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Case Report

Fetal surgery for occipital encephalocele: A case report

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ABSTRACT

Background: Occipital encephalocele is a congenital defect of the neural tube at the level of the cranial midline, which results in herniation of meninges and brain tissue. The results of the management of myelomeningocele study determine the maternal and fetal risks for an open fetal surgery and have motivated the constant review of the concepts and strategies which the pediatric neurosurgeon can employ for the treatment of neural tube defects in the prenatal period.

Case Description: We present a case of a female patient in utero of 26 gestational weeks with the diagnosis of an occipital encephalocele treated by open fetal surgery. During week 20 of gestation, the diagnosis of occipital encephalocele was made by ultrasound, which was corroborated by fetal magnetic resonance that showed cranial protrusion of neural and meningeal content in the occipital region, measuring 1.6 × 2.8 × 3.3 cm with an approximate volume of 7.7 cc through a bone defect of 6 mm. The closure of the defect was performed by the postnatal surgical technique adapted to the open fetal surgery. Later, the patient was born transabdominal with a 2.8 cm occipital wound, with suture points and approximated borders, normocephalic, without clinical signs of sepsis, hydrocephalus, or overt neurologic compromise.

Conclusion: Open fetal surgery is a therapeutic option in the face of an isolated occipital encephalocele. This case report demonstrates the viability of the surgical procedure by the adaptation of a postnatal surgical technique to a prenatal surgery. Further studies are needed to evaluate the long-term functional results, comparing them with those seen in patients who undergo a postnatal procedure.

Keywords: Fetal surgery, Cranial dysraphism, Occipital encephalocele

INTRODUCTION

Occipital encephalocele is a congenital defect of the neural tube at the level of the cranial midline, which results in herniation of the meninges and cerebral tissue.[3] It has a variable incidence according to geographic location and race, going from 0.8 to 3/10,000 alive newborns and corresponding to 15–20% of all cases of craniospinal dysraphism.[3,7]

Encephalocele is an anomaly of the embryonic mesoderm resulting from a faulty differentiation of the superficial ectoderm with the neural ectoderm.[7] It manifests as an occipital protrusion of different sizes confined to the midline between the lambdoid suture and the foramen magnum. According to its relationship with the torcula, it is classified into supra or infra torcular, and the bone defect may vary in size from a few millimeters to over 20 cm; in some cases, the sac is as big as the newborn's head (giant encephalocele).[6]

Although it can occur as an isolated disorder, it can also be associated with other congenital anomalies or genetic syndromes.[7] The majority of these patients die at birth, and most of these deaths occur in the 1st year of life.[3,7] Those who survive will exhibit a certain degree of neurodevelopment retardation or hard-to-control convulsive crisis.[3,7] Risk factors associated with mortality are as follows: the presence of microcephalus, neonatal sepsis, size of hernial sac, and herniated neural tissue within the sac.[6]

Ultrasound is sufficient for an accurate diagnosis with a sensitivity detection of over 90% which can be supplemented or not with a fetal magnetic resonance.[6,7]

The results of the management of myelomeningocele study (MOMS study) determine the maternal and fetal risks for open fetal surgery and have motivated the constant review of the concepts and strategies which the pediatric neurosurgeon can employ for the treatment of neural tube defects in the prenatal period.[1,2]

CASE DESCRIPTION

Female patient *in utero* of 26 weeks' gestation by obstetric ultrasound product of a third pregnancy from a 30-year-old woman with a history of gestational diabetes diagnosed at 12 weeks' gestation, treated with Metformin. During week 20 of gestation, an obstetric ultrasound is performed, which reveals an occipital encephalocele [Figure 1].

A fetal magnetic resonance scan demonstrated a single product of approximately 26 weeks by fetometry with a midline cranial protrusion of neural and meningeal content similar to cerebrospinal fluid (CSF) in occipital region measuring 1.6 × 2.8 × 3.3 cm and an estimated volume of 7.7 cc as well as an occipital bone defect of 6 mm [Figures 2-4].

The multidisciplinary team evaluated the potential risks described for open fetal surgery and, along with the ethics committee of the hospital institution and the signed informed consent, decided to perform the postnatal surgical technique for the repair of an occipital encephalocele adapted to an *in-utero* procedure.

Surgical technique

The surgical procedure was performed at the National Medical Center La Raza Mexican Institute for Social Security in Mexico City on April 27, 2023. The steps followed in the surgical obstetric technique were like those proposed by Adzick *et al*. in the MOMS study.[1]

The abdominal wall is opened with an extended Pfannenstiel incision; the gravid uterus is extracted from the abdominal cavity, followed by a detailed mapping of placenta localization and identification of the parts of the umbilical cord by means of a transoperative echography. The site of the hysterotomy

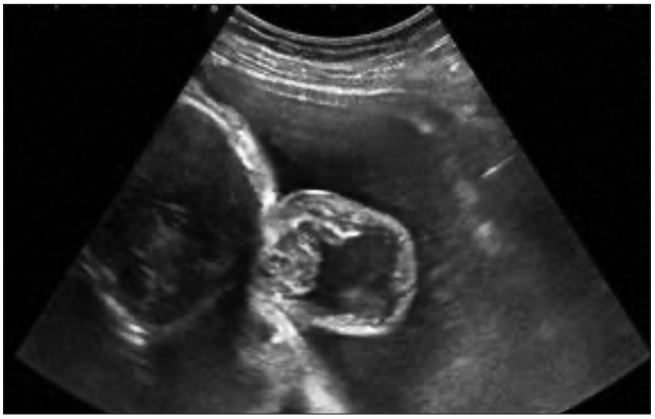

Figure 1: Obstetric ultrasound during week 20 of gestation with occipital encephalocele.

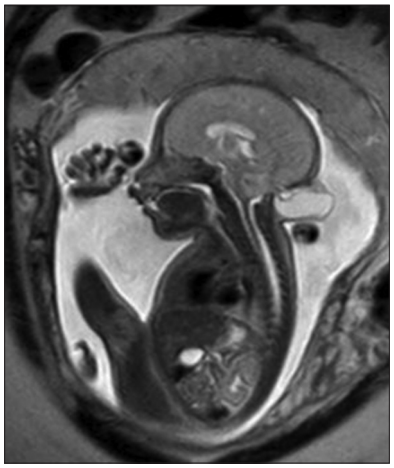

Figure 2: Sagittal view of fetal magnetic resonance T2 weighted image showing an occipital protrusion measuring 1.6 × 2.8 × 3.3 cm

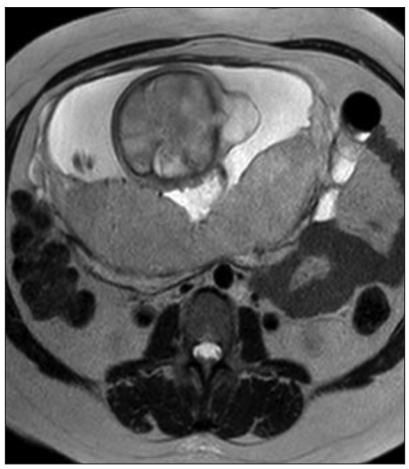

Figure 3: Axial view of fetal magnetic resonance T2 weighted image showing an occipital protrusion with a bone defect of 6 mm.

depends on the placenta localization. A 3 cm longitudinal mini-hysterotomy was performed [Figures 5a-b].

The neural surgical steps begin with the positioning of the head of the fetus on the site of the uterine opening guided by transoperative ultrasound. The repair of the encephalocele starts with a midline incision of the hernial sac [Figure 5c]. Hemostasis is performed with a bipolar clamp at a very low intensity. Then, exposure of meninges and circumferential dissection is carried out, followed by cutting at the level of the external table of the occipital bone by means of micro scissors and monopolar at a very low intensity [Figure 5d].

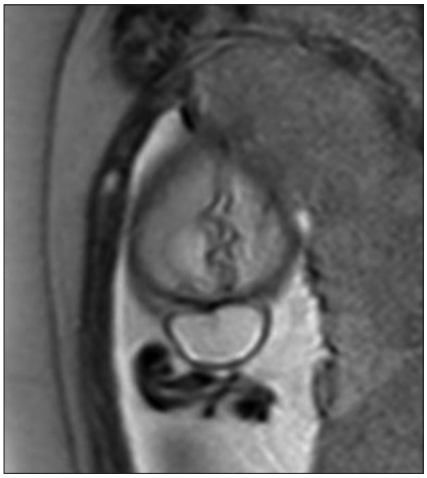

Figure 4: Coronal view of fetal magnetic resonance T2 weighted image showing an occipital protrusion with an estimated volume of 7.7 cc.

A safety margin of 1.5 cm was maintained in the meningeal layer to have a tight closure without complications. The protruded neural tissue was returned to its intracranial position, and a tight closure of the meningeal layers with a 5-0 propylene running suture was completed [Figure 5e].

The cutaneous remnant was excised, and the skin was reconstructed, avoiding redundant tissue with a 25 3-0 poliglecaprone running suture. After the repair of the occipital encephalocele, the fetus was returned to the uterine cavity [Figure 5f]. The immediate postsurgical management was done in an obstetric intensive care unit where the mother stayed for approximately 24 h and was discharged home 72 h later.

FOLLOW-UP AND RESULTS

Four weeks after fetal surgery, the mother entered the operating room with the following diagnoses: Pregnancy of 30.4 weeks gestation and gestational diabetes. A premature female patient was born transabdominally, with regular respiratory effort and a cardiac frequency of over 100 beats/min. Capurro of 30 weeks gestation, Silverman-Anderson O with a weight of 1480 g, height of 36 cm, and a cephalic perimeter of 26 cm with a 2.8 cm was obtained at birth. The wound in the occipital region with suture points and borders closed with a running suture without a leak of CSF or blood [Figure 5g].

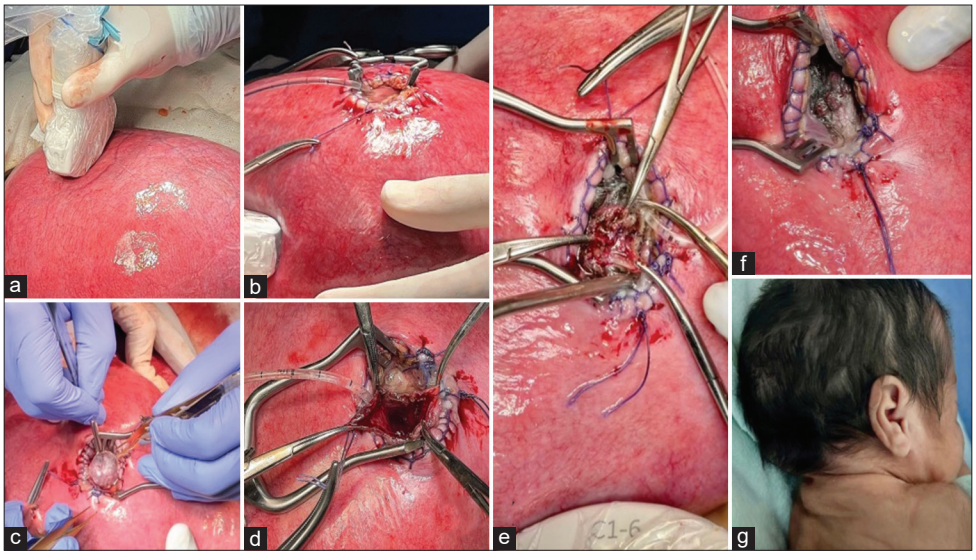

Figure 5: Surgical technique. (a) Localization of placenta by transoperative echography, planning of mini hysterotomy. (b) A 3 cm mini hysterotomy and widening by a Weitlaner retractor. (c) Exposure of occipital encephalocele. (d) Midline incision of encephalocele sac and exposure of meninges. (e) Tight closure of meninges by a 5-0 propylene running suture. (f) Complete closure of occipital defect without leak of cerebrospinal fluid (CSF). (g) Postnatal wound of clinical case with borders closed with a running suture, without leak of CSF.

She is hospitalized for three weeks, reaching the ideal weight for dismissal, without presenting clinical data of neonatal sepsis, hydrocephalus or overt neurologic compromise and having adequate respiratory effort.

After one month of follow-up, the patient is active, reactive to stimuli, normocephalic, with rhomboidal anterior fontanelle, sharply pointed posterior fontanelle, short neck with redundant skin, and a close tight wound in the occipital region with adequate healing.

DISCUSSION

Encephalocele comprises 10–20% of all craniospinal dysraphisms.[3] The exact cause of an encephalocele is unknown. It is probably multifactorial, including both environmental and genetic factors. Although it can occur as an isolated disorder, it can also be associated with other congenital or genetic syndromes.[3,7] Encephalocele can be a part of over 30 different syndromes, such as Meckel, Frasier, Roberts, Knobloch, and Walker-Warburg.[7]

Sixty per cent of patients with an occipital encephalocele are going to present another malformation and/or chromosomic defect, and at least 15–20% of newborns will have other anomalies such as defects of the neural tube, microcephaly, Chiari malformation type 2 or 3, craniosynostosis, and syringomyelia.[7]

Signs and symptoms may vary significantly depending on the size, localization, and type of content which protrudes through the bone defect.[7] Hydrocephalus is an important clinical manifestation, and it is estimated that it can occur in 60% of patients with occipital encephalocele.[6,7]

Prenatal diagnosis can be accomplished with a three-dimensional ultrasound even from week 11 of gestation, but in the majority of cases the diagnosis is made in the second trimester.[6] The assessment of an encephalocele by ultrasound should include the localization of the cranial defect, the content of the sac, and other associated findings like the presence of hydrocephalus.[6,7]

Ultrasound is sufficient for an accurate diagnosis with a detection sensitivity greater than 90%, although it can be complemented or not with a fetal magnetic resonance.[6,7]

The latter is indicated when the processes of maturation and neuronal myelination need to be evaluated.[6] High-resolution images and ultrarapid sequences of one pondered burst in T2 or of one turbo burst allow the execution of the study without the need for fetal sedation and are sufficiently precise to carry out an analysis of the fetal anatomy.[4]

The most important thing in the treatment of this pathology is the adequate prenatal diagnosis utilizing all the ancillary laboratory tests available, such as blood makers, obstetric ultrasound, magnetic resonance, and fetal karyotype.[3,4,8]

Surgery remains the only treatment for encephalocele.[2-4,6,7] The main objectives of surgery are the following: To reintegrate neural tissue, meninges, or CSF into the cranial cavity without causing neurologic deficit, repair the dural defect, and complete a cranioplasty if possible.[11] The majority of surgical techniques recommend the repair at the time of birth or within the first three postnatal months.[7,10]

Surgical indication must be considered in cases where there is a risk of rupture of the sac and/or CSF fistula, meningitis, the content of herniated tissue, presence of vascular structures, associated hydrocephalus, and cosmetic appearance.[3,4]

The complications that accompany postnatal surgery of an occipital encephalocele include the presence of postsurgical meningitis, CSF fistula, hydrocephalus, wound infection, wound dehiscence, and death.[3-5]

Fetal surgery is performed in many centers around the world for a great number of pathologies.[2] The results of the MOMS study have resolved the technical challenges of fetal surgery for neural tube defects: Successful access to the uterine cavity and closure of the gravid uterus, intraoperative fetal monitoring, and the strict control of pre-term delivery.[1,2]

The surgical repair of an occipital encephalocele is not an experimental procedure because it is the same technique utilized in the postnatal period: early identification of the meningeal layers, preservation of neural tissue (whenever possible), and tight closure of the dura mater and skin.[2,3]

At present, there are no inclusion or exclusion criteria for the selection of candidates for fetal surgery of an isolated occipital encephalocele. However, there is evidence in the literature about recommendations such as gestational age between 20 and 27 weeks gestation, mother's age equal to or over 18 years, normal fetal karyotype, and cystic hernial sac with <20% of neural tissue.[3]

Furthermore, there are recommendations which contraindicate the treatment *in utero*: the presence of syndromic fetal anomalies, chromosomopathies, neural tissue >20% of the volume of hernial sac, and the presence of vascular structures or from the encephalic trunk within the sac.[1-3]

On the other hand, other criteria that should be considered in the preoperative assessment are the following: risk for premature delivery due to adverse conditions of the mother or the course of pregnancy such as short cervix, placenta previa, oligohydramnios, chorioamnionitis, uncontrolled gestational diabetes or arterial hypertension, human immunodeficiency virus infection, hepatitis B or C, and refusal of the procedure.[1,3]

The maternal and fetal risks that are well-defined for the procedure by an open fetal surgery should be balanced and

clearly focus on the benefit of the fetus.[2,3] In our case, there was no additional risk to the integrity of the pregnancy because the mother was in her third pregnancy, referred to a satisfactory parity. Though she had previous C-sections, there was not a risk of rupture in a new pregnancy because the size of the mini-hysterotomy (3 cm) does not increase the risk.

The only drawback of the report is that the patient was born prematurely to the expected casued by multifactoral conditions. However, she did not present complications inherent to a postnatal occipital encephalocele, completed her scheme of pulmonary maturation, and once the ideal weight was reached, she was discharged home without an overt neurologic deficit.

Fetal surgery requires the active participation of a multidisciplinary group.[1,2] Teamwork is indispensable, the fetal surgeon makes a timely initial diagnosis, plans the aces to the gravid uterus with the less number of complications and the pediatric neurosurgeon decides the treatment, surgical technique and the long-term follow-up of the patient.[2]

The performance of some critical steps, such as optimal localization of the hysterotomy, opening of a gravid uterus with minimal bleeding, fetal stabilization throughout the procedure, and intraoperative tocolysis before extubation, permits a successful evolution of the binomial.[3,4]

It is important to emphasize the necessity of ample experience with prenatal and postnatal neurosurgical procedures for neural tube defects.[3,4] Some authors recommend the use of transillumination of the hernial sac to identify and localize vascular or nervous structures within the cyst.[3]

Cavalheiro *et al*. suggest the placement of an absorbable miniplate between the bone and dura, with the theoretical objective of limiting the pressure on the defect and preventing the progression of the encephalocele. One of the advantages of carrying out the closure of the defect *in utero* is the easy manipulation of the hernial sac, which is smaller compared to that of the postnatal period, and the amount of neural tissue is less, allowing the reintegration into the cranial cavity.[3] However, the frail nature of the fetal tissues requires attention to the details.[2]

Cavalheiro *et al*. compare the results of patients who underwent fetal surgery versus patients operated on during the postnatal period, reporting that fetal surgery is associated with a better prognosis in cognitive development.[3,4]

The results are encouraging, particularly if the herniated neural content is below 20% of the total volume of the encephalocele.[2,3] The risk of rupture and associated complications decreases by limiting the exposure of the neural tissue to the dynamic environment of the uterine cavity. The long-term functional results seem superior to those of patients who undergo postnatal surgery.[2-4]

Progressive herniation *in utero* plays an important role in the development of the clinical prognosis.[5,8] Gadgil *et al*. documented in their cohort that progressive herniation is defined as an increment of >5% in the absolute value of neural tissue within the encephalocele.[5,9]

An additional investigation is necessary, including the use of a larger database and studies in animals to demonstrate the progressive hernia of functional neural tissue, which later presents damage within the uterine cavity, as well as the role it plays in the pathophysiology of an occipital encephalocele.

CONCLUSION

Open fetal surgery is a therapeutic option in the face of an isolated occipital encephalocele. This report demonstrates the viability of the surgical procedure by the adaptation of the postnatal technique to the prenatal surgery, with the aim of expanding the alternatives for the treatment of neural tube defects.

Further studies are needed to evaluate long-term functional results and demonstrate whether the procedure *in utero* plays an important role in the modulation of progressive herniation of neural tissue and in the prevention of postnatal complications such as hydrocephalus or seizures hard-to-control.

Ethical approval

Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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Commentary

The authors describe a case of occipital encephalocele that was operated in utero. Using their personal experience in fetal myelomeningocele (MMC) repair and the postnatal surgery of encephalocele, and following the pioneering work of Sergio Cavalheiro[2] in Sao Paulo, Brazil, this complicated surgery was performed successfully, for which the authors have to be commended.

However, some comments have to be made to put this case into proper perspective. Fetal surgery for MMC has a solid pathophysiological basis: direct exposure of the placode to the neurotoxic amniotic fluid leads to progressive damage to the neurons throughout the pregnancy, and persistent leakage of cerebrospinal fluid (CSF) leads to tonsillar herniation, which plays a role in the hydrocephalus seen in almost all children with MMC. Fetal surgery aims to reverse these factors. The fact that these objectives could be reached was shown in animal experiments before the use of the technique in humans. The randomized MOMS trial[1] finally gave scientific evidence that prenatal surgery is superior to postnatal surgery for children with MMC.

Occipital encephalocele is a different disorder: it is a closed defect without CSF leakage and exposure of the brain tissue to the amniotic fluid. The natural history during the fetal period is not well known, and although some cases are clearly progressive, others definitely are not. There is no clear definition of progression: a larger fluid volume without damage to the brain tissue, e.g., is not the same as progressive herniation of brain tissue. There are no good surgical animal models of encephalocele[3] that could be used to do preclinical studies. Evidence from the MOMS trial and other types of fetal surgery also illustrate the risks for the child inherent to the procedure: fetal demise in 1-4%, prematurity with all resulting complications in approx. 10%, etc. In conclusion, this surgical procedure for encephalocele is still experimental, and all the issues mentioned above have to be carefully considered before embarking on this kind of surgery in a fetus.

One other aspect also should be considered: 'fetal surgery' is not only fetal surgery but also maternal and uterine surgery. The risks to the mother are not negligible (chorioamniotic membrane separation, spontaneous rupture of membranes, uterine rupture, infection, and even maternal death), and future pregnancies are compromised. From an ethical point of view, it is very difficult to determine beneficence and non-maleficence when one has to consider both the mother and the fetus. As a minimum and in every case, not only the consent of the pregnant woman, who is in a vulnerable position and under a lot of emotional stress to make a decision but also the approval of a well-informed ethical review board should be sought before considering this surgery.

In summary, occipital encephalocele is a disorder for which more (pre)clinical studies are highly needed—the present case by Dr. García Méndez and his colleagues shows that fetal surgery for occipital encephalocele is feasible but does not offer scientific proof that this is the procedure of choice.

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