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SCIENTIFIC ARTICLE - CASE REPORT

FETAL MYELOMENINGOCELE REPAIR: A CASE REPORT

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ABSTRACT

Neural tube defects are common in fetal life, particularly those involving neural tube closure, and are often associated with deficiencies in essential nutrients, such as folic acid and vitamin B12. Among the main malformations, myelomeningocele stands out as the most severe form of spina bifida. The purpose of this article is to report the surgical treatment of a 26-week pregnant patient with a fetus diagnosed with myelomeningocele, who was monitored in the malformed fetus outpatient clinic and underwent correction via laparotomy. The surgery was successful, and the newborn was in good general condition without neurological injuries. Early diagnosis should be made during prenatal care, as early correction in the second trimester of pregnancy can be effective in reducing possible motor sequelae. The treatment approaches are laparotomy, the traditional method, and fetoscopy, a newer and less invasive technique.

Keywords: Fetal surgery, Intrauterine surgery, Meningocele surgery.

INTRODUCTION

Neural tube defects are among the most common issues during fetal life, particularly those involving neural tube closure, which involve the failure of the spine or head of the fetus to close properly. During fetal development, the human spine undergoes a process of posterior vertebral closure which, in certain circumstances, does not occur correctly. A deficiency in essential elements, such as folic acid and vitamin B12, especially during the periconceptional phase—the period close to conception—can impair this closure process, resulting in severe defects.

Neural tube defects (NTDs) are serious congenital malformations that affect the central nervous system and spine, resulting from a failure in neural tube closure during development. The most common NTDs include anencephaly, spina bifida, and encephalocele.[1] Anencephaly is characterized by a partial or complete absence of skull bones with minimal brain remnants. Spina bifida occurs when the vertebrae do not fully close over the neural tube, exposing the spinal cord and nerves, and is compatible with survival.[2] Encephalocele involves a protrusion of

the brain and/or its membranes through the skull.[2]

Maternal folic acid deficiency before and during early pregnancy is one of the most common and preventable risk factors.[3,4] Folic acid supplementation is an effective primary prevention strategy, and mandatory food fortification with folic acid has been implemented in several countries to reduce the incidence of NTDs.[4] Furthermore, prenatal diagnosis through ultrasound and molecular markers is crucial for early detection and for implementing treatment strategies.[5]

These defects can have significant consequences on an individual's life, including neurological, motor, and cognitive impairments. Among the most common effects are hydrocephalus, motor difficulties in the lower limbs (including congenital clubfoot), and complications in bowel and bladder control. The impairment of these functions results in conditions such as urinary and fecal incontinence, as well as impaired glycemic control, directly impacting the patient's quality and longevity.

The estimated global prevalence of NTDs is around two cases per 1,000 births, with significant variations.[4,6] Effective prevention of NTDs requires awareness of the importance of folic acid supplementation and the implementation of public health policies to ensure fortification.[4]

CASE REPORT

A 26-week pregnant patient, followed in the malformed fetuses outpatient clinic at the Hospital das Clínicas of the Federal University of Goiás (HC-UFG), presented with a suspected diagnosis of myelomeningocele (MMC) in the lumbar region during prenatal consultations. To confirm the diagnosis, a pelvic MRI was requested, as shown in Figure 1. Following the diagnosis, the treatment was carried out by the surgical team at HC-UFG, coordinated by Dr. Waldemar Naves do Amaral. The surgery was performed by him, along with the neurosurgery team. The chosen access route for correction was laparotomy through the maternal abdominal wall, followed by exposure of the spine and correction.

The surgery progressed successfully, without complications, and the patient maintained good progress throughout the pregnancy. The delivery occurred safely, with the newborn in good general condition, showing proper healing of the spine and no signs of motor impairment in the lower limbs, Figure 4. Currently, the newborn has mild hydrocephalus, with no indication for surgical interventions.

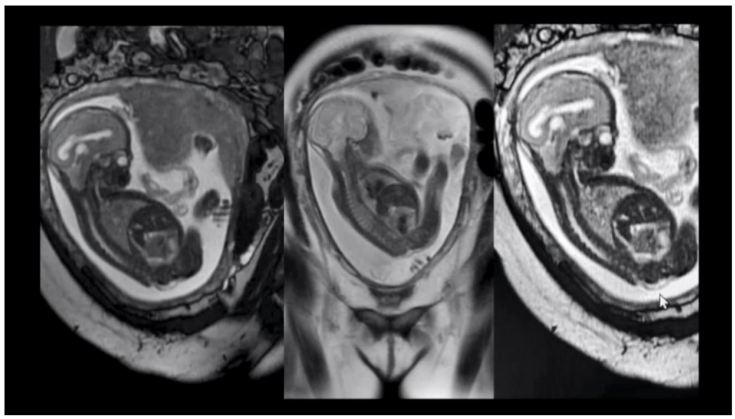

Figure 1: Pelvic MRI for the diagnosis of MMC

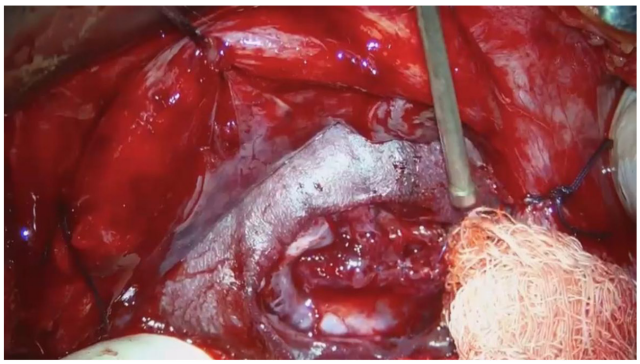

Figure 2: Exposure of MMC

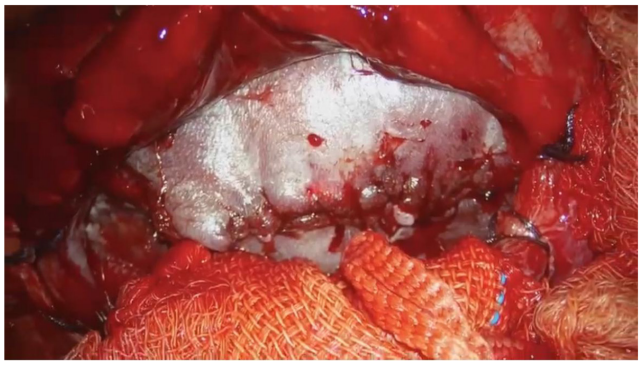

Figure 3: Surgical correction of MMC

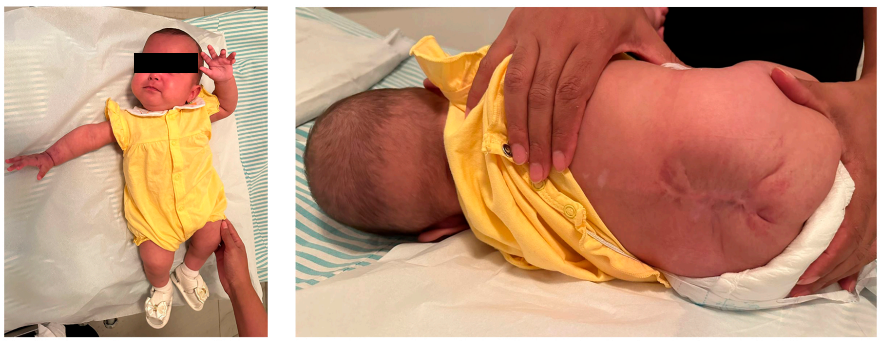

Figure 4: Postnatal outcome with proper healing of the myelomeningocele surgery

DISCUSSION

This study presents a case report aimed at describing the fetal treatment of the patient through laparotomy. MMC is the most severe form of open spina bifida and is one of the most common severe congenital malformations. Historically, MMC repair surgeries were performed postnatally, covering the exposed spinal cord and preventing infections, but still carrying the risks of neurological damage, hernia, and hydrocephalus associated with incomplete neurulation or mechanical and chemical trauma. Thus, prenatal closure has emerged as an excellent option, resulting in improved motor function, reduced hindbrain hernia, and decreased need for cerebrospinal fluid diversion.[7]

Fetal ultrasound is the primary diagnostic tool, often performed between 18 and 20 weeks of gestation. It is important to clearly define the location and size of the NTD and confirm whether it is open or closed. Additionally, it is possible to associate the condition with other abnormalities, so the exam for secondary findings, such as hydrocephalus, should be conducted, and fetal echocardiography should be considered. The measurement of acetylcholinesterase in amniotic fluid helps differentiate between open and closed NTDs and is a component of many preoperative evaluations for fetal closure.[8]

Genetic evaluation with amniocentesis should also be recommended, particularly for those considering fetal closure. The identification of genetic abnormalities in the fetus has important implications for counseling on prognosis, pregnancy management, and whether fetal closure of the NTD is an option. Fetal MRI may also be considered, particularly if there are unclear findings on ultrasound.[8]

The treatment is fetal intervention, which can be performed during pregnancy. There are two main approaches to correct these defects before birth: the traditional approach, which involves open surgery in the second trimester of pregnancy, and the more recent approach, fetoscopy, which is minimally invasive.

The surgery is performed between the 20th and 28th week of gestation. The procedure involves a laparotomy, where an incision is made in the pregnant woman's abdomen to access the uterus. A window is then created in the uterus, and the fetal spine is exposed with the help of ultrasound. The neurosurgery and obstetrics teams, working together, perform the correction of the meningocele, restoring the alignment of the spine and repairing the defect by placing a mesh, when necessary. This approach can reduce or even eliminate the sequelae associated with neural tube defects, reduce the need for ventriculoperitoneal shunting, and reverse the hindbrain hernia associated with Chiari II malformation, significantly improving the patient's quality of life after birth. However, the open maternal-fetal surgical approach is associated with a relatively higher risk for the patient, the pregnancy as a whole, and future pregnancies.[9]

In some cases, fetoscopy can be performed, a more recent and less invasive technique. Fetal repair of open spina bifida through fetoscopy minimizes maternal risks while providing similar neurosurgical outcomes for the fetus. The percutaneous approach avoids laparotomy and uterine exteriorization, and it is associated with a lower risk of anesthesia and better maternal post-surgical recovery.[10] Through a small access in the uterus, using a fetoscope, it is possible to visualize the lesion in the fetal spine and perform the correction with precision. This technique involves the insertion of small cameras and surgical instruments, with accesses no larger than 5 millimeters, minimizing risks for both the pregnant woman and the fetus. The correction can be performed by implanting meshes and repairing spinal hernias, with excellent long-term results.

CONCLUSION

MMC is a common severe congenital malformation associated with deficiencies in folic acid and vitamin B12. Early diagnosis, combined with appropriate interventional treatment, allows for resolution during the fetal stage. Currently, there are two main approaches for fetal myelomeningocele surgical correction: laparotomy, the traditional approach, and fetoscopy, a less invasive and more recent technique. This approach helps reduce the damage and sequelae derived from neural tube formation defects.

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