CASE REPORT

Phnx Med J. November 2023, Volume 5 No 3
DOI:10.38175/phnx.1292845

A Newborn Spina Bifida Case Report

Yenidoğan Spina Bifida Vakası

Kamuran Suman[1] Serhat Korkmaz[2] Murat Suman[3]

1 Afyonkarahisar State Hospital, Clinic of Perinatology, Afyonkarahisar, Türkiye.
2 Afyonkarahisar Health Sciences University, Medical Faculty, Department of Neursurgurey, Afyonkarahisar, Türkiye.
3 Afyon Çay State Hospital, Clinic of Pediatric, Çay, Afyonkarahisar, Türkiye.

ABSTRACT

In North America, over seventy thousand cases of disability are attributed to central nervous system anomalies, with the majority of these cases resulting from neural tube defects. Among these defects, meningomyelocele is the most commonly observed condition. Meningomyelocele is characterized by the protrusion of spinal structures outside the skin, leading to deficits in motor function of the lower limbs and challenges related to urinary and fecal elimination. The severity of these clinical symptoms can vary depending on the location of the defect along the neural tube.

The root cause of meningomyelocele is associated with the incomplete closure of the neural tube during fetal development. In our specific case, the diagnosis of spina bifida was made prenatally by the perinatology clinic, and the family received comprehensive information about the condition well before the delivery. Despite being informed, the family made the decision to proceed with the birth, and necessary arrangements were made to support this choice. Following the delivery, the newborn underwent an examination confirming the presence of spina bifida, in line with the previous ultrasound diagnosis.

ÖZET

Kuzey Amerika'da, merkezi sinir sistemi anomaları 70 binden fazla engelli vaka ile ilişkilendirilmektedir ve bunların büyük bir kısmı nöral tüp defektlerinden kaynaklanır. Bu defektler arasında en yaygın görüleni ise meningomyelosel olarak bilinir. Meningomyelosel, omurilik yapılarının deri dışına çıkmasıyla karakterizedir ve bu durum genellikle alt ekstremitelerin motor fonksiyonlarında eksikliklere ve idrar ile dışkı boşaltımıyla ilgili sorunlara yol açar. Hastalığın şiddeti, defektin nöral tüp boyunca yerleşim yerine bağlı olarak değişebilir. Meningomyeloselin temel nedeni, fetal gelişim sırasında nöral tüpün düzgün bir şekilde kapanmamasıyla ilişkilidir. Bu tür bir durumun oluşumu, prenatal dönemde perinatoloji kliniği tarafından tanımlanabilir ve aileye durumla ilgili gerekli bilgiler doğum öncesinde sağlanabilir. Ancak, bazı aileler bu zorlu süreçte doğum yapma kararı alabilirler.

Bahsettiğiniz durumda, aileye doğum sonrası yaşanabilecek zorluklar konusunda bilgilendirme yapılmış olmasına rağmen, onlar doğum yapma kararı almıştır. Bu zorlu süreci kolaylaştırmak amacıyla uygun düzenlemeler yapılmış ve doğum gerçekleşmiştir. Bebek, doğumdan hemen sonra yapılan bir muayeneyle ultrason ile teşhis edilen spina bifida durumunu doğrulamıştır.

Bu tür durumlarda, ailenin kararı ve doğum sonrası sürecin yönetimi büyük önem taşır. Sağlık profesyonelleri, ailenin ihtiyaçlarına uygun destek ve rehberlik sağlayarak, bebek ve ailenin yaşamlarını en iyi şekilde yönlendirmeye çalışırlar. Bu tür çabalar, hastalıkla yaşamak zorunda olan çocuğun ve ailenin yaşam kalitesini artırmada kritik bir rol oynar.

Keywords:
Spinal dysraphism
Meningomyelocele
Prenatal diagnosis
Folic acid

Anahtar Kelimeler:
Spinal disrafizm
Meningomyelosel
Prenatal tanı
Folik asit

INTRODUCTION

Spina bifida is a congenital disorder with a variable clinical presentation, ranging from subtle to severe, depending on the location and size of the neural tube defect (1). Symptoms commonly include lower limb muscle weakness, mobility issues, and urological problems, such as urinary retention and the need for catheterization. The level of the lesion is typically related to the severity of the symptoms; however, this relationship is not absolute (2). The underlying cause of central nervous system dysfunction is abnormal development of the neural tube, which can also result in Chiari type 2 malformation, characterized by a hypoplastic cerebellum and herniation of the lower brainstem into the cervical canal (3). Additionally, obstructive hydrocephalus is a common complication of myelomeningocele, resulting from defects in cerebrospinal fluid absorption. Spina bifida is a multifactorial condition with folic acid deficiency being a well-established risk factor. Prenatal diagnosis of neural tube defects is usually performed through amniocentesis, which is typically combined with fetal ultrasonography (3). Elevated levels of alpha-fetoprotein in amniotic fluid are suggestive of a neural tube defect, with abnormal levels at 13-15 weeks of gestation being particularly indicative of spina bifida (4). A definitive diagnosis of spina bifida can be made via amniocentesis between 15th and 18th weeks

Correspondence: Murat Suman, Afyon Çay State Hospital, Clinic of Pediatric, Çay, Afyonkarahisar, Türkiye.
E-mail: muratsuman@hotmail.com
Cite as: Suman K, Korkmaz S, Suman M. A Newborn Spina Bifida Case Report. Phnx Med J. 2023;5(3):230-232.

Received: 05.05.2023 **Accepted:** 31.07.2023 **Online Published:** 24.10.2023

of gestation. Diagnostic imaging, such as X-rays, MRI, or CT scans, can be used to confirm the diagnosis of spina bifida and assess the severity of the defect (5). This report presents a case of myelomeningocele in a neonate with severe spina bifida, which was diagnosed prenatally.

CASE REPORT

The neonate's mother is a 32-year-old woman with an unremarkable medical history and no known genetic disorders. Prior to this pregnancy, she had three successful pregnancies with no reported complications or adverse outcomes. The parents are not consanguineous. The neonate was born with a weight of 2.76 kg to a mother who experienced pre-eclampsia and presented with respiratory distress and lower back pain. Upon examination, a visible aperture in the lower back was observed, prompting admission to the neonatal intensive care unit due to respiratory distress. At 16 weeks of gestation, routine antenatal visits revealed dilated lateral ventricles, obliterated cisterna magna, and an approximate 60-degree angulation of the lumbosacral spinal cord in the neonate (Figure 1).

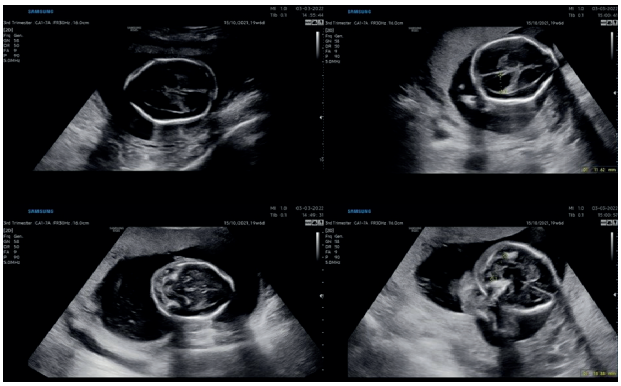

Figure 1: Lateral ventricle measures at 19w6d.

Upon examination, the newborn was found to have an open spina bifida with a meningocele sac containing approximately 8 vertebrae that were not visible. The family was promptly informed of the situation, and the available treatment options were discussed with them. Historically, most medical centers treated all viable newborns with aggressive surgical intervention without giving the family an option to choose. However, at our hospital, treatment decisions are based on the parents' preferences after careful counseling regarding treatment options and expected outcomes, including the possibility of terminating the pregnancy, performing additional prenatal testing, choosing a delivery setting, and, if necessary, fetal surgery. Postnatal treatment options, including surgical closure of the defect and the potential need for ventriculoperitoneal shunting, are also discussed. In our case, the family chose not to terminate the pregnancy, and the defect was carefully assessed immediately after birth. The size, location, and level of the defect were noted, and if a sac was present over the skin, its integrity and potential for cerebrospinal fluid (CSF) leakage were carefully evaluated. To avoid the risk of infection and heat loss, the defect was covered with plastic sheets and a sterile saline-soaked dressing until the neurosurgeon arrived. The prone position was recommended for resting the newborn, and

Figure 2: Four months after surgery.

all systemic examinations were performed to evaluate any potential neurological or other systemic issues. During the surgery preparations, vomiting and an increase in head circumference were noted, highlighting the importance of careful monitoring of neurological findings from the beginning of treatment. Infections are also a significant concern for patients with large defects and are often prevented by using broad-spectrum antibiotics. To reduce the risk of CNS infections, the surgical procedure should be completed as soon as possible, and the total process should not be postponed for more than three days. The surgical technique used to close the defect involves an approach from the lateral margin of the neural plate aperture, which forms a proper neural tube and provides optimal closure. A tethered spinal cord is a risk during this type of surgery, but it can be detached during the subsequent surgical procedure. In our case, the ventricular enlargement observed during the fetal period was also present, and a ventriculoperitoneal shunt was successfully placed. During surgery, the defect was closed, and the wound was managed with routine care. It is crucial to continue careful monitoring of neurological findings and infection risks to ensure the best possible outcomes for the patient (Figure 2).

DISCUSSION

The majority of congenital central nervous system (CNS) diseases can be attributed to a failure in neural tube closure during early weeks of gestation (6). This results in a variety of neural tube defects that can affect different

CNS structures, resulting in a diverse range of clinical presentations based on the specific area involved. Cultural factors can also impact the prevalence of these defects, as families may have varying attitudes toward prenatal counseling and different approaches to management. A recent review comparing the incidence of these defects among different cultures found significant differences in prevalence (Table 1).

Table 1: Incidences of neural tube defects around the world.

Region	Number of the Family affected (n/10.000)
Mediterranean	21.9
Southeast Asia	15.8
Africa	11.7
Americas	11.5
Europe	9.0
Western Pacific	6.9

Note: most recent data available (2016).

The latest available data indicates that the United States of America has an incidence of 6.5 cases per 10,000 live births of congenital central nervous system (CNS) diseases, which are attributed to the inability of the neural tube to close during the early weeks of conception, leading to a defect in the closure of most CNS structures (7). The presentation of clinical symptoms varies depending on the affected CNS structure. The number of cases reported among families worldwide may vary due to cultural differences in prenatal counseling, resulting in a more diverse range of affected cases as reported in recent studies. The exact cause of the CNS defects is yet to be elucidated. However, the risk of recurrence following the index case is increased by approximately 4%. This risk increases to 10% if there are more than two cases. Inadequate consumption of folic acid during pregnancy plays a crucial role in the development of neural tube defects (8). Studies recommend that women of childbearing age who may become pregnant take 0.4 mg of folic acid daily, while those at high risk should take a tenfold dosage of up to 4 mg. Infants with myelomeningocele often present with more severe deficits as they extend into upper thoracic levels (8). Chiari type II malformation, a common complication of myelomeningocele, typically results in hydrocephalus. The emergence of hydrocephalus may vary from slow to rapid, depending on the size of the defect. Symptoms of Chiari type II malformation typically affect the hindbrain, resulting in feeding difficulties, stridor, apnea, and vocal cord paralysis, which may be fatal if left untreated. Treatment for children with myelomeningocele requires a multidisciplinary approach involving surgeons, therapists, and pediatricians as coordinators (9). Most cases involve the genitourinary system, and parents must learn to regularly catheterize a neurogenic bladder to minimize urinary tract infections and prevent further damage. Regular assessment of renal function is essential through urine cultures, serum electrolytes, renal scans, vesiculourethrograms (VCUG), renal ultrasonography, and cystometrograms. Urinary tract assessment can reduce the need for urologic detour procedures. Mortality rates of untreated CNS defects rise to 90%-100%, while untreated myelomeningocele has a chance of only 28% of living to the age of 7. Episodes of meningitis can impair intellectual and cognitive functions, and renal dysfunction is a significant determinant of mortality (10). While only a few centers offer surgical closure of a spinal lesion in utero, parents are faced with the choice of preparing for the delivery of an affected infant, considering in utero intervention, or terminating the pregnancy after the sonographic diagnosis of NTD. Additional fetal testing to identify associated anatomic or chromosomal abnormalities, referral to a spina bifida clinic to discuss possible fetal interventions, postnatal management, and prognosis, and outreach to other parents of affected children can help them make this decision.

Conflict of Interest: No conflict of interest was declared by the authors.
Ethics: The patient informed consent form was obtained.
Funding: There is no financial support of any person or institution in this research.
Approval of final manuscript: All authors.

REFERENCES

1. Iskandar BJ, Finnell RH. Spina bifida. New England Journal of Medicine. 2022;387(5):444-450.
2. Ho P, Quigley MA, Tatwavedi D, Britto C, Kurinczuk JJ. Neonatal and infant mortality associated with spina bifida: A systematic review and meta-analysis. PloS one. 2021;16(5):e0250098.
3. Ushakov F, Sacco A, Andreeva E, et al. Crash sign: new first-trimester sonographic marker of spina bifida. Ultrasound in Obstetrics & Gynecology. 2019;54(6):740-745.
4. Kancherla V, Wagh K, Pachón H, Oakley Jr GP. A 2019 global update on folic acid-preventable spina bifida and anencephaly. Birth Defects Research. 2021;113(1):77-89.
5. Jain L. Improving the Precision of Neonatal Neuroimaging. Clinics in Perinatology. 2022;49(3):xv-xvii.
6. Kancherla V, Mowla S, Räisänen S, Gissler M. Early Neonatal Mortality among Babies Born with Spina Bifida in Finland (2000–2014). American Journal of Perinatology. 2021; 83(1):41-49.
7. Waitzman NJ, Jalali A, Grosse SD. Preterm birth lifetime costs in the United States in 2016: An update. Elsevier; 2021:151390.
8. Shlobin NA, Yerkes EB, Swaroop VT, Lam S, McLone DG, Bowman RM. Multidisciplinary spina bifida clinic: the Chicago experience. Child's Nervous System. 2022;38(9):1675-1681.
9. Wolfe ID, Lillegard JB, Carter BS. Parental request for non-resuscitation in fetal myelomeningocele repair: an analysis of the novel ethical tensions in fetal intervention. Journal of Perinatology. 2022;42(7):856-859.
10. Gajagowni S, Altes T, Vachharajani AJ. Diagnostic utility of spinal ultrasounds in neonates. American Journal of Perinatology. 2023;110(1):71-79.