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Arthrogyposis in a Case of Chiari Malformation II: First Case Report in a Mediterranean Population

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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Conflict of interest: None declared

Patient: Male, 28
Final Diagnosis: Arthrogyposis in a Chiari II malformation
Symptoms: Failure to thrive
Medication: —
Clinical Procedure: —
Specialty: Orthopedics and Traumatology

Objective: Congenital defects/diseases





Background: Arthrogyposis multiplex congenita is a multifactorial syndromic or non-syndromic group of conditions consisting of multiple congenital contractures of the body, of unknown etiology. It is associated with a heterogeneous group of disorders that include but are not limited to processes such as myopathic and neuropathic. Neural tube defect is a neuropathic disorder that incorporates myelomeningocele that might be either isolated or within a spectrum of multiple diseases.

Case Report: This is a case report of a 28-day-old male born with lower limb arthrogyposis with myelomeningocele and Chiari II malformation in a Mediterranean population.

Conclusions: Lower extremity arthrogyposis with myelomeningocele and Chiari II malformation is a prenatal diagnosis that requires high clinical suspicion, early multidisciplinary intervention, and genetic counselling. As long as new approaches are being explored in the management of such cases, babies born now with neural tube defects can expect better quality of life.

MeSH Keywords: Arnold-Chiari Malformation • Arthrogyposis • Meningomyelocele

Full-text PDF: <https://www.amjcaserep.com/abstract/index/idArt/914870>

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Background

Arthrogryposis has been identified as multiple congenital contractures in the body, of undisclosed etiology [1]. It has been used as a disease diagnosis, but now it is referred to as one of the symptoms of a syndrome called arthrogryposis multiplex congenita. 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 upper and/or lower extremities. It is a group of conditions with varied inauguration including myopathic processes, neuromuscular end-plates, connective tissue abnormalities, intra-uterine space limitation, vascular compromise, maternal factors, metabolic disturbances, and neuropathic processes. Disturbances in both central and peripheral neurologic processes are one of the leading etiological factors [2]. Arthrogryposis multiplex congenita has a complex clinical feature with multiple prenatal congenital contractures in several joints of the body [3]. It has been associated with myelomeningocele leading to secondary limitation of active fetal movements and congenital multiple joints contractures [2–4]. Nonetheless, to the best of our knowledge, this report presents the first case of lower limb arthrogryposis with Chiari II malformation in the Mediterranean population.

Case Report

A 28-day-old male, whose parents were non-consanguineous, was delivered by planned C-section to a G2P2A0 mother known to have Thalassemia minor. The infant has an APGAR score of 7 at 1 minute and an APGAR score of 10 at 5 minutes. During prenatal follow-ups, the mother was on iron supplements only and underwent echocardiography and morphological scans. Echocardiography was done at the end of the fifth month of

pregnancy and showed hydrocephalus and spina-bifida (suspected). At the beginning of 6 months of pregnancy, a morphological scan was performed to confirm the presence of spina-bifida, hydrocephaly, and lower limb paralysis. During admission, at the age of 1 week, the patient presented with a fever of 38.5°C, had a weight of 2.65 kg (at birth it was 2.4 kg), a head circumference of 39.5 cm, and a body length of 44 cm. He was transferred to the neonatal intensive care unit, was diagnosed with sepsis, and was administered broad spectrum antibiotics for 2 weeks. Upon physical examination (Figure 1A–1C), the patient was conscious, non-cyanotic, non-icteric, and in no distress. Cardiovascular, respiratory, and abdominal examinations were intact. Facial features included frontal protrusion and abnormal eyelid movements. During neurological examination, the patient had normal upper extremity reflexes including palmar reflex and deep tendon reflexes. However, the lower extremities were fully extended at the knee joint and flexed 45 degrees at the hip joint with extensive contraction and limited passive range of motion due to joint contractures and hyper spasticity. The deep tendon reflexes could not be elicited. In addition, our patient had hypertonicity, spastic lower extremity hemiplegia, and bilateral club feet deformity. On examination, the patient had several erythematous purulent clustered lumbosacral lesions with greenish discharge on his back. Magnetic resonance imaging (MRI) of the brain was done during admission and showed Bud Chiari II malformation with ventricular dilatation, suggestive of hydrocephalus (Figure 2). Moreover, MRI of the spine showed lumbar dysraphism with large subcutaneous myelomeningocele and tethered cord syndrome (Figure 3). After several days, our patient was hemodynamically stable, taking food by mouth and gaining weight, and discharged home.



Figure 1. (A–C) Hypertonicity and spastic lower extremity hemiplegia, with fully extension at knee joint and 45-degree flexion at the hip joint and extensively contracted with limited passive range of motion. Bilateral club feet deformity. Erythematous purulent clustered lumbosacral lesion with greenish discharge.

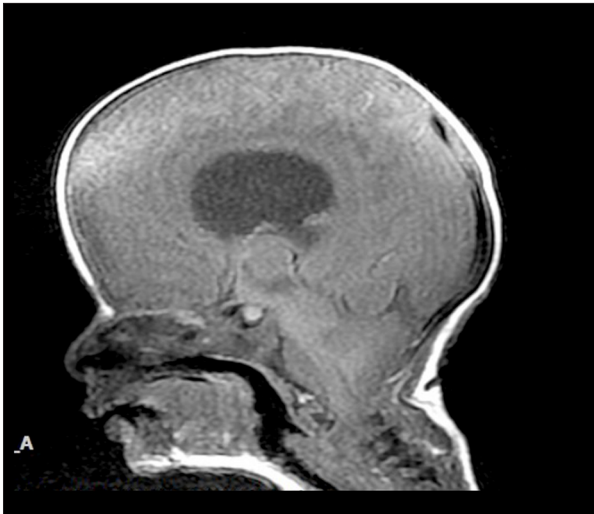


Figure 2. Magnetic resonance imaging of the brain without intravenous contrast showed small posterior fossa with downward migration of tonsils and segment of hemisphere and associated cervicomedullary kinking. There was also moderate platybasia, flattening and obliteration of the fourth ventricle. The ventricle system is dilated, bilateral well defined cystic and seen revealing blood fluid level with focal deformity of frontal bone suggesting perinatal intracranial hemorrhage. The findings suggest Chiari II malformation with associated significant ventricular dilatation.

Discussion

Arthrogryposis multiplex congenita can be neurogenic, myopathic, or of other subtypes. The neurogenic subtype is by far more common than the other subtypes, and the associated congenital anomalies are more frequent in the neurogenic form [5]. Nervous system involvement in arthrogryposis multiplex congenita was studied by Banker and Engel. Their study showed that patients with the neurogenic form of arthrogryposis multiplex congenita had dysgenesis of the anterior horn, spinal cord, and/or brainstem [5]; yet some patients had dysgenesis of both the spinal cord and the entire brain [2]. Other neurologic abnormalities leading to arthrogryposis multiplex congenita include nerve formation, structural and/or functional 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 the lower extremities [2]. Pathophysiology can be divided into 3 groups: 1) functional muscles lacking innervated antagonists, 2) denervated muscles reacting spastically rather than flacidly due to an intact reflex arc without an intact spinal pathway, and 3) weight-bearing cumulative effect across an unbalanced joint. Nevertheless, the deformities roughly stratify



Figure 3. Magnetic resonance imaging of the lumbar done using T1, SE, T2 TSE followed by axial T1, SE, T2 TSE. No intravenous contrast dye was used. It showed extensive posterior element defect extending from T1 thru S1 vertebral level with large posterior epidural 3.6×1.5 cm myelomeningocele. It also showed L5 S1 ill-defined ending suggestive of tethered cord syndrome. There was also evident kyphosis and thoracolumbar deformity.

according to the level of the neurologic involvement. Club feet are by far the most common deformity seen in patients with myelomeningocele. It is present in 30% to 50% of these patients, most commonly in the mid-lumbar lesions (L3 and L4); these patients are the most difficult to treat [6]. The most severe presentation, however, as noted by Sharrard et al., is that of an L4 lesion with accompanied spasticity [7].

Hydrocephalus is due to the impaction of the posterior fossa contents on the foramen magnum. If cerebrospinal fluid outflow is blocked or impaired at the foramina of Luschka and Magendie, progressive ventriculomegaly results. In McLone and Knepper's theory, both open neural tube defects and incomplete spinal occlusion allow cerebrospinal fluid to drain through the central canal, and thus, it is not maintained in the ventricular system [8]. Moreover, ventricular distention

is required to induce both neural and calvarial development. Without this ventricular cerebrospinal fluid driving force, the posterior fossa will never fully develop [8].

The MRI of the brain and spinal cord of our patient revealed a Chiari malformation II with tethered cord and lumbosacral dysraphism. Chiari II malformation (brainstem herniation) mainly presents with respiratory symptoms including inspiratory stridor (due to cranial nerve X injury) and prolonged expiratory apnea with cyanosis (PEAC), gastrointestinal symptoms, neurogenic dysphagia with chronic aspiration, choking, and prolonged feeding time which can be due to cranial nerve IX and X injury. In addition, paraparesis or quadriparesis, hypotonia, opisthotonus, nystagmus, weak cry, and developmental delay are all neurologic symptoms that can also be associated with Chiari II malformation. Nonetheless, we report the first case of Chiari malformation II with arthrogryposis.

In regard to prevention of this patient's neural tube defect (myelomeningocele), the World Health Organization recommends that any woman considering pregnancy or pregnant should receive folic acid (400 mcg folic acid daily) from the moment she tries to conceive until 12 weeks of gestation. For the management of Chiari II malformation, decompression is recommended to be done either urgently or planned accordingly to severity of symptoms. Finally, the main treatment goal of arthrogryposis is to optimize quality of life which is done as

early as possible through a triad of treatment goals: First, by rehabilitation which includes physiotherapy, manipulation of contractures, and later social and occupational rehabilitation. Second, individually tailored orthotic management whether for maintenance or correction of joint mobility, and for prevention of recurrent deformities. Third, a broad spectrum of surgical techniques for correction of musculoskeletal deformities which is typically found in congenital contractures [9].

Conclusions

An aggressive approach to the potentially symptomatic Chiari II malformation child is warranted since good outcomes can be expected when this course is taken. Lower extremity arthrogryposis with myelomeningocele and Chiari II malformation is a prenatal diagnosis that requires high clinical suspicion, early multidisciplinary intervention, and genetic counselling. Early surgical approach and close follow-up are recommended due to the high recurrence rate of foot deformities, especially club foot deformities. As long as new approaches are being explored in the management of such cases, babies born now with neural tube defects can expect a better quality of life.

Conflict of interest

None.

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