratory complications in approximately 10-15% of AMC cases, but the true incidence may be higher due to underreporting.[18] The combination of laryngomalacia with underlying neurological impairment created a challenging respiratory scenario, emphasizing the need for proactive pulmonary assessment in all AMC patients.

The right femoral shaft fracture highlights an often overlooked aspect of AMC management. The combination of decreased fetal movement, potential osteopenia of prematurity, and mechanical stress of delivery can predispose these infants to fractures.[19] This discovery has significant ramifications for obstetric management and delivery planning, as cases with severe contractures may benefit from individualized delivery strategies to minimize trauma.

The left teratologic hip dislocation represents one of the most challenging orthopedic aspects of AMC management. Unlike typical developmental dysplasia of the hip, teratologic dislocations occur in utero and are often associated with more severe contractures and poorer outcomes.[20] The timing of surgical intervention in preterm infants with AMC remains controversial, as early surgery must be balanced against the risks of anesthesia and surgery in unstable neonates.

This case reinforces the critical importance of early multidisciplinary intervention in AMC management. The involvement of multiple specialties from the immediate postnatal period exemplifies best practice approaches.[21] The integration of various specialties

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allows for comprehensive assessment of all affected systems and coordinated treatment planning. The early initiation of passive range-of-motion physiotherapy aligns with current evidence supporting aggressive conservative management to maintain joint mobility and prevent contracture progression.[12]

The provision of genetic counseling addresses an important aspect of AMC management extending beyond immediate medical care. With over 400 recognized conditions within the AMC spectrum and diverse inheritance patterns, accurate genetic counseling requires expertise in both clinical genetics and the specific phenotypic features present in each case.[9] The heterogeneous nature of AMC means that recurrence risks can vary dramatically.

This case demonstrates key clinical imperatives for AMC management: systematic evaluation for associated anomalies beyond joint contractures, proactive respiratory assessment, early multidisciplinary intervention, and genetic counseling. Healthcare providers caring for AMC patients should maintain high suspicion for occult spinal dysraphism and respiratory complications, as early recognition can significantly impact management and outcomes.

References

1. Lowry RB, Sibbald B, Bedard T, Hall JG. Prevalence of multiple congenital contractures including arthrogryposis multiplex congenita in Alberta, Canada, and a strategy for classification and coding. Birth Defects Res A Clin Mol Teratol 2010; 88: 1057-1061
2. Hermansen MC, Hall JG, Rode C, Heuther C, Leppig KA. The range of publications on arthrogryposis multiplex congenita from 1995 to 2022--A scoping review. Am J Med Genet A 2023; 191: 1570-1580
3. Bamshad M, Van Heest AE, Pleasure D. Arthrogryposis: a review and update. J Bone Joint Surg Am 2009; 91 Suppl 4: 40-46
4. Hall JG. Arthrogryposis multiplex congenita: etiology, genetics, classification, diagnostic approach, and general aspects. J Pediatr Orthop B 1997; 6: 159-166
5. O'Flaherty P. Arthrogryposis multiplex congenita. Neonatal Netw 2001; 20: 13-20
6. Kalampokas E, Kalampokas T, Sofoudis C, Deligeoroglou E, Botsis D. Diagnosing arthrogryposis multiplex congenita: a review. ISRN Obstet Gynecol 2012; 2012: 264918
7. Drachman DB, Banker BQ. Arthrogryposis multiplex congenita. Case due to disease of the anterior horn cells. Arch Neurol 1961; 5: 77-93
8. Stevenson DA, Carey JC, Palumbos J, Rutherford A, Dolcourt J, Bamshad MJ. Clinical characteristics and natural history of Freeman-Sheldon syndrome. Pediatrics 2006; 117: 754-762
9. Hall JG, Reed SD, Greene G. The distal arthrogryposes: delineation of new entities--review and nosologic discussion. Am J Med Genet 1982; 11: 185-239
10. Gordon N. Arthrogryposis multiplex congenita. Brain Dev 1998; 20: 507-511
11. Staheli LT, Chew DE, Elliott JS, Mosca VS. Management of hip dislocations in children with arthrogryposis. J Pediatr Orthop 1987; 7: 681-685
12. Palmer PM, MacEwen GD, Bowen JR, Mathew PA. Passive motion therapy for infants with arthrogryposis. Clin Orthop Relat Res 1985; 194: 54-59
13. Sarwark JF, MacEwen GD, Scott CI Jr. Amyoplasia (a common form of arthrogryposis). J Bone Joint Surg Am 1990; 72: 465-469
14. Fahy MJ, Hall JG. A retrospective study of pregnancy complications among 828 cases of arthrogryposis. Genet Couns 1990; 1: 3-11
15. Hall JG. An approach to congenital contractures (arthrogryposis). Pediatr Ann 1981; 10: 15-26
16. Lim KB, Tan BH, Peter SD, Eng AK, Thng CH, Tan KL. Spina bifida occulta in arthrogryposis multiplex congenita: a report of six cases. Singapore Med J 2001; 42: 26-29
17. Swinyard CA, Bleck EE. The etiology of arthrogryposis (multiple congenital contractures). Clin Orthop Relat Res 1985; 194: 15-29
18. Herring JA. Tachdjian's Pediatric Orthopaedics. 3rd ed. Philadelphia, PA: W.B. Saunders Company; 2002

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19. Beals RK, Hecht F. Congenital contractural arachnodactyly. A heritable disorder of connective tissue. J Bone Joint Surg Am 1971; 53: 987-993

20. Weinstein SL. Natural history of congenital hip dislocation (CDH) and hip dysplasia. Clin Orthop Relat Res 1987; 225: 62-76

21. Sells JM, Jaffe KM, Hall JG. Amyoplasia, the most common type of arthrogryposis: the potential for good outcome. Pediatrics 1996; 97: 225-231

Figures

Figure 1. Clinical photograph showing arthrogryposis multiplex congenita in a preterm neonate Clinical photograph demonstrates characteristic limb contractures including fixed flexion and adduction deformities of both thumbs (cortical thumb deformities), bilateral calcaneovalgus foot deformities, and generalized joint contractures affecting upper and lower extremities. Patient shown with necessary medical monitoring equipment in the neonatal intensive care unit. Written informed consent obtained from patient's legal guardians.

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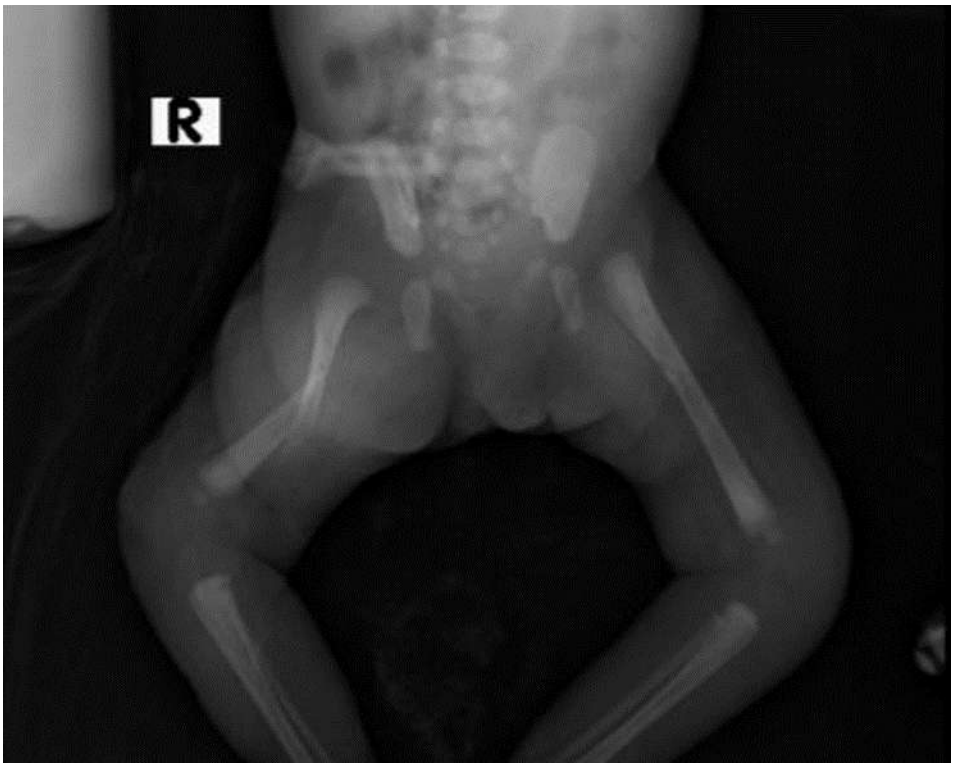
Figure 2. Anteroposterior radiograph demonstrating orthopedic complications in arthrogryposis multiplex congenita

Radiographic image shows left teratologic hip dislocation with femoral head displaced superiorly and laterally, and right femur mid-shaft fracture with transverse pattern and minimal displacement. Hip dislocation occurred in utero and is commonly associated with arthrogryposis multiplex congenita. Femoral fracture likely occurred during delivery due to decreased fetal movement, potential osteopenia of prematurity, and mechanical stress associated with the condition.



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