



Arthrogryposis Multiplex Congenita And Myelomeningocele In Lebanon: Case Report And Review Of Literature

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Abstract:

Arthrogryposis multiplex congenita is a rare, non-progressive congenital disorder characterized by multiple joint contractures associated with akinesia and connective tissue fibrosis that can be either generalized or limited to the upper and/or lower extremity. AMC is a group of conditions with varied etiologies including myopathic processes, neuromuscular end-plates, connective tissue abnormalities, intra-uterine space limitation and vascular compromise, maternal factors, metabolic disturbances and neuropathic processes [2]. Neural tube defects including myelomeningocele have been identified as a cause of arthrogryposis in lower extremities [2]. We present, to the best of our knowledge, the first reported case in Lebanon of lower limb arthrogryposis due to lumbosacral myelomeningocele.

Keywords:

Arthrogryposis, Myelomeningocele, Neural Tube Defects, Club Foot

Introduction:

Arthrogryposis has been identified as multiple congenital contractures for over a century [1]. It has been used as a disease diagnosis, but now it is referred to as a symptom that is included but not limited to a syndrome called Arthrogryposis multiplex congenita (AMC). AMC is a group of conditions with varied etiologies including myopathic processes, neuromuscular end-plates, connective tissue abnormalities, intra-uterine space limitation and vascular compromise, maternal factors, metabolic disturbances and neuropathic processes [2]. AMC has complex clinical features which include multiple prenatal congenital contractures in several joints of the

body [3]. Disturbances in both central and peripheral neurologic processes are one of the etiological factors leading to AMC. Lower limb arthrogryposis has been seen with myelomeningocele due to abnormal neural tube development leading to secondary limitation of active fetal movements and congenital multiple joints contractures [2,4]. We present, to the best of our knowledge, the first reported case in Lebanon of lower limb arthrogryposis due to **lumbosacral myelomeningocele**.

Case study:

An 8 days old female presented to the pediatric department with a history of multiple contractures in

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Fig-1:
Lumbosacral Myelomeningocele.

lower limbs since birth and oozing lumbosacral mass suggestive of myelomeningocele (**Fig-1**). A detailed neonatal, maternal, prenatal, and family history was taken. The patient was G₁PA₀, neonate of an uneventful pregnancy, born by C-section due to the breech presentation with normal **APGAR** score (7 and 9 at 1 and 5 minutes respectively). No identifiable risk factor for arthrogryposis was identified except his lumbosacral meningomyelocele. On the 5th month, a screening prenatal ultrasound showed spinal anomaly. Both parents were healthy, young and non-consanguineous. Mother received folic acid supplementation starting the first month of pregnancy. No family history of similar abnormalities

was reported.

Upon examination, the patient had a weight of 2890 gms, length of 35 cm and head circumference of 35 cm. She had no facial dysmorphism or skin lesion except oozing lumbosacral myelomeningocele with foul smelling discharge. Cardiac and respiratory exams were normal. On musculoskeletal exam, the patient had an internal rotation with the extension of both hip joints; internal rotation and extension of the left leg and right leg; both knees were extended with a bilateral club foot (**Fig-2,3**). Neurologically, she had bilateral spastic lower extremities with absent bilateral deep tendon reflexes.



Fig- 2,3:
Internal rotation with extension of both hip joints; internal rotation and extension of the left leg and right leg; both knee were extended with bilateral club foot

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A diagnosis of lower extremity arthrogryposis was considered with early onset infected myelomeningocele. Full blood count result was unremarkable except with a mildly high white blood count and CPR. Brain ultrasound has shown an increased size of the ventricles mainly on the right side suggestive of hydrocephalus. Urea and electrolytes were normal except for a mild increase in potassium. Abdominal ultrasound was normal. The CT scan of the brain (**Fig-4**) showed dilatation of the lateral ventricles more on the left side suggestive of mild left hydrocephalus. CT lumbar spine showed

7.5x6 cm myelomeningocele (**Fig-5,6**).

She was managed using antibiotics (vancomycin and meropenem) for open wound infection and for sepsis prophylaxis. The cerebrospinal fluid culture showed no growth. The lower limbs kept a fixed position with plaster-of-Paris cast after an orthopedics consultation. She was doing well, and after 1 week she was discharged against medical advice due to financial reasons. Thus, electroencephalogram, muscle biopsy, cytologic studies, genetic studies were not done.

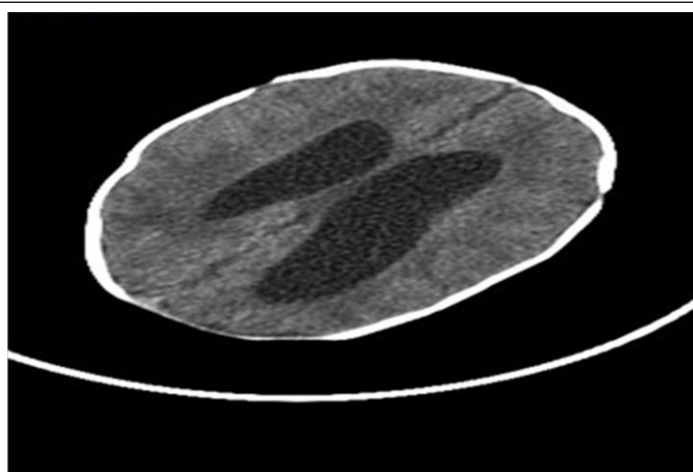


Fig-4:

T2 axial image of the brain without contrast showing mild dilatation of the ventricles, more on the left lateral ventricle. The cisterns and sulci are within normal limits.



Fig-5:

Axial image of lumbar spine without IV contrast showing 7.5x6 cm fluid containing, well defined formation lined by dural isodense material. It is rising from an 8 mm posterior elements defects at the S1-2 level and bulging posteriorly in the lower back suggestive of myelomeningocele.

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Fig-6:
Mid-sagittal CT scan of the lumbar spine showing sacral myelomeningocele.

Discussion:

Arthrogryposis multiplex congenita is a rare, non-progressive congenital disorder characterized by multiple joint contractures associated with akinesia and connective tissue fibrosis that can be either generalized or limited to the upper and/or lower extremity. Multiple etiologies were identified and they include but are not limited to myopathic, neurogenic, space limitation, neuromuscular end-plate, and connective tissue abnormalities. Neurogenic AMC is far more common than the other subtypes [5], and the associated congenital anomalies are more frequent in the neurogenic form. The involvement of the nervous system in AMC was studied by Banker and Engel who showed that patients with neurogenic AMC had dysgenesis of the anterior horn, spinal cord and/or brainstem [6]. Some patients had dysgenesis of both the spinal cord and the entire brain [2]. Other neurologic abnormalities leading to AMC include nerve formation, structure and/or function, a defect in myelination, failure to prune axons, and central nervous system dysfunction.

Neural tube defects including myelomeningocele have been identified as a cause of arthrogryposis in lower extremities [2]. It can be secondary to functional muscle lacking innervated antagonist, denervated muscles reacting spastically rather than flaccidly, due to intact reflex arc without otherwise intact spinal pathways or it can be acquired from the cumulative effect of weight bearing across an unbalanced joint.

Nevertheless, the deformities roughly stratify according to the level of neurologic involvement. **Clubfeet** are by far the most common deformity seen in patients with myelomeningocele present in 30 to 50% of patients, found most commonly in the midlumbar lesions (L3 and L4) and are the most difficult to treat [6]. Sharrard et al. noted that the most severe presentation was those of an L4 with accompanied spasticity [7].

To the best of our knowledge, this is the first case report in Lebanon of myelomeningocele associated with lower extremity arthrogryposis. Our patient presented with a combination of lumbosacral myelomeningocele and diffuses spastic lower extremity contracture associated with akinesia and bilateral club foot. According to the literature, the management of our case should be as early as possible to benefit the most from the suppleness of the newborn. Therapy should encompass a multidisciplinary approach including a prenatal diagnosis, genetic counseling, genetic studies to rule out any possible syndrome depending on the clinical evaluation, physical therapy, Orthopedic care with surgical procedures for the limb deformities and the contractures including soft tissue release, talectomy, external fixator correction and serial casting that should be delayed till 4 months. Neurosurgical care for correction of the myelomeningocele is needed with a close follow-up due to the high recurrence rate of foot deformities.

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Conclusion:

Lower extremity arthrogryposis with myelomeningocele is a prenatal diagnosis that requires high clinical suspicion and early multidisciplinary intervention, genetic counseling and surgical approach to decrease the morbidity of such entity with a close follow-up due to a high recurrence rate of the foot deformities, especially club foot and improving quality of life.

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