

Case Report



Clinical Manifestation and Initial Management of Newborn with Spina Bifida Aperta: A case report

Indra Sandinirwan^{1*}, Wargian Hadisaputra²

¹Department of Pediatrics, Hermina Hospital Sukabumi, Sukabumi, West Java, Indonesia

²Department of Neurosurgery, Hermina Hospital Sukabumi, Sukabumi, West Java, Indonesia

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Abstract

Spina bifida is a congenital disorder that occurs due to the disruption of the neural tube formation, with an incidence of 1–10 cases per 1000 live births. Spina bifida could appear as an occulta or aperta type; the latter consisting of meningocele, myelomeningocele, and myeloschisis. We reported a case of a term male newborn who was presented with lethargy, bradypnea, suction reflex abnormalities, and paralysis of lower extremities. A defect was found in the thoracolumbar region of the spine, showed a placode with no meningeal and skin covering, suggestive of a myeloschisis. The patient was treated in the neonatal intensive care unit (NICU), placed on NIV with successful weaning, parenteral nutrition, systemic antibiotics regimen, and wound care. The patient was consulted with a neurosurgeon and was subjected to an ultrasound examination of the head, which resulted in hydrocephalus. After clinical improvement and gaining the oral tolerance, the patient was discharged and scheduled for surgery.

Introduction

Caring for newborns is the routine of pediatricians and physicians. Although it is not frequent, sometimes neonates are born with congenital anomalies. Spina bifida is one of the common congenital anomalies. Considering the recent advancements, nowadays, the newborns with spina bifida have a greater chance to survive. Many aspects are of concern in terms of clinical aspects and complications, as well as interdisciplinary cooperation, especially neurosurgery.

Neural tube defects (NTDs) are serious congenital disorders that arise at the time of embryogenesis and are the result of the failure in the neural tube closure. This congenital NTD has an incidence of 0.5-10 in 1000 live births.^{1,2} The clinical phenotype of the defects depends on the point at which the embryonic neural tube closure has failed. Failure of the neural tube closure in the forebrain area results in anencephaly, while the failure in the spinal area results in spina bifida. Craniorachischisis occurs when a failure occurs at both points. Among all of the congenital abnormalities mentioned, spina bifida is the most common type of NTDs with a higher chance of survival and compatible with postnatal life.^{1,3}

There are two forms of spina bifida, spina bifida occulta and spina bifida aperta. In the spina bifida occulta, the defect is still covered with skin, and the spinal cord is not exposed. In the spina bifida aperta, the defect is not

covered with skin, so the spinal cord is exposed with or without a herniated sac.^{4,5} Various subtypes of the clinical phenotype can occur, which are influenced by the pathophysiology at the embryogenesis phase. This case report focuses on the clinical manifestation and initial management of a newborn with spina bifida aperta in a private regional hospital with a neonatal intensive care facility. Informed consent is in the Supplementary File.

Case Report

A 1-day-old term male neonate was born spontaneously in our hospital delivery room, who had not cried immediately so he was given positive pressure ventilation. The 1 and 5 minutes Apgar scores were 5 and 7, respectively. Breathing appeared irregular and showed bradypnea with a rate of fewer than 30 breaths/minute, a heart rate of 150 beats/minute, temperature of 36.8°C, and a normal capillary refill time. The newborn was the second child of a mother with an obstetrician's routine antenatal follow-up. The previous history of antenatal ultrasound examination revealed dilation and excess fluid in both fetal ventricles.

In the physical examination, the birth weight was 2530 g, the body length was 45 cm, and the head circumference was 32 cm. There was a defect in the spine in the thoracolumbar region (T12–L2) that showed a placode with no meningeal and skin covering, suggestive of a myeloschisis. A wound in the size of 8 cm in 8 cm, with

*Corresponding Author: Indra Sandinirwan, Email: indra14006@mail.unpad.ac.id

a depth of approximately 1 cm was evident, as shown in Figure 1. The upper limbs still had the active motor movement, while the lower limbs experienced paralysis and the absence of tendon reflexes.

The newborn was then treated in the neonatal intensive care unit (NICU). Routine laboratory workup and radiologic examinations were carried out. Airway management with non-invasive ventilation (NIV), umbilical vein catheterization for parenteral nutrition, orogastric tube insertion, administration of systemic antibiotics, and aminophylline (8 mg/kg loading dose infusion over 30 minutes, and maintenance dose 1.5 mg/kg every 8 hours) were administered. Wound care and the dressing were done carefully. The patient was referred to a neurosurgeon, who advised a head ultrasound, and neurosurgery if the patient was stable.

The plain radiograph showed agenesis of the right fifth rib, and lumbarization of S1 vertebrae and hemivertebrae of S2, as shown in Figure 2. The results of the initial laboratory tests were as follows: Hemoglobin 17.2 g/dL, hematocrit 47.9%, leukocytes 18 900/mm³, platelets 263 000/mm³, differential count results within normal limits, blood glucose at 64 mg/dL, natrium 133 mmol/L, kalium 3.63 mmol/L, chloride 99.3 mmol/L. On the third day of treatment, the neonate's breathing pattern improved and bradypnea was no more present. The patients weaned the NIV and received continuous positive pressure ventilation. On the fourth day, the patient started trophic feeding through an orogastric tube. The neonate's diuresis was good even though urinary incontinence was noted. The neonate had abdominal distension and delayed defecation, so he was given rectal laxatives until he could pass the stool. No vomiting was observed.

On the fifth day of treatment, the neonate experienced an increase in body temperature to 38°C. Laboratory workup was obtained, resulted in a hemoglobin of 13.9 g/dL, hematocrit 38.8%, leukocytes 21 300/mm³, platelets 166 000/mm³, basophil 0, eosinophil 1, band 3, segment 71, monocytes 13, lymphocytes 12, suggesting a bacterial



Figure 1. A defect in the thoracolumbar region showed myeloschisis.



Figure 2. The plain radiograph showed agenesis of the right 5th rib, lumbarization of S1 vertebrae, and hemivertebrae of S2.

infection. We changed the antibiotic to meropenem (40 mg/kg, every 8 hours) instead of the previously use cefotaxime (50 mg/kg/dose, every 12 hours).

On the seventh to the ninth day of treatment, the neonate's condition gradually improved, jaundice appeared with an increase in the total bilirubin levels, marked 15.8 mg/dL, and phototherapy was performed. The orogastric tube was removed. The patient breathed easily without supplemental oxygen. The oral intake tolerance was also good while continuing the oromotor therapy. The head ultrasound showed hydrocephalus with dilated ventricles. On the tenth day of treatment, the umbilical vein catheter was removed. The neonate was able to feed orally, the motoric function of upper extremities was remarkably good, laboratory parameters were within normal limits. Counseling and education were carried out to the parents. On the following day, the neonate was allowed for discharge.

At the first month follow-up, the patient's head circumference increased considerably to 38 cm, with vomiting symptoms, and sunken eyes. A head CT scan of the patient was performed and showed dilatation of the ventricular system, as shown in Figure 3. The results of cardiopulmonary and laboratory assessments were within the normal limits, so that the patient underwent a ventriculoperitoneal shunt, with good results. With routine wound care, the myeloschisis defect gradually covered by skin and granulation tissue, as shown in Figure

4. The patient was scheduled for tethered cord release procedure and correction of spina bifida.

Discussion

In this case, there was a defect in the spine in the lumbar region, showing a reddish placode and suggestive of a spina bifida aperta, an 8 cm in 8 cm wound with a depth of approximately 1 cm. From the physical examination, we strongly suspected that the type of spina bifida in this newborn was spina bifida aperta (myeloschisis subtype) because there was no meninges and skin covering the wound. The upper extremities still had active motor movement, while the lower limbs experienced paralysis with the absence of tendon reflexes.

The term spina bifida refers to a fairly varied spinal

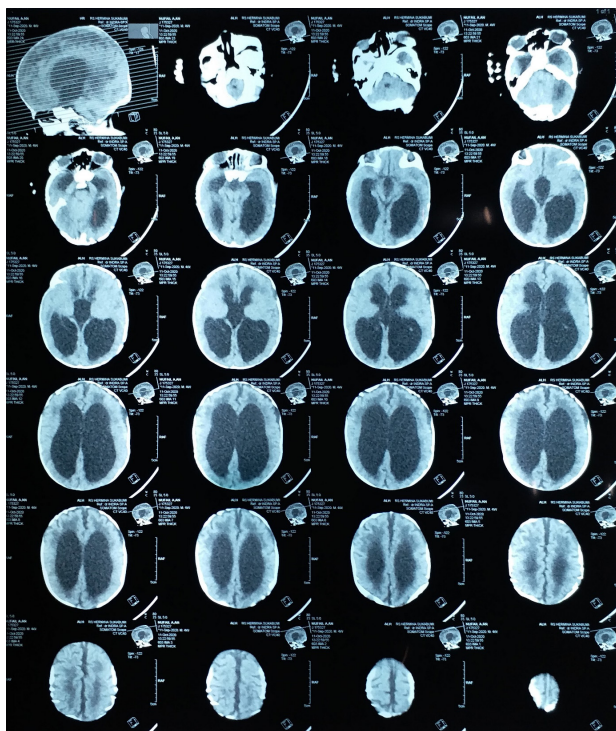


Figure 3. A head CT scan showed dilatation of the ventricular system.



Figure 4. The myeloschisis defect gradually covered by skin and granulation tissue.

abnormality with the absence of spinal protrusions and vertebral bone abnormalities. In general, spina bifida can be classified into two groups, spina bifida occulta, and spina bifida aperta.^{4,6} In spina bifida occulta, one vertebral arch is not formed, and there is still skin covering. On inspection, some neonates have indentations, dark spots, hairy lesions, or swelling on the defective area. In this condition, the spinal cord and nerves are not damaged.^{1,7}

On the contrary, spina bifida aperta is characterized by defect of the vertebral arch or protrusion of the meninges. If more than one vertebra is affected, the meninges will protrude, and if they are covered with thin skin, it is a meningocele. This protruding sac-like structure containing the meninges and the cerebrospinal fluid (CSF) may rupture. If the spinal cord also protrudes, apart from the meninges, the defect is called a meningocele, which is often accompanied by moderate to severe neurological symptoms and even disability. The most severe form is myeloschisis; a neuronal tissue called placode that is neither protected by the skin nor the meninges. This type of spina bifida is accompanied by the most complications.^{1,6}

Although the shared feature picture of NTDs is incomplete neural tube closure, the available evidence suggests both genetic and environmental factors. The defects occur due to various risk factors simultaneously disrupting the neural tube closure process, but not sufficiently damaging if it stands alone.^{1,8} Lack of folic acid during pregnancy will increase the risk of having a child with spina bifida.^{8,9} Evidence from a recent study suggests a relationship between mothers who work or live in agricultural areas, exposed to chlorinated substances, magnetic radiation, anesthetic gases, and organic pollutants with the occurrence of NTDs. Other evidence suggests that maternal stress and consumption of certain drugs during pregnancy will also increase the risk of NTDs. Genetic markers that appear to be associated with the onset of NTDs are mutations in the methylenetetrahydrofolate reductase and dihydrofolate reductase genes.⁸

Most of the time, spina bifida is not a syndrome. However, it can also appear as part of certain genetic disorders, such as Jarcho-Levin syndrome, DiGeorge syndrome, and Turner syndrome. The proportion of spina bifida as part of the syndrome is less than 10% of all NTDs.^{3,4}

Before the 1970s, the mortality rate for patients with spina bifida was almost 40%, but now it has decreased due to advances in health care and medical equipment support. The survival rate for spina bifida patients is now close to 75%.¹⁰ The management of spina bifida is currently improving due to the aggressive measures in the neonatal period, preventive catheterization of bladder functions, home care, and neuroimaging technology that allows suitable radiological evaluation.¹¹⁻¹³

A prospective study showed that sensory function at

birth is correlated with long-term outcome in spina bifida patients. Patients with the knee-level sensory function had a higher survival rate (60%) compared with the patients who did not have sensory function below the umbilical area (17%). Patients who do not require CSF diversion or do not undergo shunt correction tend to be more independent and mobile. When followed for 50 years, there were 37 survivors (31%). For those born with the sensory function below L3, the survival rate was 54%, while for those born with the sensory function at the L3 level or above, the survival rate was only 22%. Nearly more than half of people with spina bifida who survive to age 50 years of age can live independently. Those who did not experience hydrocephalus or undergo shunt revisions can live more independently.¹⁴

Conditions associated with the spina bifida include hydrocephalus, Chiari type II malformation (CM-II), as well as gastrointestinal and genitourinary impairment.¹⁵ Low Apgar scores, hydrocephalus, and CM-II with central respiratory problems found at birth are several factors associated with early mortality. Hydrocephalus is the most frequent finding, with nearly 90% of patients requiring insertion of a ventriculoperitoneal shunt or endoscopic third ventriculostomy by median age of 5 days. Nearly 90% of patients with defects at L5 or higher require a shunt, and about 50% of patients with defects as high as S1-2 require a shunt.¹⁶

Type II Chiari malformation is an inferior displacement of the cerebellar vermis, medulla, and tonsils.¹⁵ Although closely related to myelomeningocele, this symptom only occurs in one-third of patients. Symptomatic CM-II patients will suffer from swallowing problems, airway-breathing difficulties, or even apnea and may require multiple surgical interventions, including surgical decompression.¹⁷

In patients with symptomatic CM-II, the primary goal is to reduce intracranial pressure by CSF diversion and surgical decompression. Young patients with CM-II also need physiotherapy in terms of bladder function, respiratory-related disorders, and digestive-related disorders.¹⁵

This newborn had lower-limb paralysis, which was also accompanied by sensory abnormalities, urinary and fecal incontinence, and breathing and feeding difficulties. The breathing problems in this neonate improved, and at the end of the hospitalization period he could breathe easily without supplemental oxygen. Aminophylline administration appeared to be effective in this patient. Feeding and oral tolerance were gradually improved with the continuation of oromotor therapy. Although he had a fever with elevated leucocytes ($21\,300/\text{mm}^3$) in the midway of the treatment period and was strongly indicative of having an infection, the modified Tollner score for sepsis did not meet the criteria for sepsis, the IT ratio was within normal limits.

Considering the clinical condition of the neonate, the

occurrence of CMII appears to be excluded, although it needs to be monitored for long term follow up. At our hospital, there is no magnetic resonance imaging (MRI) examination available, so it was not carried out. The head ultrasonography was the only non-invasive radiological examination performed in this patient. We also administered empirical systemic antibiotics because there were no facilities for culture and sensitivity testing. Hence, we tried maximizing the measures we can take, especially in the aspect of initial airway management, parenteral nutrition, and wound care that were carried out very carefully. After ten days of hospitalization, he was allowed for discharge and scheduled for surgery.

Newborn with spina bifida generally survive with proper medical support but suffer from neurological disorders, the severity of which is determined by the level of the defect. A clinician must be ready to anticipate any clinical condition associated with spina bifida and its complications, and empathetically explain the neonate's condition to the parents, given that the cases are quite often encountered in daily practice.

Conflicts of Interest

The authors declare no conflict of interest in this study.

Ethical Approval

Informed consent was obtained from of parents of the patient for publication of this case report.

Authors' Contribution

IS conceptualized the study and was involved in data acquisition, drafting the manuscript, and critical revision. WH was involved in data acquisition and manuscript revision.

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Supplementary Files

Supplementary File contains informed consent.

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