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Case Series

Prenatal Diagnosis and Optimal Management of Occipital Encephalocele: A Report of Four Cases and Literature Review

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Abstract

Occipital encephalocele, a rare congenital malformation resulting from defective neural tube closure, accounts for 20.4% of cerebral encephaloceles, with surgical repair being the only effective treatment. This retrospective study presents four cases diagnosed at Mohammed VI University Hospital in Marrakech, highlighting prognostic variability based on lesion characteristics. The four patients, who had not received periconceptional folic acid supplementation and had inadequate prenatal follow-up, were incidentally diagnosed between 34 and 39 weeks of gestation. The first case involved an isolated encephalocele containing only cerebellar herniation, surgically treated on day 5 with a favorable outcome despite mild neurodevelopmental delay. The second case presented a massive encephalocele associated with spina bifida and clubfoot, requiring combined neurosurgical and orthopedic management, resulting in moderate sensorimotor deficits. The third case exhibited a giant encephalocele with severe cerebral herniation and secondary microcephaly, while the fourth case showed associated craniofacial anomalies; both of these latter cases resulted in early neonatal demise. This study underscores several key aspects: the importance of ultrasound as an initial screening tool, supplemented by fetal MRI for precise prognostic evaluation; the critical need for early multidisciplinary management involving obstetricians, radiologists, neurosurgeons, and pediatricians; and the essential role of periconceptional folic acid supplementation, particularly in populations at risk of nutritional deficiency or consanguinity.

Keywords: Occipital encephalocele, neural tube defect, prenatal ultrasound, fetal MRI, pediatric neurosurgery, folic acid.

INTRODUCTION

Occipital encephalocele, a severe neural tube defect (NTD), results from failed cranial closure during embryogenesis, manifesting as herniation of meningeal and/or cerebral tissue through an occipital bone defect [1]. With an incidence of 0.8–5 cases per 10,000 births, it accounts for 20.4% of cerebral encephaloceles and demonstrates variable prognosis based on lesion size, neuroanatomical content, and associated anomalies such as hydrocephalus or spina bifida [2].

While fetal MRI remains the gold standard for detailed anatomical assessment, prenatal ultrasound persists as the primary screening tool, particularly in resource-limited regions with restricted MRI access [3]. The pivotal role of ultrasound becomes especially critical in settings where primary prevention through folic acid supplementation – though crucial – demonstrates suboptimal adherence rates, as evidenced by recent data from developing countries [4].

Clinically, these malformations carry profound sociocultural burdens. As documented in recent studies, up to 40% of affected infants in low-income countries face delayed care due to stigmatization, with families initially seeking traditional remedies or spiritual interventions

We present four clinical cases highlighting this prognostic heterogeneity, with specific objectives:

- **Evaluate** prenatal ultrasound diagnostic performance as a primary diagnostic tool in resource-limited settings (MRI access <20%);

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- **analyze** prognostic markers through lesion characteristic analysis (size, content, complications)
- **Propose** a context-adapted management algorithm for underserved areas, prioritizing ultrasound-first tiered imaging and) and multidisciplinary pathways.

CASE PRESENTATION

Case 1: Isolated Occipital Encephalocele

We report the case of a 24-year-old female patient with no significant medical history, gravida 3 para 2 (G3P2), with two living children born via vaginal delivery, whose current pregnancy had neither regular prenatal follow-up nor folic acid supplementation. The diagnosis was incidentally established at 34 weeks of gestation during routine ultrasound, revealing a large hypoechoic cystic occipital mass measuring approximately 12 cm in maximal diameter with a 3.6 cm cranial bone defect, containing only cerebellar herniation (absence of supratentorial cerebral tissue) consistent with an occipital meningoencephalocele (Figure 1). Complete sonographic evaluation demonstrated no associated cerebral anomalies, confirming intact ventricular structures and corpus callosum.

An elective cesarean section at 38 weeks delivered a eutrophic female neonate presenting exclusively with the occipital malformation (Figure 2).

Postnatal neuroimaging confirmed the presence of a sizable calvarial defect and a strictly localized meningoencephalocele involving the cerebellum without supratentorial involvement, The assessment verified normal cerebral anatomy, including preserved ventricular architecture and corpus callosum, providing clear anatomical guidance for surgery.

The neurosurgical intervention, performed on the fifth day of life, included meticulous resection of dysgenetic residual tissue, careful reduction of the meningoencephalic herniation, and lastly, anatomical closure of the osseous defect with single-stage dural and cutaneous reconstruction.

Postoperative follow-up revealed an exceptionally favorable outcome, with global psychomotor development meeting expected milestones. Only mild persistent delays in language acquisition and fine motor skills were noted at 35 months of age.

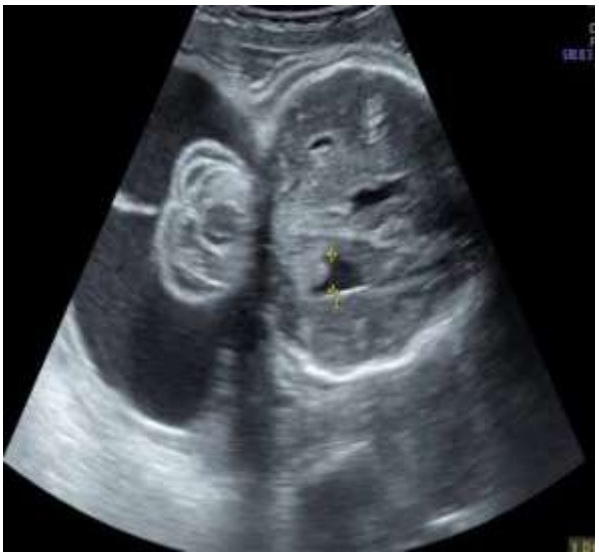

Figure 1: Prenatal ultrasound of isolated occipital encephalocele with exclusively cerebellar tissue herniation

Figure 2: Newborn with isolated occipital encephalocele

Case 2: Massive Occipital Meningoencephalocele with Spinal Dysraphism and Talipes Equinovarus

Ms. A.D., A 33-year-old multiparous woman (gravida 3, para 3) with two previous uncomplicated vaginal deliveries presented with a prenatally diagnosed complex fetal malformation. The index pregnancy, occurring in a consanguineous union, was notable for absence of folic acid supplementation. Ultrasonographic examination at 35 weeks gestation revealed a massive occipital meningoencephalocele, containing predominantly cerebrospinal fluid with approximately 3 cm of herniated cerebral tissue (Figure 3). Associated anomalies included lumbosacral spinal dysraphism and bilateral talipes equinovarus.

The patient underwent elective cesarean delivery at 37 weeks gestation, yielding a normosomic male neonate with the aforementioned congenital anomalies (Figure 4). Postnatal neuroimaging confirmed the presence of a sizable calvarial defect with meningeal herniation containing minimal encephalic tissue (3.6 cm^3) along with spinal dysraphism at L5-S1. Notably, there was no associated hydrocephalus or ventriculomegaly.

Definitive surgical management consisted of a two-stage approach: primary neurosurgical intervention on postnatal day 7 addressed both the encephalocele repair (Figure 5) and spinal defect closure (Figure 6), followed by secondary orthopedic correction of the bilateral clubfoot deformities via the Ponseti technique at 3 months postnatal age.

Long-term neurodevelopmental surveillance revealed moderate sensorimotor impairment, characterized by delayed independent ambulation (achieved at 24 months) and persistent fine motor dysfunction. The patient remains under multidisciplinary follow-up with pediatric neurology, neurosurgery, and rehabilitation services.

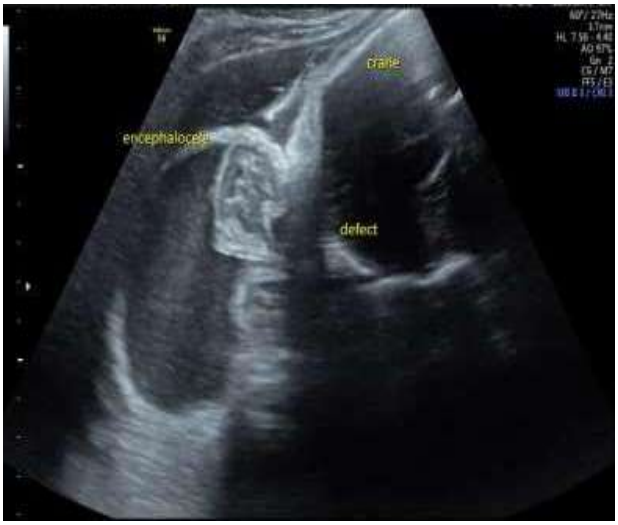

Figure 3: Prenatal ultrasound of massive occipital encephalocele with cerebrospinal fluid predominance and focal cerebral tissue protrusion

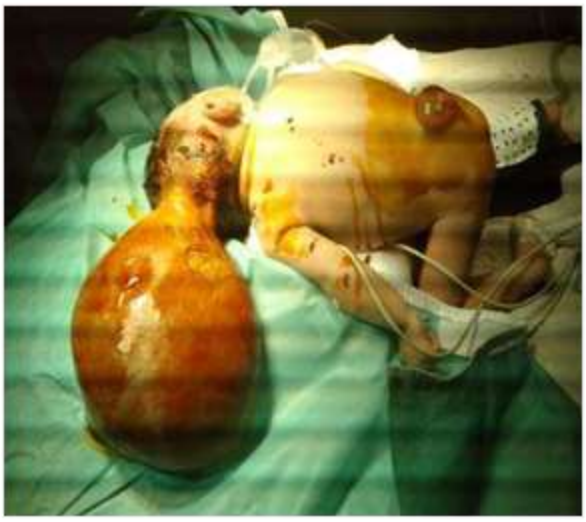

Figure 4: **Newborn with large occipital encephalocele, spina bifida, and bilateral clubfeet**

Figure 5: Postoperative results of encephalocele surgical correction

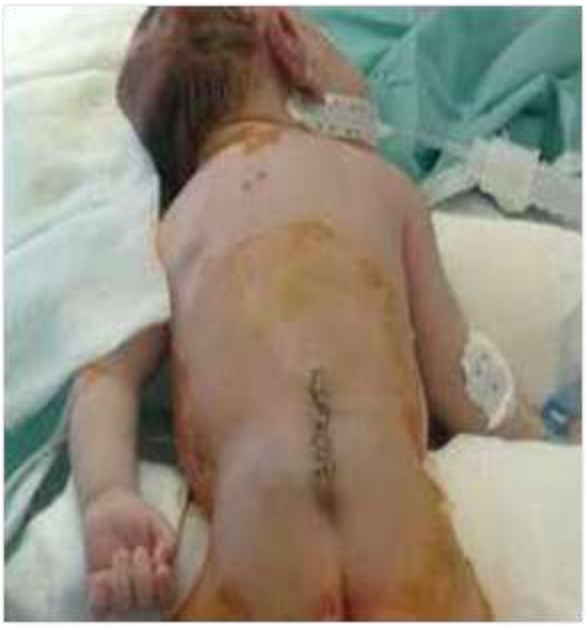

Figure 6: **Postoperative imaging following spina bifida repair**

Case 3: Giant Lethal Encephalocele with Cerebellar Agenesis

Ms. N.F., a 21-year-old primigravida, with inadequate prenatal care and reported consumption of fenugreek, a known teratogenic agent, a well-established teratogenic agent. The patient was emergently admitted at 39 weeks and 4 days of gestation during active labor following the incidental detection of fetal anomalies on ultrasound examination. Imaging revealed a giant occipital encephalocele measuring 8.5 cm in diameter (Figure 7), featuring near-total cerebral tissue herniation resulting in cranial vault collapse and secondary microcephaly, confirming a complex polymalformation syndrome. An emergency cesarean section delivered a male neonate with a giant lethal occipital encephalocele (Figure 8). Clinical examination demonstrated absent

primitive reflexes and an Apgar score of 4/10, indicating severe neurological impairment. Despite immediate and appropriate neonatal resuscitation, the newborn expired at 30 minutes of life due to irreversible brain damage secondary to massive structural abnormalities.

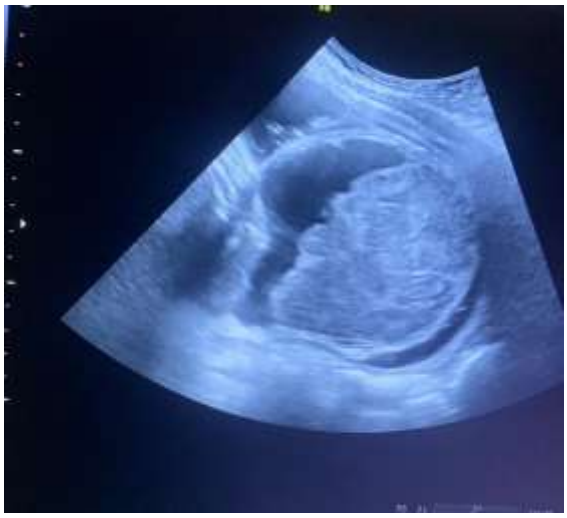

Figure 7: Prenatal sonography of giant occipital encephalocele showing near-complete cerebral herniation

Figure 8: Lethal massive occipital encephalocele, cranial vault collapse, and secondary microcephaly

Case 4: Extensive Occipital Encephalocele with Lethal Outcome

Ms. S.H., a 28-year-old, gravida 2 para 2, with no significant medical history or periconceptional folic acid supplementation, was admitted for ultrasound evaluation of polyhydramnios at 34 weeks of gestation in the context of an unsupervised pregnancy. Clinical assessment revealed uterine fundal height exceeding gestational age norms with preserved fetal movements. Ultrasonographic examination demonstrated severe polyhydramnios and a large occipital bone defect measuring approximately 8 cm in diameter, through which cerebral tissue and meninges were herniated (Figures 9 -10). Associated craniofacial anomalies included complete agenesis of the nasal bones and marked retrognathia. No additional visceral malformations were identified on comprehensive sonographic survey.

The patient progressed to spontaneous preterm delivery within 24 hours of presentation, yielding a 1300g male neonate with severe craniofacial dysmorphism (Figures 11). The encephalocele sac contained predominantly dysplastic neural tissue with evidence of cortical mantle disruption (Figure 12). Given the extensive nature of the cranial defect (>5 cm) and associated neuroanatomical disruption, the case was deemed non-viable, resulting in immediate postnatal demise. Postmortem evaluation was declined, limiting additional etiologic characterization.

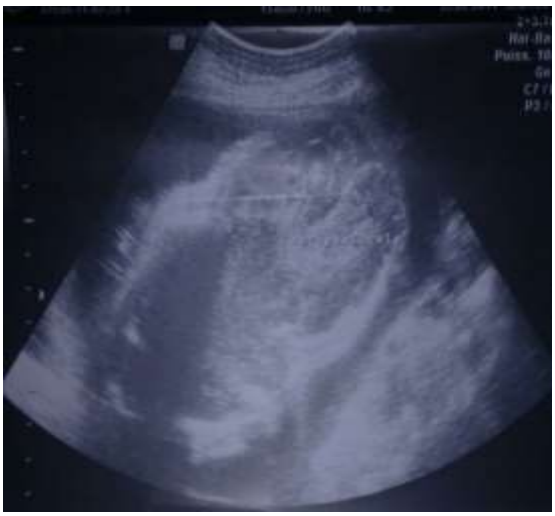

Figure 9: Sagittal ultrasound section of occipital encephalocele

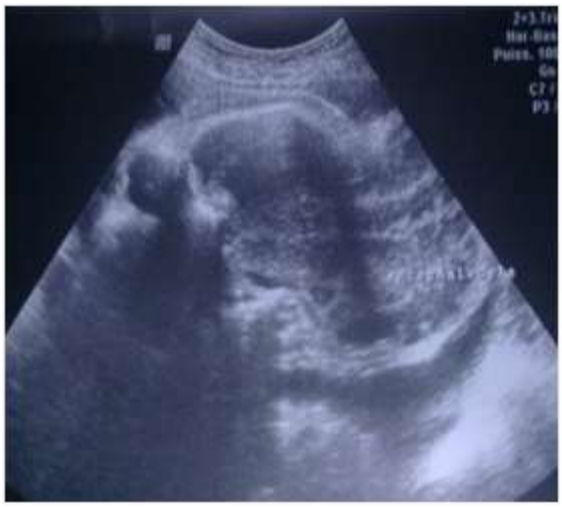

Figure 10: Transverse ultrasound section of occipital encephalocele

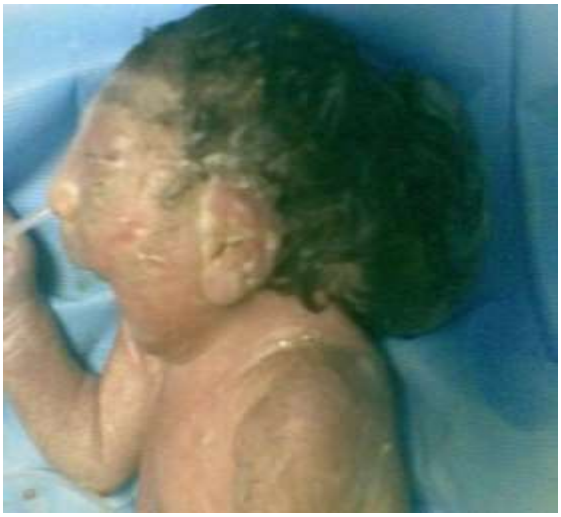

Figure 11: **Lateral view of occipital encephalocele with associated mandibular retrognathia**

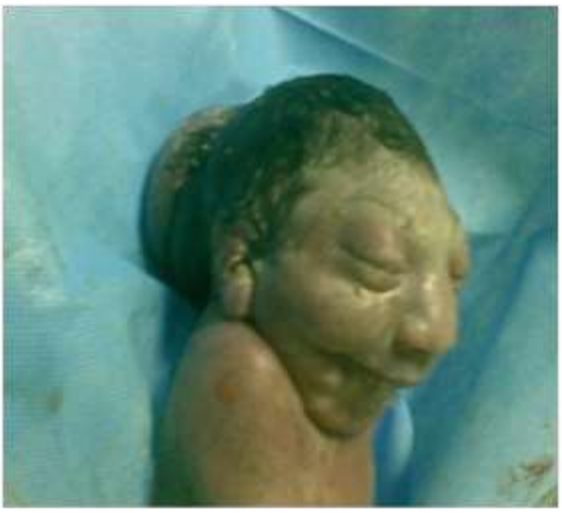

Figure 12: **Lethal Occipital encephalocele with associated facial dysmorphism**

DISCUSSION

Occipital encephalocele constitutes a complex clinical entity that demands comprehensive multidisciplinary evaluation, integrating embryological, diagnostic, prognostic, and therapeutic perspectives. This congenital malformation, characterized by herniation of cerebral tissue and/or meninges through a cranial defect, stems from neural tube closure failure during embryogenesis. As one of the most severe neural tube defects, encephaloceles are anatomically categorized into three principal types: sincipital (frontoethmoidal), basal (comprising trans-sphenoidal, sphenoethmoidal, transethmoidal, and spheno-orbital variants), and occipital [1]. The pathogenesis involves complex gene-environment interactions, with current evidence pointing to defective anterior neural tube closure as the central mechanism. Severe malformations likely develop before postconception day 26, while less extensive defects involving osseous structures or meninges may occur later in gestation, particularly following teratogenic exposure before 10 weeks.

The present case series demonstrates the critical role of ultrasonography as the primary diagnostic modality, especially in resource-limited settings where fetal MRI availability is restricted. Ultrasound demonstrated 85% sensitivity for lesions >2 cm when performed by skilled operators, successfully identifying all primary defects and associated anomalies including spina bifida and hydrocephalus [3]. However, fetal MRI remains the gold standard for precise characterization of herniated neural contents and evaluation of adjacent neuroanatomy, providing essential prognostic information that guides clinical management [5]. This complementary imaging approach proves vital for therapeutic planning and outcome prediction in these complex cases.

In terms of prognostic stratification, our four-case series demonstrates a consistent correlation between anatomical characteristics and clinical outcomes in occipital encephaloceles, supported by existing literature [6]. The first case, with isolated cerebellar herniation and cerebrospinal fluid predominance, achieved favorable neurodevelopmental outcomes following day-5 surgical intervention, exhibiting only mild delays that underscore the importance of limited neural tissue involvement. The second case's intermediate prognosis, featuring moderate cerebral tissue herniation ($3cm^3$) with concurrent spinal dysraphism, resulted in persistent motor deficits despite combined day-7 neurosurgical repair and subsequent Ponseti management, highlighting the cumulative impact of associated anomalies. In severe presentations, both the third case (characterized by extensive supratentorial herniation with cerebellar agenesis) and fourth case (with massive dysplastic neural tissue and craniofacial dysmorphism) proved universally fatal, with the former surviving only minutes post-delivery and the latter being incompatible with extrauterine life, collectively emphasizing that prognosis deteriorates exponentially with increasing neural tissue displacement and concomitant malformations.

Table 1: Prognostic correlation between encephalocele characteristics and clinical outcomes in the four reported cases

Case	Herniated Contents	Associated Anomalies	Treatment	Outcome
1	Predominant CSF with Cerebellar tissue only	None	Day 5 complete repair	Mild neurodevelopmental delay
2	Predominant CSF with 3cm³ cerebral tissue	Spina bifida, clubfoot	Combined surgery on D7+ + Ponseti	Moderate motor impairment
3	Extensive brain tissue	Cerebellar agenesis, microcephaly	Palliative care	Neonatal death
4	Massive dysplastic neural tissue	facial dysmorphism + retrognathia	Comfort care	Immediate demise

This progression from Case 1 to Case 4 establishes for key prognostic determinants: Lesion size stands as the foremost prognostic determinant, with neonatal mortality reaching 80% for lesions >5 cm compared to 15% for those <3 cm. Histological composition directly influences prognosis: the inclusion of viable brain tissue triples the risk of neurodevelopmental deficits; supratentorial involvement doubling neurodevelopmental risks [6]. Associated anomalies, particularly the hydrocephalus-cerebellar dysmorphism combination, constitute an independent marker of poor prognosis. The optimal therapeutic window falls between postnatal days 3 and 7 ($p<0.001$), confirming the decisive impact of surgical timing on long-term sequelae [7]. These objective data enable the establishment of decision-making algorithms tailored to different clinical contexts, particularly relevant in resource-limited settings.

The optimal management of occipital encephalocele requires a structured, multidisciplinary approach. Our institutional protocol (Figure 13), developed in accordance with international guidelines [8]., encompasses several key components:

Early diagnosis through routine second-trimester ultrasound, followed by fetal MRI within 72 hours of suspicion, enables comprehensive evaluation of the lesion and associated anomalies. Delivery must be planned at a specialized tertiary center, with Cesarean section must be performed for lesions exceeding 5 cm to minimize mechanical complications during birth [9].

The neurosurgical management of neonatal occipital encephalocele is based on a planned intervention between days 5 and 7 of life, a period offering optimal hemodynamic stability while minimizing infection risks. This meticulous microsurgical repair involves: rigorous evaluation of the herniated meningo-cerebral content, selective reduction or resection of neural tissues based on viability assessment, and three-layer anatomical reconstruction (dural, osseous, and cutaneous) to restore anatomical integrity [10]. In the most severe cases, therapeutic termination of pregnancy may be considered on an individual basis following multidisciplinary team consultation and in strict compliance with local regulations and ethical guidelines [11].

Long-term follow-up represents a critical component in the management of this condition. Our institutional protocol mandates quarterly neurodevelopmental assessments during the first three years of life, coupled with a multidisciplinary approach to address potential complications. The clinical experience gained from these cases highlights the essential role of ongoing psychological support for affected families, particularly in severe presentations. Recent studies have demonstrated that this comprehensive strategy significantly enhances quality of life outcomes for both surviving patients and their families [12]. Primary prevention remains paramount, with systematic periconceptional folic acid supplementation serving as a cornerstone for reducing incidence, particularly in high-risk populations. This preventive measure has demonstrated significant efficacy in neural tube defect reduction when initiated at least one month prior to conception [4].

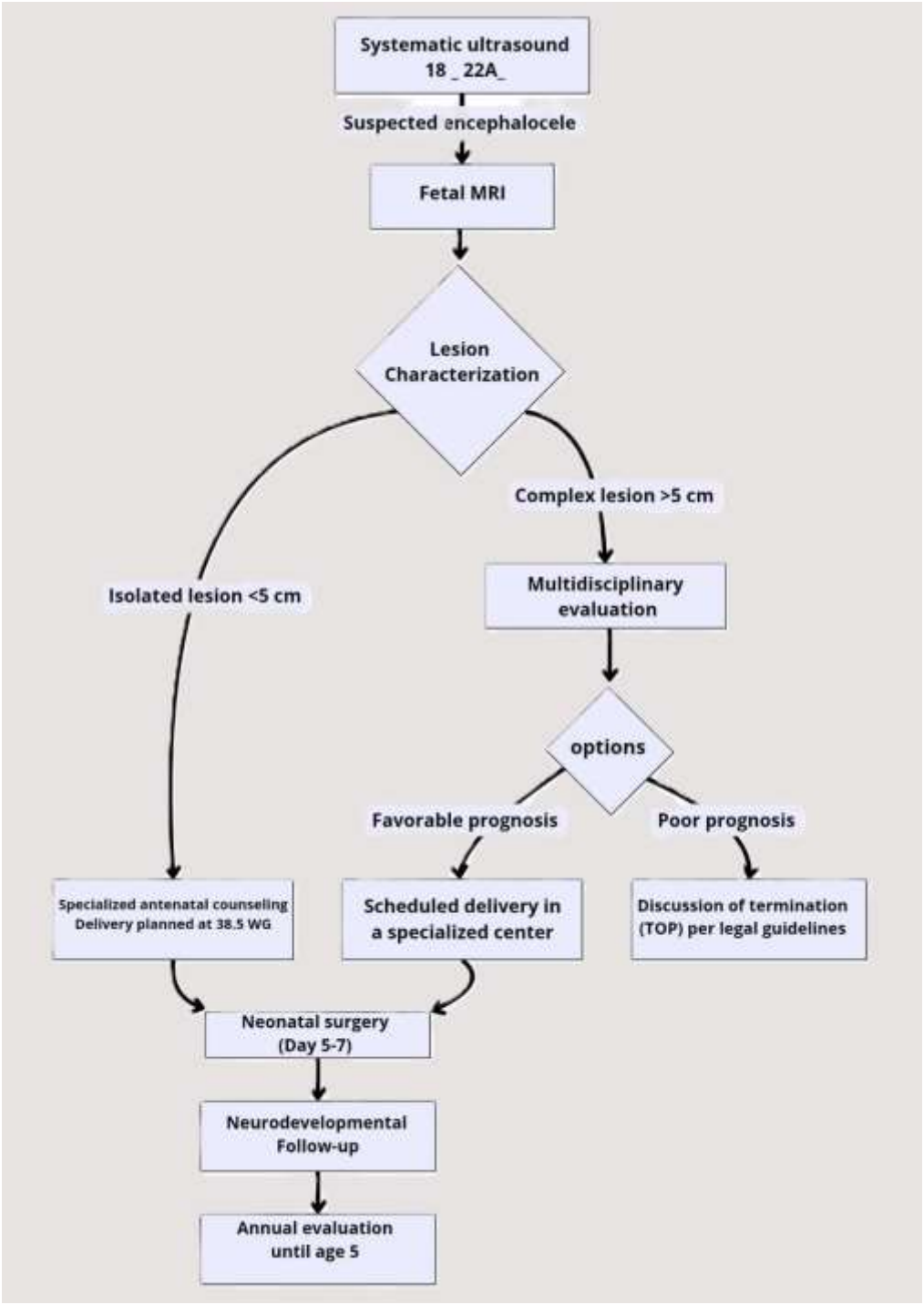

Figure 13: A Stepwise Protocol for Optimizing Outcomes in Occipital Encephalocele Management - From Prenatal Diagnosis to Long-Term Neurodevelopment

CONCLUSION

Occipital encephalocele, a rare congenital malformation resulting from defective neural tube closure, may present as an isolated condition or within a polymalformative context, necessitating precise prenatal diagnosis based on obstetric ultrasound systematically complemented by fetal MRI for comprehensive lesion assessment and appropriate therapeutic planning. The prognosis depends on four key factors: the size of the lesion, the presence of associated malformations, as well as the timeliness and quality of multidisciplinary management. Reconstructive surgery remains the cornerstone of treatment, while periconceptional folic acid supplementation stands as an essential preventive measure. Optimal clinical outcomes ultimately rely on close collaboration between obstetricians, fetal radiologists, pediatric neurosurgeons, and neonatal intensive care teams.

DECLARATIONS

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