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Spina Bifida Complicated by Hydrocephalus: A Case Report

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Abstract

Spina Bifida is a congenital anomaly that arises from incomplete development of the neural tube. It is commonly used as a nonspecific term referring to any degree of neural tube closure. It can be further subdivided into spina bifida occulta and spina bifida aperta. Spina bifida occulta, or closed spinal dysraphism, is the mildest form of the neural tube defects (NTD) which involves a hidden vertebral defect and minimal neural involvement. Spina bifida aperta, or open spinal dysraphism, refers to a defect in which neural tissues communicate with the external environment such as meningocele and myelomeningocele. These conditions result in a varied spectrum of neurological effects due to the degree of neuralization. Spina bifida is commonly associated with several other developmental abnormalities which makes a multidisciplinary medical plan paramount to survival and positive outcomes.

Keywords*: Spina bifida, folic acid, genetic, ultrasound, delivery.*

Introduction

Worldwide, about 150 000 infants are born with spina bifida yearly, making this condition one of the most common fetal central nervous system anomalies compatible with life.The etiology is often multifactorial, which includes environmental, maternal, and genetic factors. Over the last two decades, major changes in the prenatal diagnosis and management of spina bifida have been introduced **[**1**]**.

Case Report

We report the case of a young 28 year old parturient, with no pathological history, non-consanguineous marriage, primiparous, the pregnancy is not followed up until her consultation at the end of 39 weeks of amenorrhoea for pelvic pain without hydrorrhoea, the examination finds a patient in good general condition at the beginning of labour, at the ultrasound scan we find an evolving monofetal pregnancy in cephalic presentation, with a quadriventricular hydrocephalus with laminated brain parenchyma, the lateral ventricles are dilated to more than 4cm with septal rupture, the examination of the feotal spine revealed a solidokystic image at the level of the sacral spine measuring 4*3cm well limited evoking a

spina bifida or a sacrococcygeal teratoma. The rest of the foetal ultrasound exploration showed no obvious abnormalities subject to an ultrasound at advanced term.

The decision was to perform a caesarean section for the patient given the hydrocephalus with a biparietal diameter greater than 10 cm with the birth of a newborn with obvious macrocrania, spina bifida and bilateral clubfeet, referred to paediatric surgeons for further management.

Discussion

Neural Tube Defects (NTD) result from incomplete closure of the neural fold at 3-4 embryonic weeks. The anatomic site at which the abnormal closure occurs, defines the type of NTD**[**1**]**. Failed closed of the cranial part of the neural tube results in acrania. Spina bifida is the result of failed caudal closure. More than 90% of spina bifida cases will be open defects where the neural elements are exposed at the level of the skin. A lesser portion of spina bifida cases are closed defects, where skin still covers the neural tube. Within open spina bifida, different subtypes exist, defined by what is contained within the defect. While meningoceles contain only dural sac, myelomeningoceles (MMC) also contain neural elements that are attached to the herniated dural sac. Both present as a 'bulge' on the lower spine. The term myeloschisis is used when the neural placode is exposed without associated dural herniation and presents as a 'flat' lesion **[**2**]**.

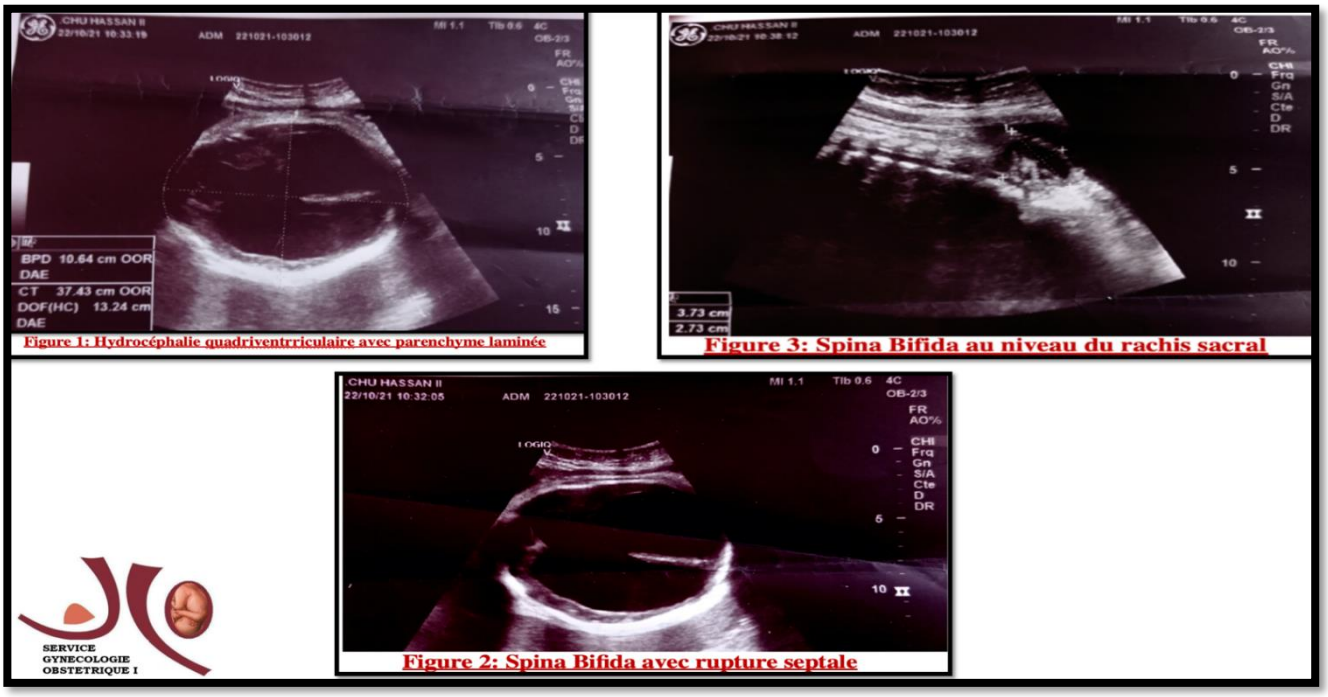


Figure 1: Hydrocéphalie quadriventriculaire avec parenchyme laminée

Figure 3: Spina Bifida au niveau du rachis sacral

Figure 2: Spina Bifida avec rupture septale

Most cases of neural tube defects are considered to be multifactorial in origin and are influenced by a combination of geographic, ethnic, genetic and dietary factors. The prevalence of neural tube defects ranges around 0.5 to 1 per 1000 pregnancies in Europe, China, and the Americas, but reaches 4 per thousand pregnancies in India **[**3**]**. Part of this geographic variation in incidence can be explained by dietary variations. Indeed, micronutrients play an important role in the etiology of NTDs. The most notable of these is folate (also known as folic acid or vitamin B9). Folate is an essential nutrient formed by humans, and must be obtained through dietary intake. Low serum folate levels are associated with higher incidences of fetal neural tube defects **[**4**]**. In addition to folate, low vitamin B12 levels, have also been suggested as a possible risk factor for NTD. In less than 10% of cases, spina bifida is due to genetic syndromes or chromosomal anomalies, including the common trisomies 13 and 18, triploidy and microdeletions or duplications **[**1**]**.

Spina bifida can be detected prenatally and more than 90% of cases will be detected before birth in countries with a national screening program for birth defects.

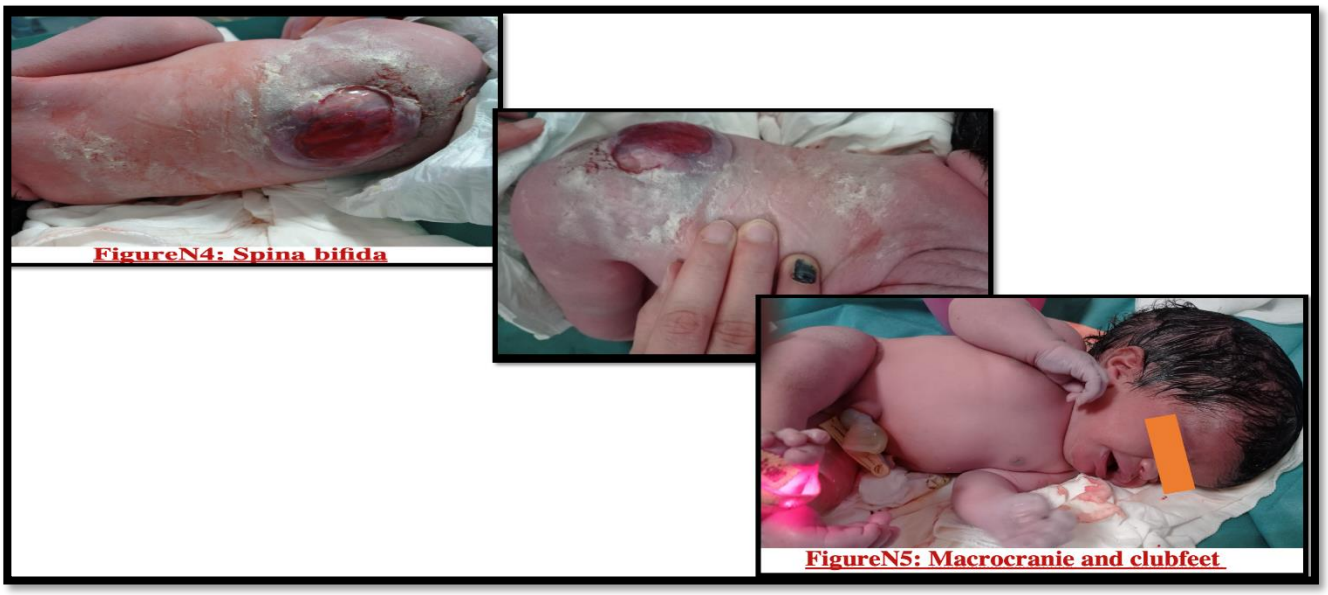
Currently, ultrasound is considered the gold standard screening tool for anatomic anomalies, as it is non-invasive, inexpensive, safe and more specific than biochemical screening. Sonographic features of open spina bifida can be detected in the 2nd trimester, and include an abnormal skull shape with flattened temporal bones (‘lemon sign’), an abnormal appearance of the cerebral ventricles (pointy appearance of the posterior horns) and ventriculomegaly (figureN1). Changes to the posterior fossa can also be visualized, such as cerebellar herniation (Chiari II malformation, ‘banana sign’) and non-visualization of the cisterna magna. The defect at the level of the spine, including a saccular or cystic lesion (figure N3) and abnormal or incomplete closure of the posterior vertebral arches is often more difficult to detect **[**5**]**. When looking at the fetal lower extremities, infrequent or lack of lower limb movement and abnormal positioning of one or both feet (talipes) may be noted, especially later in pregnancy. Successful visualization of the intracranial translucency, which is the sonographic equivalent of the fourth ventricle can be achieved in >97% of cases in expert centers. Using the intracranial translucency as a screening for fetal open spina bifida has a sensitivity of 53.5% and a specificity of 99.7%

When fetal spina bifida is suspected based on screening tests, the pregnancy should be referred to a tertiary care center for further assessment and counselling. There, a detailed anatomic survey should be completed to rule out associated anomalies, which are seen in 20-30% of cases. Additionally, the level of the spinal defect and the presence of eventual secondary findings (such as ventriculomegaly and club feet) will be assessed. This can be done using neurosonography, three-dimensional ultrasound

or fetal MRI for spinal level of the lesion to determine where the lesion lies in the vertebral column and the associated innervation emanating[6].

Further genetic counselling and invasive genetic testing should be offered to exclude chromosome anomalies. counselling and management of pregnancies complicated by fetal spina bifida, should occur with a multidisciplinary team, experienced in the management of this condition. Ideally, this involves fetal medicine specialists, (neuro)radiologists, geneticists, neurosurgeons, neonatologists, (neurodevelopmental) pediatricians, urologists as well as psychologists and social workers [6].

It is important that the information presented to parents was objective, value neutral, individualized and comprehensive, encompassing all aspects from fetal to adult[7].



FigureN4: Spina bifida

FigureN5: Macrocranie and clubfeet

Obstetric complications are rare and most pregnancies complicated by isolated fetal spina bifida will continue to term, yet the risk of preterm birth before 37 weeks is around 15-20%[2]. In open spina bifida, prolonged exposure of the neural tissue to neurotoxic amniotic fluid, microtrauma at the level of the exposed neural elements as well as continuous leakage of cerebrospinal fluid during gestation may result in progressive development of hydrocephalus and loss of nerve function[8]. Postnatally, this presents as musculoskeletal weakness, bowel dysfunction, genitourinary dysfunction, and sustained hydrocephalus [1].

Ongoing antenatal obstetric care is critical. In addition to social and mental health support, antenatal appointments provide opportunities to educate parents about the disease. Moreover, we recommend fetal ultrasound assessment every 4 weeks to assess for fetal biometry, paying close attention to head

measurements (head circumference and biparietal diameter) as these may determine mode of delivery (caesarean section in case of severe macrocephaly). We would typically also assess the degree of ventriculomegaly as well as fetal limb movements. The latter, however, would not affect pregnancy management.

Fetuses with spina bifida should be delivered in a center with neurosurgical services on site. There is no clear evidence that cesarean section improves outcomes in infants with spina bifida and most centers will allow for vaginal delivery in the presence of small to moderate size lesions. At the time of delivery, use of latex-containing materials should be avoided, as infants with spina bifida are at high risk of developing latex allergies **[**9**]**.

After birth, the spinal defect will be closed surgically within the first 24 hours of life, to prevent ascending infection55. The risk of perinatal death in recent cohorts is less than 5%. Within 6 months of life, approximately 60-80% of infants with spina bifida will need either ventriculo- peritoneal shunting or third ventriculostomy to help treat hydrocephalus and the common co- occurrence of Chiari II malformation **[**8**]**.

It is important to initiate the conversation regarding prevention of NTDs in subsequent pregnancies, Recurrence rates for a woman who has had a previous pregnancy affected by a fetal NTD vary based on the underlying cause of the NTD. The recurrence risk of a multifactorial NTD, in a low prevalence population is estimated to be 2-4%. This can be reduced to 1% with high-dose (4mg/day) folic acid supplementation, ideally initiated at least 3 months prior to conception, and most importantly throughout the first trimester **[**10**]**. Vitamin B12 supplement is also recommended, even though the evidence for this is less strong. Finally, women who have had a previous child with spina bifida, are recommended to undergo a first trimester fetal anomaly screening in subsequent pregnancies for early detection of spina bifida **[**1**]**.

Conclusion

Spina bifida is a severe congenital anomaly. Despite the availability of fetal and postnatal treatment options, infants with spina bifida face many challenges through to adult life. As such, it is important that prospective parents are well informed when making decisions regarding their pregnancy. Spina bifida counselling and interventions should be performed by multidisciplinary teams with expertise in this area and we have here provided some guidance as to what can be discussed.

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