

Myelomeningocele: A Case Report

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Abstract

Neural tube defects are the second leading cause of congenital malformations worldwide. In Cuba, most malformations are detected prenatally, contributing to a significant decrease in the birth of malformed children. In recent years, there has been an increase in the diagnosis of these malformations postnatally, although mostly of small size. The following article is carried out to describe a case of the postnatal diagnosis of a large, closed myelomeningocele, as well as review the literature in this regard. It was concluded that most myelomeningoceles are associated with a maternal folate deficiency during the first trimester of gestation. Many cases are diagnosed prenatally, although there may be cases with postnatal diagnosis, which represents a challenge for doctors and the patient due to the significant morbidity and mortality associated with the pathology.

Keywords: mielomeningocele, spina bifida, neural tube defect.

Introduction

Neural tube defects (NTDs) are the second most prevalent cause of congenital malformations, only preceded by heart disease. [1] These are a heterogeneous group of structural congenital disabilities that arise from a complex series of genetic and environmental factors and adversely affect the structure and function of the brain and spinal cord in the form of cranial defects and open or closed spinal dysraphism. [2] Open spinal dysraphisms or open spina bifida are often seen on physical examination as visible lesions and include malformations such as myelomeningocele (MMC), the most common anatomic type of spina bifida. [3,4]

Currently, MMC is a pathology that is among the 10 leading causes of neonatal mortality and is associated with significant morbidity, including the effects of hydrocephalus, whose incidence related to MMC after postnatal closure is 57 to 86 %, to different degrees. Paralysis of the lower limbs according to the affected vertebral level and mixed incontinence (50-90 %). [5,6,7,8]

The risk increases to 2–3% if there is a previous birth with MMC and 6–8 % after two affected children. The risk also increases in families where close relatives have given birth to children with MMC, especially when the relationship is maternal. Prenatal folate (in the form of folic acid) reduces the incidence of MMC. Theworldwide incidence of MMC is approximately 1 to 2 cases per

1000 live births. In South America, the prevalence is 6 % in births, and neural tube closure represents 24 cases per 10,000 births, afigure that exceeds the prevalence in the rest of the world. [9,10]

In Cuba, spina bifida with MMC is 0.6 to 0.7 per 1,000 births. Most of the malformations are detected by the National Program for the Detection of Congenital Malformations, giving the mother the possibility of interruption of pregnancy if desired, contributing to a significant decrease in the birth of children with open spina bifida.[10]

In the central region of our country, in recent years, an increase in the diagnosis of these malformations has been observed, although the majority are small in size. The following article is carried out to describe a case of the postnatal diagnosis of a primarily closed myelomeningocele, as well as review the literature in this regard.

Case report

Newborn patient, son of a 22-year-old mother with previous obstetric health history. Eutocic delivery at 40.5 weeks, membrane rupture time 10 min, cord and placenta of typical characteristics, weighing 3750 g, and Apgar score 8-9.

He was immediately transferred to neonatology due to presenting a 9 x 9 cm rounded volume increase in the lumbosacral region with

well-limited edges, wholly covered with skin suggestive of neural tube closure defect. The rest of the physical examination was regular. Blood analysis, transfontanellar, and abdominal ultrasound were routine.

Spinal ultrasound is performed where it is reported: It is observed vertebral bodies without closure defects at the upper edge of the

lumbar spine. Towards the middle and low portion of the lumbar spine, a closure defect in the longitudinal axis is observed, which allows the exit of the pocket with nerve roots that are mobilized during crying. The purse is surrounded by a 42 x 15 mm echogenic image reminiscent of lipomatous tissue.

The patient underwent emergency surgery, closing the defect without surgical complications.



Figure 1. Spinal ultrasound image shows the defect in lumbar spine and its content.



Figure 2. The picture shows the lesion in the lumbar spine of the patient.

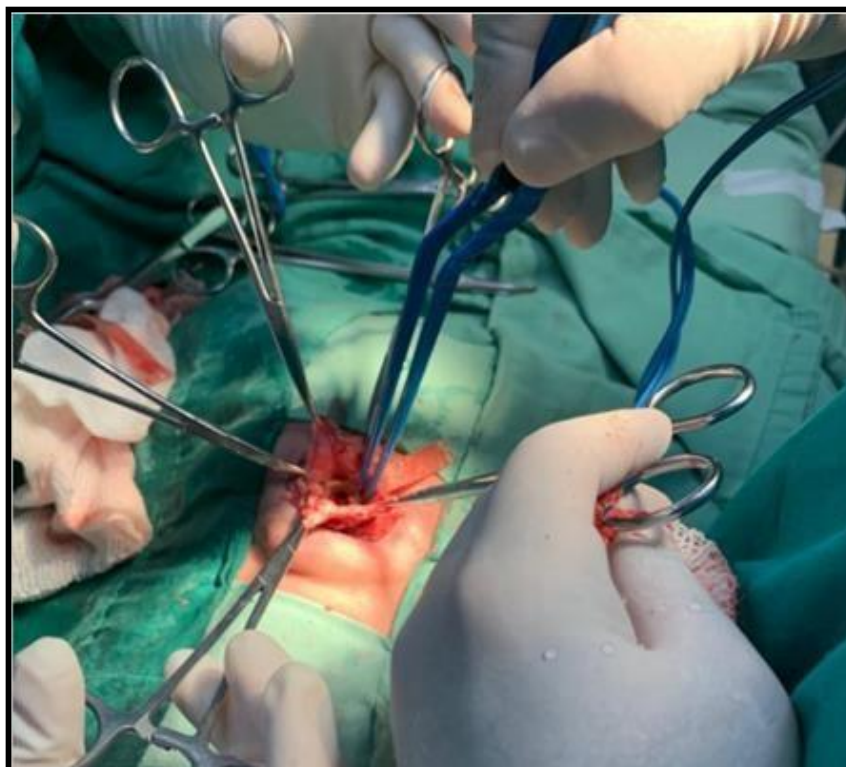


Figure 3. The picture shows the patient during surgical procedure.

Discussion

Myelomeningocele is a spinal dysraphism in which the spinal cord and its contents herniate through a congenital bony defect in the posterior elements of the spine. This is a serious malformation associated with high morbidity and mortality rates.

Myelomeningocele is the most common presentation of spinal dysraphism, accounting for approximately 80 % of cases. [1,2]

These malformations represent a relatively early failure of neurulation, which justifies the concurrence of other primary and complex macro and microscopic malformations along the neural tube, which motivate its semiological diversity and the usually bad prognosis of these patients. As a result, the overlying mesodermal and ectodermal elements fail to form, resulting in an open spinal defect almost always associated with a Chiari II malformation and its cranial manifestations. [1,2,3]

Recent experimental studies using toxic agents and animal mutants have supported the non-closure theory. Toxic agents include cytochalasin, vinblastine, calcium channel blockers, phospholipase c, concanavalin a, retinoic acid, hydroxyurea, and mitomycin c. Folate and its pathophysiology have been the focus of most recent research. Hibbard suspected the association between insufficient folate and NTDs in 1964. A randomized, double-masked study in 1991 showed that couples in the United States with a history of a baby with a neural tube defect have a 2% to 3% chance of having a second child born with a neural tube defect. If 4 mg of folic acid was consumed during the critical period (before and during pregnancy), the risk of having a second child born with spina bifida decreased by 71 % [26], suggesting that some, but not all, defects are related to abnormal folate supplementation. In our case, the mother's lack of folic acid intake before and during the first trimester of pregnancy was recorded as a history, which was identified as one of the possible causes. [1,2,3,5,10]

The most useful investigation in the early detection of open spina bifida is the determination of Alpha-fetoprotein in the amniotic fluid or the mother's blood; sometimes, the meningeal herniated sac can be seen by ultrasonographic study. Determination of maternal serum alpha-fetoprotein levels in the early part of the second trimester is the initial test for neural tube defects. In amniotic fluid, alpha-fetoprotein concentration is 100 times lower than in fetal Cerebro Spinal Fluid (CSF). The optimal time to sample alpha-fetoprotein is 16-18 weeks, although alpha-fetoprotein levels can be determined

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between 14-21 weeks gestation. Seventy-nine percent of open defects pregnancies and 3 % of normal singleton pregnancies have an alpha-fetoprotein level of 2.5 multiples of the median (MoM) at 16 to 18 weeks gestation. Amniocentesis may be indicated if the alpha-fetoprotein level and imaging studies suggest the presence of aneural tube defect. [4,10] In this case, the patient did not present altered alpha-fetoprotein concentration, and the lesion was not seen in prenatal ultrasound studies.

In the lesion description, the cardinal sign is the presence of a sac or a tumor, almost always covered by atrophic skin. Sometimes, other alterations appear in the neighboring skin, such as cutaneous angiomas, hair, or lipomas. (6; 8) In our case, a mass was observed in the posterior midline, wholly covered with skin and without other alterations.

No other malformations were detected in our patient, but other associated alterations were frequently observed, the most frequent being hydrocephalus, stenosis of Silvio's aqueduct, agenesis of the corpus callosum, Chiari malformation, cortical neuronal migration disorders, hydromyelia, among others. ; the most constant is the Chiari malformation, which is the descent of the elements of the posterior fossa (brain stem, cerebellum, and IV ventricle) below the foramen magnum. It is present in 80 % of patients with myelomeningoceles. The different malformations that occur in the MMC can either evolve with the development of the individual or manifest throughout it, forming the dysgraphic state, varied and, above all, dynamic, according to which the lives of these patients are marked out and threatened by a diversity of complications whose prevention, early diagnosis and treatment are the objectives that the care team must attend to, our patient did not present any associated malformation, which led to a good post-surgical evolution. [1,3,7,11,12]

Conclusion

Most myelomeningoceles are associated with a maternal folate deficiency during the first trimester of gestation. Many cases are diagnosed prenatally, although there may be cases with postnatal diagnosis, which represents a challenge for doctors and the patient due to the significant morbidity and mortality associated with the pathology.

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