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Primary Repair of Open Neural Tube Defect in Adulthood: Case Example and Review of Management Strategies

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Abstract

Background Context: Neural tube defects are congenital malformations that develop when the neural plate fails to close during embryogenesis. The most common open neural tube defect, myelomeningocele (MMC), is declining in frequency in North America. If identified, an MMC must be closed in the perinatal period to prevent lethal complications. Lesions presenting in older adults are, thus, very uncommon.

Purpose: To describe the surgical management of an adult with an unrepaired ulcerated lumbosacral MMC presenting with persistent cerebrospinal fluid leakage and review the management strategies for adult patients with unrepaired MMC.

Study Design: Case report

Methods: The patient was a 62-year-old woman with an unrepaired ulcerated lumbosacral myelomeningocele and associated lower extremity weakness. She sought medical care for persistent lumbar tenderness and ulceration after sustaining a fall four months prior to admission. Physical and radiological assessment revealed a lumbosacral myelomeningocele at the L5/S1 level and a tethered cord. Surgical resection of the placode and de-tethering were performed.

Results: One week after repair, the patient was readmitted for management of continued cerebrospinal fluid leakage and hydrocephalus, requiring external ventricular drainage, wound revision, and placement of lumboperitoneal shunt. The patient experienced complete resolution of back pain without additional episodes of cerebrospinal fluid leakage.

Conclusions: This rare case and review of management strategies suggests that proper surgical

management of open MMC in adulthood can successfully be performed and improve patient symptoms and prevent further complications.

Key words: myelomeningocele; spinal dysraphism; tethered cord; lumboperitoneal shunt; hydrocephalus; cerebrospinal fluid (CSF)

Running Title: Primary repair of open neural tube defect in adulthood

Abbreviations: MMC, myelomeningocele; CSF, cerebrospinal fluid

Introduction

Neural tube defects are a family of congenital malformations that are thought to develop when the neural plate fails to close during embryogenesis [1]. Myelomeningocele (MMC), the most common type of open neural tube defect, is characterized by herniation of meningeal membranes and spinal cord tissue through a bony defect in the vertebral column, frequently in the lumbosacral region [2]. According to Centers for Disease Control and Prevention estimates, MMCs occur in 3 out of every 10,000 live births, accounting for the chronic disability of nearly 100,000 individuals in the United States (http://www.cdc.gov/ncbddd/spinabifida/data.html) [1].

As a result of increased prenatal screening, the vast majority of neural tube defects are diagnosed early in pregnancy and if pregnancy is carried to term, the defect is surgically repaired within 72 hours of birth. Although the benefits of prenatal and early postnatal repair are well established [3], primary MMC repair in adolescent and adult patients has been sporadically reported [4-6]. The risks of not treating MMC include progressive neurological dysfunction, chronic meningitis, and death [7]. Here, we report a unique case of a 62-year-old patient with open lumbosacral MMC undergoing primary surgical repair and review management strategies for similar published cases. This is one of a few cases of delayed MMC repair in an adult requiring surgical intervention for persistent cerebrospinal fluid (CSF) leakage following an injury.

Case Report

History and Examination

This 62-year-old woman from a rural area had a lifelong history of an open lumbosacral MMC that had been judged non-operable at the time of birth. Minimal CSF leakage from the

ulcerated lesion had begun 8 years prior to her presentation and had been managed with daily wound dressing and intermittent oral antibiotic treatment. The patient sought medical attention four months after suffering a fall that ~~precipitated~~ led to focal tenderness and copious CSF drainage; it was unclear whether pain was related to muscular strain or irritation from CSF leakage. At admission, the MMC measured 10 × 10 cm and was noted to be primarily epithelialized with small ulcerations and CSF leak in multiple areas (Figure 1).

Since birth, the patient's clinical condition had been characterized by lower extremity weakness, limited ambulation with use of a front-wheeled walker, as well as bowel and overflow incontinence due to urinary retention. On examination, the patient demonstrated reduced distal lower extremity strength bilaterally: dorsiflexion (3/5), plantarflexion (3/5), and extensor hallucis longus (3/5). Sensation of the ankles and feet was diminished, with normal reflexes and no hyperreflexia. Her general health was otherwise unremarkable and unchanged after her fall. She had no previous history or treatment of severe meningitis requiring hospitalization, and during the last 8 years she had experienced minimal CSF leakage.

Radiographic studies revealed a large complex MMC with associated tethered spinal cord and posterior defect involving nearly the entire sacrum (Figure 2). Computed tomography (CT) imaging demonstrated a Chiari II malformation but no ventriculomegaly.

Initial operative intervention

Because of the persistent CSF leakage after her fall, surgery was pursued. Surgical treatment consisted of primary closure of the MMC and debulking of the surrounding dysplastic scar tissue. The subperiosteal dissection was carried down to the level of the lamina at L5, identifying the normal anatomy surrounding the defect. The epithelial tissue planes were

carefully dissected free from the scar tissue and adjoining placode (Figure 3A,B). The subdural space was dissected and the tethered structures were released. The dura was inadequate to fully cover the placode, therefore a 4 × 6-cm AlloDerm patch was used to achieve a watertight closure. Figure 4 illustrates the layers of closure of a MMC repair and neural placode.

Postoperative course and secondary operative intervention

Postoperatively, the patient's neurological condition was unchanged, but she developed leakage of CSF from her wound on postoperative day 5 as well as increased ventricular size on cranial imaging. On postoperative day eight, an external ventricular drain was inserted, but subsequent attempts to wean the drain were unsuccessful. Thirteen days after the initial repair, the patient underwent a second surgical procedure for wound revision and placement of lumbar drain because of wound dehiscence and continued CSF leakage. Prior to the patient's discharge, the drain was converted to a slit valve lumboperitoneal shunt system.

After shunt placement, the patient experienced complete resolution of back pain without additional episodes of CSF leakage. She complained of recurrent occipital headaches that were responsive to over-the-counter medications. Eleven months after surgery, her Oswestry Disability Index score was 26%, and magnetic resonance imaging demonstrated an intact repair without evidence of pseudomeningocele (Figure 5). There was no change in lower extremity neurological function, and she was free of headaches with a well-healed wound 2 years after surgery.

Discussion

MMC is the most common type of open spinal dysraphism, with an incidence of

approximately 1/1000 to 1/3000 [2,8,9]. Declining rates are thought to be due to dietary supplementation with folic acid and improved prenatal screening. Open neural tube defects are thought to result from the sequential disruption in the maturation process of neuroectodermal and mesodermal structures early in the gestational process, between the 22^{nd} and 26^{th} days of embryogenesis [2]. MMCs are characterized by the cystic protrusion of the meninges and spinal cord tissue through a midline defect of the spinal column. These lesions can result in neurological injury at the root or cord level, depending on the level of the defect. Treatment is typically aimed at primary closure of the defect while preserving any viable neural structures.

Traditionally, MMC closure is performed within the first 48 hours after delivery. Delay beyond 72 hours is associated with higher risk of meningitis, ventriculitis, and development of hydrocephalus-related complications. Recent observational and prospective studies highlight the benefits of prenatal repair. Burrows et al. [3] reported a randomized trial of prenatal MMC repair compared with postnatal repair in 183 patients. The trial was terminated after interim analysis demonstrated improved neurological outcomes and less shunt dependence in the prenatal cohort, despite increased risk of premature birth and maternal morbidity.

We report a case of primary open MMC repair in an adult patient with persistent CSF leakage and chronic neurological dysfunction. To date, very few cases of open MMC closure in adulthood have been reported (Table 1) [4-6,10]. Akay et al. [4] performed standard repair on 5 adolescents (mean age 21) with open MMC. Notably, all 5 patients in the series were neurologically intact and underwent surgery primarily for pain relief of low back pain and improvement of aesthetic appearance. Denaro et al. [5] resected an open cervical MMC presenting with new onset weakness and paresthesias in a previously asymptomatic 52-year-old man. In a case similar to the current case, Alberio and colleagues [6] described open MMC repair

in an adult paraplegic patient with split cord malformation and persistent CSF leakage. Brokinkel et al. [10] described a ventral cervical MMC in a 26-year-old-woman with Klippel-Feil Syndrome with progressive paresthesias and weakness in her upper extremities. Remarkably, the majority of patients had been previously diagnosed with MMC but had not been surgically treated. Both the case reported by Alberio et al. [6] and our own raise the question of why a CSF leak would occur in a delayed fashion and why it was not associated with infection earlier in life. It is possible that the delayed presentation resulted from gradual erosion from an underlying dural defect through the skin, and perhaps this inflammatory milieu prevented bacterial migration. While the reasons for the delay in surgery in this patient population are unknown, they may relate to socioeconomic status, availability of healthcare or, lack of neurological dysfunction. In this case the patient did not seek medical attention due to lack of significant, progressive dysfunction and the belief that her lesion was "inoperable."

Our review of the literature suggests a similar risk profile in adult and neonatal MMC closure. Early complications of neonatal MMC repair include neurological worsening (9.5%), hydrocephalus (80–90%), CSF leakage (18%), and associated wound-healing problems [11,12]. Of the nine cases of adult MMC repair we identified in the literature, patients experienced neurological deficit (1/9), hydrocephalus requiring additional surgery (2/9), and CSF leakage (1/9) (Table 1) [4-6]. Akay et al. [4] reported a low incidence of complications, with only one case of bladder incontinence in a previously neurologically intact patient that improved spontaneously after 3 months without additional revision surgery. Two cases with significant preoperative neurological deficits developed hydrocephalus within 2–8 days of repair, with one requiring an external ventricular drain and the other a ventriculoperitoneal shunt [6]. The present case demonstrated concurrent CSF leakage and wound dehiscence despite meticulous dural

closure. The patient required repeat dural closure and extended hospital stay with aggressive CSF drainage to allow for wound healing [11]. As in neonatal cases, the CSF leak was likely the result of increased CSF pressure that occurs 4–8 days after repair once the chronic CSF fistula was closed [11].

Although the etiology of hydrocephalus and increased CSF pressure following MMC repair is not clearly delineated, a multifactorial origin is likely. Both non-communicating hydrocephalus, due to aqueductal stenosis and fourth ventricle obstruction related to the Chiari II malformation, and communicating hydrocephalus resulting from underdevelopment of arachnoid granulations as contributory mechanisms have been proposed as potential causes [11,13]. Despite an uncertain causality in neonates, in the current case the patient did not demonstrate hydrocephalus preoperatively, yet demonstrated ventricular enlargement 8 days after closure of the chronic CSF leak, consistent with communicating hydrocephalus (Figure 6) [11,14].

Strikingly, the majority of patients undergoing adult repair demonstrated resolved or improved symptoms after surgery (Table 1). In Akay et al. [4], surgical indications included pain and aesthetic concerns, with 75% of patients demonstrating decreased pain postoperatively. Denaro et al. [5] reported resolution of preoperative weakness and paresthesias within 6 months after surgical repair of a cervical MMC. As can be expected, neither patient with lifelong neurological dysfunction demonstrated dramatic improvement in neurological status; however, Alberio et al. [6] described improved lower extremity tone and bowel control.

Conclusions

We report an extremely rare case of a lumbosacral MMC repaired in late adulthood. Although the patient had been diagnosed at birth, the malformation remained unrepaired and was

characterized by lower extremity weakness and continuous CSF drainage. Despite a complicated postoperative course and revision surgery, the patient reported an improved quality of life following surgery. Our case and review of the literature suggest that proper surgical management of open MMC in adulthood can improve patient's quality of life.

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Figure 1. Photograph demonstrating MMC measuring 10 × 10 cm. There were several areas noted to be ulcerating, with CSF leaking from multiple areas along the defect.

Figure 2. Axial (A) and sagittal (B) T1-weighted MR images of large complex MMC shows neural placode extending beyond skin surface because of expansion of underlying subarachnoid space. (C) Sagittal T2-weighted MR image at midline. (D) Sagittal CT image.

Figure 3. (Upper) Photograph following excision of the surrounding dysplastic scar tissue and primary closure of the MMC. (Lower) The epithelial tissue planes were carefully dissected free from the scar tissue and adjoining placode and subsequently closed primarily.

Figure 4. Illustration outlining the layers of a MMC repair and neural placode.

Figure 5. Postoperative axial (A) and sagittal (B) T1-weighted and axial (C) and sagittal (D) T2-weighted MR imaging at 11-month follow-up.

Figure 6. Axial non-contrast head CT obtained preoperatively (A) and one obtained 8 days following repair and closure (B) demonstrating dilation of the lateral ventricles consistent with communicating hydrocephalus.

Table 1: Primary myelomeningocele repair in adulthood

Author (year)	No. of patients	Patient age (years)	Location	Primary presenting symptom	Neurological deficits	Complications

Akay et al. 2003[2]	5	Mean 21 (20–23)	Lumbosacral	Pain (4)	Normal (5)	Urinary incontinence (1)
Alberio et al. 2010[3]	1	47	Lumbosacral	CSF leakage	Paraplegia, anesthesia below T12	Hydrocephalus*
Denaro et al. 2008[6]	1	53	Cervical	Neurological	Weakness, paresthesias	None
Brokinkel et al. 2013[5]	1	26	Cervical	Paresthesias	Weakness, paresthesias	None
Present study	1	62	Lumbosacral	CSF leakage, pain	Bilateral LE weakness	CSF leak, hydrocephalus*

1 CSF, cerebrospinal fluid; LE, lower extremity

2 *Patient required additional unplanned surgery

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