



Case Report

Myelomeningocele with Associated Anomalies – Case Report and Literature Review

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Abstract

Myelomeningocele is a common defect of the development of the neural tube. It is a complex congenital malformation of the central nervous system (CNS) that can be associated with other concurrent anomalies. We report on a case of lumbar myelomeningocele with concomitant CNS malformations we followed up over a period of 15 years. A concise literature review has also been performed. The current report illustrates that the myelomeningocele is a complex anomaly that is commonly associated with a variety of other CNS malformations such as hydrocephalus and Chiari malformation. It may follow chronic progressive course with exacerbation of clinical symptoms in the long term. Patients that have undergone surgical correction of this spinal defect should be closely monitored over a long period of time because of the possibility of clinical deterioration of the concomitant anomalies such as hydrocephalus, Chiari malformation and siringomyelia.

Key words:

myelomeningocele, Chiari malformation, hydrocephalus, neural tube defect, CNS anomaly

INTRODUCTION

Myelomeningocele (MM) is one of the most common forms of spinal dysraphism characterized by bulging of the meninges and spinal cord through open vertebral arches as a result of misclosure of the neural tube during the last neurulation stage in the 3rd and 4th week of gestation.¹ Apart from genetic predisposition, some exogenous factors can also contribute to the development of MM such as concomitant maternal diseases, low socioeconomic status, folic acid deficit, etc.^{1,2} Despite prophylactic use of folic acid before and during pregnancy, incidence of MM remains high, ranging between 0.1 and 10 per 1,000 live births.¹ This anomaly can affect the entire spinal axis, but the most commonly affected area remains the lumbosacral region followed by the thoracic spine.³ The most common malformations that can be associated with MM are hydrocephalus and Chiari malformation type II.⁴

CASE REPORT

We report on a case of a 16-year-old male patient who was born of a second normal pregnancy of a 33-year-old mother who did not use folic acid prophylactically. The boy was born with soft tumor-like formation located in the lumbosacral region measuring 8 cm by 9 cm. At birth, the child preserved leg movements and only mild urinary and bowel retention. A computed tomography scan of the head and spine demonstrated MM in the lumbosacral region without associated malformations. At 2 months, he was operated on and the sac of the MM was partially excised after the placode had been freed from surrounding adhesions. In the following years the boy had normal psychological development. He started to experience speech stuttering. He experienced some difficulty in walking due to weakness in the legs, predominantly on the right and mild urinary and bowel retention since the age of 1.5 years. His right calf became hypotrophic. The electromy-

ography demonstrated anterior damage to L₄-S₂ nerve roots on the right. Upon his second hospital admission at the age of 16, he had complaints of weakness in the legs, predominantly in the right distal part of the leg, hypesthesia in the area of his right L₄-S₁ dermatomes and diminished dorsiflexion of the right foot. The magnetic resonance imaging (MRI) of the craniovertebral junction and lumbosacral region showed presence of Chiari malformation type II (CMII), syringomyelia at C₆-T₁ spinal level, tethered spinal cord with conus medullaris ending at L₄-L₅ spinal level, lumbosacral lipomatosis located in the anterior epidural space, dislocating posteriorly the low lying medulla (Fig. 1).

A second surgical repair of the lumbosacral MM was performed. After circumferential dissection of the sac to the level of the opened lamina of L₄ to S₂ vertebra, we performed microsurgical liberation and relocation of the neural elements inside the meningeal sac. We performed maximal safe resection of the epidural lipomatosis. The spinal cord was found to be divided which was consistent with split cord syndrome type II (Fig. 2). Eventually, a dural repair was performed. Histological examination of the epidural formation demonstrated lipid and fibrous tissue with congested blood vessels.

The patient had transient inferior paraparesis following surgery that gradually resided. Due to persistent headache,

we performed MRI of the head which demonstrated hydrocephalus consistent with colpocephaly that necessitated placement of ventriculoperitoneal shunt (Fig. 3).

DISCUSSION

Associated malformations of MM include anomalous development of the skull, supratentorial and infratentorial parts of the brain.⁴ CMII is a complex congenital anomaly that involves the midbrain and hindbrain (i.e. pons, medulla, and the cerebellum) and cervical spinal cord which occurs universally and exclusively in spina bifida with MM (Figs 1A, 1B).⁴ Additional features of spina bifida with MM can include tectal beaking, hydrocephalus, and corpus callosum hypoplasia and dysgenesis, enlarged massa intermedia, anomalous gyrification of the neocortex, but these features are variable.⁵ Associated anomalies of the spine and spinal cord and other organs and systems are rarely observed.⁶

There are several theories that discuss the formation of CMII but none of them thoroughly explains all associated anomalies.⁴ The theory of hindbrain dysgenesis as a result of MM clarifies the alterations in the posterior cranial fossa but not those affecting the supratentorial space.⁴ Accord-



Figure 1. MRI of the craniovertebral junction and lumbosacral region: A) Sagittal T1-weighted image of craniovertebral junction showing low lying brainstem (black and white arrow) and herniation of the cerebellar tonsils in the great occipital foramen (orange arrow); B) Sagittal T2-weighted image demonstrating syrinx at C6-T1 spinal levels (black and white arrow); C) Sagittal T2-weighted image – tethered spinal cord ending at L4-L5 spinal level (white arrow) and presence of epidural lipomatosis D) Sagittal T1-weighted image – cystic formation compressing filum terminale (white arrow), epidural lipomatosis dislocating posteriorly the spinal cord (black and white arrows).



Figure 2. Intraoperative image: the two halves of the split and tethered spinal cord (black and white arrow); dura mater (white arrow); nerve roots of cauda equina (arrow head).

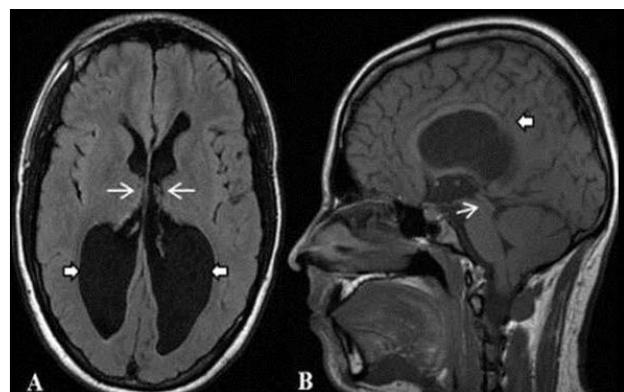


Figure 3. MRI of the head: A) Axial T1-weighted image showed colpocephaly (black and white arrows) and enlarged massa intermedia (white arrows); B) Sagittal T1-weighted image demonstrated corpus callosum hypoplasia in the region of the splenium (black and white arrow), aqueductal stenosis (white arrow), and polygyria.

ing to the hydrodynamic theory, proposed by Chiari himself, the caudal herniation of hindbrain is caused by the increased intracranial pressure as a result of hydrocephalus. The traction theory explains the development of CMII with the fixation of the spinal cord that causes hindbrain descensus. Recently, the theory put forward by McLone and Knepper became widely accepted.⁷ It states that there is CSF leakage through the spinal defect to the amniotic fluid which impedes the normal embryonic development of the ventricular system. The latter halts the skull development in the region of the posterior cranial fossa and reduces its size.⁷ Thus, the growing cerebellum reduces its volume and undergoes traction and herniation along the vertical and anteroposterior axis.⁸

The hydrocephalus is considered as inevitable complication of the opened neural tube defects. However, some 10-20% of the pediatric population with MM and CMII never develop hydrocephalus that requires shunting.¹ According to a Swedish study, the incidence of MM associated with hydrocephalus decreases from 0.33/1000 live births in the period 1989-1998 to 0.18/1000 live births in the period 1999-2002, mainly as a result of improved prenatal diagnostics and interruption of pregnancy for medical reasons.⁹ Hydrocephalus may be absent at birth but develop later in life (**Fig. 2**).¹⁰ Some patients with MM without hydrocephalus at birth may develop ventricular enlargement as a result of surgical repair of MM, as presented in our case.¹ In other cases, the prenatal ventriculomegaly does not progress over time despite the presence of elevated intracranial pressure.¹⁰ In such cases the problem revolves around the so called “contained hydrocephalus” that is a matter of debate due to the possibility of developing late fatal complications related to non-treated patients and risks associated with shunting.¹¹ The avoidance of shunt placement may have devastating effect on the development of such patients.¹⁰ Based on this belief, we decided to shunt our patient.

Corpus callosum (CC) dysgenesis and hypoplasia is observed in 70-90% of the cases with MM. It may affect the entire or different parts of the CC: rostrum, genu, body and splenium (**Fig. 2B**).¹² Merely 4% of cases with MM may have intact development of CC.¹² The substantial percentage of dimorphological development of the posterior part of CC in MM determines the common cognitive deficits that are caused by the impaired interhemispheric processing and transfer of speech perception.¹³ CC anomalies result in reduction of interhemispheric transfer and can cause stuttering.¹³ Another study suggest that stuttering patients have atypical distribution of gray and white brain matter and asymmetry between the prefrontal and occipital brain regions.¹⁴ The MRI performed in our patient confirms these two conditions that can predispose to stuttering.

Colpocephaly is a congenital form of particular ventricular configuration associated with substantial dilatation of the occipital horns of the lateral ventricles (**Fig. 3A**).¹⁵ Relative ventricular enlargement is observed in fetuses prior to fifth embryonic month that gradually decreases as a

result of migration of glial cells and growth of surrounding white matter and CC.¹⁵ Each perinatal influence on this process can preserve this ventricular configuration which is exclusively connected with complete or partial CC agenesis.¹⁶ Generally, colpocephaly is diagnosed in the prenatal period or early childhood. It may be clinically manifested with intellectual decline, seizures and other anomalies such as MM.¹⁵ Colpocephaly with CC agenesis may remain asymptomatic in adulthood.¹⁷ Our case confirms that colpocephaly is commonly associated with MM and may remain asymptomatic, a fact also shared by other authors.^{16,17}

Syringomyelia is a chronic spinal cord defect in which a tubular cavity, or central cavitation, is present in several spinal cord segments and almost 90% of syringomyelias are associated with Chiari malformation (**Fig. 1B**).¹⁸ It may remain either asymptomatic or cause substantial neurological deficit. The disease may have slow progression. One of the typical symptoms is persistent cervicobrachialgia and particular sensory deficit (pain and temperature anesthesia with preserved superficial and deep sensation), muscle atrophy in distal upper extremities. Motor weakness may develop late in the evolution of the disease. In such cases surgery may be indicated.

Although the neocortex is histologically preserved, the brain cortex is altered in cases with CMII (**Fig. 2B**). The gyri are smaller and multiplied, so called ‘polygyria’, that should be differed from ‘polymicrogyria’ in which the neocortex has only four cellular layers and is not typical of CMII.⁴

Diastematomyelia (DM) is a rare malformation that is characterized by longitudinal splitting of the spinal cord. There are two types of DM. In DM type I, there is extradural intraspinal bony bar between the two halves of the split spinal cord which are wrapped with separate meninges. In DM type II, the cord is divided by fibrous septum and is covered by common dural sac. DM results from impaired development of the notochord in the early stages of gestation and is rarely associated with MM. According to one study, MM is associated with DM in 32% of the cases (4 pts with DMI and 3 pts with DMII).³ Other authors stated that in cases with MM and DM, the splitting of the spinal cord is at the level of the first anomaly which was confirmed by our case.¹⁹

CONCLUSION

The prevention of neural tube defect requires perinatal prophylactic intake of folic acid, even in low risk pregnancies. MM is a complex anomaly commonly associated with a variety of other CNS malformations. The disease may follow progressive course with exacerbation of clinical symptoms in the long term. Patients should be followed up after surgical repair of MM because of the possible clinical decompensation of the associated anomalies such as hydrocephalus, CMII and syringomyelia.

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Миеломенингоцеле с ассоциированными аномалиями – клинический случай и обзор литературы

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Миеломенингоцеле является распространённым дефектом развития нервной трубки. Это сложная врождённая мальформация центральной нервной системы (ЦНС), которая может быть связана с другими сопутствующими аномалиями. Мы сообщаем о случае менингоцеле поясничного отдела с сопутствующими мальформациями ЦНС, который прослеживался нами в течение 15 лет. Был также проведён подробный обзор литературы. Настоящий доклад показывает, что менингоцеле является сложной аномалией, которая часто связана с множеством других мальформаций ЦНС, таких как гидроцефалия и мальформация Арнольда-Киари (Chiari malformation). В этом случае может иметь место хроническое прогрессирующее течение с обострением клинических симптомов в долгосрочной перспективе. Пациенты, которые подверглись хирургической коррекции этого дефекта позвоночника, должны находиться под пристальным наблюдением в течение длительного периода времени из-за возможности клинического ухудшения сопутствующих аномалий, таких как гидроцефалия, мальформация Арнольда-Киари и сирингомиелия.