Exploring the Intricacies of Encephalocele Case in Pregnancy: Unraveling Risk Factors, Diagnostic Difficulty, and Implications for Maternal and Fetal Health

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Abstract
Introduction: Encephaloceles are a type of neural tube defect that carries a significant risk of death and health complications. Ultrasonography can be utilized for the early detection of encephaloceles starting from 11 weeks of gestation. This case report describes the diagnosis of an occipital encephalocele using ultrasonography during the third trimester of pregnancy and the subsequent monitoring of the condition until delivery. This report serves as a reminder of the potential implications and the importance of ongoing research in prenatal diagnosis of encephalocele to improve diagnostic and therapeutic strategies.
Case Report: A 23-year-old woman, gravida 2, para 1 (G1P0A0), presented with lower abdominal pain without uterine contractions at 38 weeks of pregnancy, was consulted in the Department of Obstetrics and Gynecology of Slamet General District Hospital. She did nine antenatal visits; and three were performed through ultrasonography by a general practitioner. The occipital posterior defect was visualized with protruding mass consistent with an encephalocele during the third trimester when she was admitted to Slamet General Hospital Garut. A baby born by cesarean section with an encephalocele died three hours later.
Discussion: Encephaloceles can be detected using two-dimensional (2D) and three-dimensional (3D) sonography as early as 11 weeks; however, most instances are typically diagnosed during the second trimester. Using advanced imaging technology and skilled sonographers, an encephalocele can be diagnosed at 14 weeks.
Conclusion: Prenatal screening through ultrasonography and other advanced imaging techniques plays a significant role in identifying encephalocele prenatally. Understanding potential risk factors, including genetic predispositions and environmental influences, is important to better counsel patients on preventive strategies.

Keywords: Counseling, Diagnosis, Occipital Encephalocele, Risk Factors

Eksplorasi Kasus Kehamilan dengan Ensefalokel : Faktor Risiko, Kesulitan Diagnostik, dan Implikasi untuk Kesehatan Ibu dan Janin

Abstrak
Pendahuluan: Ensefalokel adalah jenis cacat tabung saraf yang membawa risiko kematian dan komplikasi kesehatan yang signifikan. Ultrasonografi dapat digunakan untuk deteksi dini ensefalokel mulai dari usia kehamilan 11 minggu. Laporan kasus ini menjelaskan diagnosis ensefalokel oksipital menggunakan ultrasonografi selama trimester ketiga kehamilan, dan pemantauan kondisi selanjutnya hingga persalinan. Laporan ini menjadi pengingat pada implikasi potensial dan pentingnya penelitian yang sedang berlangsung dalam diagnosis ensefalokel prenatal untuk meningkatkan strategi diagnostik dan terapeutik.
Laporan Kasus: Seorang wanita berusia 23 tahun, gravida 2, para 1 (G1P0A0), datang dengan keluhan nyeri perut bagian bawah tanpa kontraksi uterus pada usia kehamilan 38 minggu ke Departemen Obstetri dan Ginekologi di RSUD Slamet Garut. Pasien telah menjalani sembilan kali perawatan antenatal, tiga di antaranya dilakukan dengan ultrasonografi oleh dokter umum. Defek posterior oksipital divisualisasikan dengan massa yang menonjol konsisten dengan encephalocele pada trimester ketiga ketika admisi di RSUD Slamet Garut. Bayi lahir melalui operasi seksio sesarea dengan klinis encephalocele lalu meninggal tiga jam kemudian.
Diskusi: Encephalocele dapat dideteksi dengan sonografi dua dimensi (2D) dan tiga dimensi (3D) sejak usia 11 minggu, namun sebagian besar kasus biasanya didiagnosis selama trimester kedua. Encephalocele dapat didiagnosis pada usia 14 minggu menggunakan teknologi ultrasonografi, tetapi deteksinya didasarkan oleh kemampuan operator.
Kesimpulan: Skrining prenatal melalui ultrasonografi dan teknik pencitraan canggih lainnya memainkan peran penting dalam mengidentifikasi ensefalokel prenatal. Memahami faktor risiko potensial, termasuk faktor genetik dan lingkungan adalah penting untuk memberikan konseling yang lebih baik kepada pasien tentang strategi pencegahan.

Kata kunci: Konseling, Diagnosis, Encephalocele Oksipital, Faktor Risiko

Introduction

Encephaloceles are uncommon congenital anomalies characterized by incomplete fusion of the skull bones, forming a space through which cranial contents can protrude.[1]patterns of encephalocele, and its postsurgical results. Materials and Methods: The study was carried from year July 2012 to June 2015. Patients with encephalocele were evaluated for epidemiological characteristics, clinical features, imaging characteristics, and surgical results. Results: 20 encephaloceles patients were treated during the study period. Out of these 12 (60% This syndrome is a congenital anomaly of the mesoderm during the initial phases of embryonic development, resulting in the partial division between the surface ectoderm and the neuroectoderm. This disorder is defined by the herniation of the brain, with or without the herniation of the meninges, through a preexisting skull defect. The brain part that extends outside the skull is frequently enveloped by a layer of skin or a delicate membrane, creating the impression of a little pouch.[2] A few cases are acquired as a result of trauma, tumors, or iatrogenic injury. E flaws are classified into four categories according to the intracranial content that protrudes through the gap in the skull. The types mentioned are meningoencephaloceles, which are characterized by the herniation of cerebrospinal fluid (CSF), brain tissue, and meninges; meningoceles, which involve the herniation of the meninges and CSF; glioceles, which are cysts lined with glial cells and contain CSF; and atretic cephaloceles, which consist of dura, fibrous tissue, and degenerated brain tissue.[2] These anomalies are a type of neural tube abnormalities, alongside spina bifida and anencephaly.[1]patterns of encephalocele, and its postsurgical results. Materials and Methods: The study was carried from year July 2012 to June 2015. Patients with encephalocele were evaluated for epidemiological characteristics, clinical features, imaging characteristics, and surgical results. Results: 20 encephaloceles patients were treated during the study period. Out of these 12 (60%

Environmental and genetic factors have been associated with the development of encephalocele. TORCH infections (toxoplasma, rubella, cytomegalovirus, herpes simplex virus) have been implicated in numerous cases. Other factors implicated include consanguineous marriages and prior NTD pregnancies.[3]Adlyaman University, was performed between 2011 and 2015. Results: Among the study population, 19 mothers had been exposed to TORCH infections (toxoplasma, rubella, cytomegalovirus, herpes simplex virus Over 30 syndromes, such as Meckel-Gruber syndrome, Walker-Warburg syndrome, Fraser syndrome, Knobloch syndrome, Roberts syndrome, morning glory syndrome, and amniotic band syndrome, have been linked to encephalocele. However, it is still unclear how the incidence of encephalocele is correlated with maternal folate use.[4]

The incidence of encephaloceles has been recorded as 1–4 cases per 10,000 live births, with the occipital area being the most commonly affected location, representing 75% of all cases. The subsequent sites are the frontoethmoidal (13%–15%), parietal (10%–12%), and sphenoidal. Anterior encephalocele is predominantly seen in Southeast Asia, whereas occipital encephalocele is more prevalent in the Western world.[5,6]

Occipital encephalocele (OE) is defined by a swelling of varying sizes along the midline of the occipital bone. Prevalence is higher in females compared to males.[7] Typically, the size of the head is small.[8] The extent of a bony defect in the skull can range from a few millimeters to over 20 centimeters. In extreme cases, the sac might be larger than the newborn's head, which is a huge occipital encephalocele. According to research, 16% of children had OE (intestinal parasites) that

measured more than 20 cm. While the lesion may first appear as a standalone condition, it is frequently linked to other congenital defects or hereditary syndromes.[9]

The morbidity and mortality rates in patients with OE remain high despite the use of modern surgical techniques. These rates have ranged from 55% to 83%.[2] Low birth weight, numerous intracranial defects, and Black race are among the factors that contribute to an increased mortality rate. Nearly 76% of deaths occur on the first day of life, and approximately 71% survive to one year of age, with 67% surviving to 20 years of age.[4]

In recent years, development in advanced high-resolution imaging, appropriate surgical interventions, and comprehensive postoperative care have led to significant improvements. Only 2% mortality was seen in research that excluded patients with various abnormalities, big lesions, a significant amount of brain tissue in the sac that posed additional hazards for repair, and microcephaly.[10]

Encephaloceles can be detected using two-dimensional (2D) and three-dimensional (3D) sonography as early as 11 weeks. However, most instances are typically diagnosed during the second trimester. The prognosis, therapy, and management of encephalocele abnormalities are contingent upon the dimensions of the sac, its contents, and any additional related observations. This case report will detail the manifestation of a solitary occipital encephalocele with dilation cardiomyopathy diagnosed utilizing 2D sonography. Nevertheless, the occipital encephalocele was just identified during the third trimester, even though previous ultrasounds were performed.

Case Report

A 23-year-old woman gravida 2, para 1 (G1P0A0), presented with lower abdominal pain without uterine contractions at 38 weeks of pregnancy and was consulted in the Department of Obstetrics and Gynecology of Slamet General District Hospital for the first time during her pregnancy. She did nine antenatal cares; and three were performed through ultrasonography by a general practitioner. She has been taking folic acid and ferrous sulfate since the 17th week of gestation. She had no complications during pregnancy and did not report any other medication treatment. Her triple elimination laboratory test (HIV, syphilis, and Hepatitis B) was negative at 17 weeks of pregnancy. Her first child was born by vaginal delivery at term without any congenital malformation. No history of a genetic disorder or congenital anomaly was seen in the patient and family,

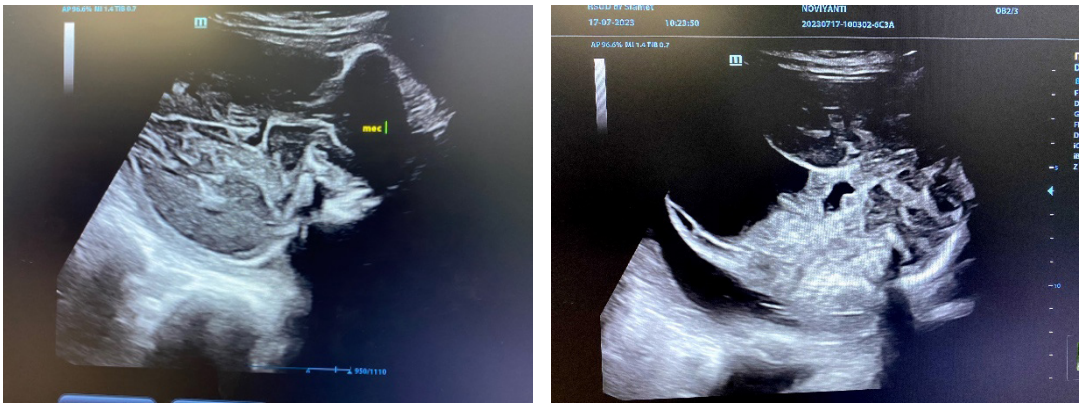

Figure 1 An Encephalocele Through the Occipital Bone Defect

and no consanguineous parents were identified.

Ultrasonography demonstrated a viable fetus measuring 38 weeks with cephalic presentation and estimated fetal weight (EFW) of 2615 grams. The occipital posterior defect was visualized with a protruding mass consistent with an encephalocele size 5cm x 5cm x 4cm (Fig. 1). A dilated cardiomyopathy was seen (Fig. 2), and polyhydramnios was found based on single deepest pocket (SDP) measuring 10.19 cm

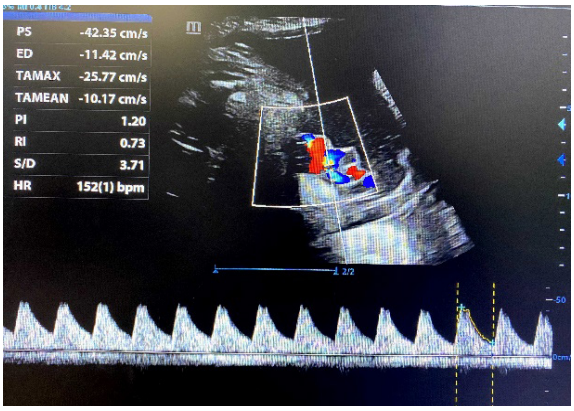

Figure 2 UItrasonography Findings of Dilated Cardiomyopathy

Figure 3 Clinical Pictures of the Neonates with Occipital Encephalocele

A cesarean section was performed due to the occipital position of the encephalocele on the fetus obstructing its delivery. An encephalocele measuring 5cm x 5cm x 4cm was observed. An infant delivered by cesarean section with an encephalocele passed away three hours after birth.

Discussion

Encephaloceles are categorized as a type of neural tube anomaly. These entities are distinguished by pedunculated growth composed of brain tissue that protrudes through an aperture in the dura mater and cranium. Cephaloceles refers to the protrusion of the brain or other intracranial structures through a skull defect. Nevertheless, the actual prevalence rate is elevated since numerous cases lead to the termination of pregnancy. The majority of encephaloceles are congenital or primary, while some might be acquired as a result of injury or trauma. Congenital encephaloceles are categorized based on their placement in the occipital, frontal, temporal, or parietal areas of the head.[11] In this case, the baby was diagnosed with occipital encephalocele.

Encephaloceles are linked to both genetic and environmental factors. Notable factors that have been documented include TORCH infections (toxoplasma, rubella, cytomegalovirus, herpes simplex virus) experienced at ages 3, 11, and 12, marriages between close relatives, and a history of the condition within the family.[3]Adlyaman University, was performed between 2011 and 2015. Results: Among the study population, 19 mothers had been exposed to TORCH infections (toxoplasma, rubella, cytomegalovirus, herpes simplex virus Like other neural tube defects, risk factors for this condition may include diabetes, obesity, hyperthermia, and insufficient levels of folic acid.[12] Pregnant women who come into contact with the Zika virus are at a higher

risk of experiencing abnormal development of the embryonic brain, which may involve the production of encephaloceles. Although encephaloceles can sometimes arise on their own, the majority (60%) of instances are accompanied by other congenital anomalies, such as cystic hygroma, single umbilical artery, amniotic band, Dandy-Walker defect, Arnold-Chiari defect, and hydrocephalus. Encephaloceles have been associated with more than 30 syndromes, including aberrant tissue band syndrome, Knobloch syndrome, Warfarin syndrome, and different chromosomal abnormalities. Encephalocele is commonly associated with Meckel-Gruber syndrome. The condition is distinguished by large polycystic kidneys, postaxial polydactyly, and an occipital encephalocele. Chiari III malformation is a congenital disorder marked by the incomplete development of the hindbrain, resulting in a specific type of brain and spinal cord herniation known as occipital/high cervical encephalocele and myelomeningocele. The exact positioning of the encephalocele and myelomeningocele can aid in differentiating between a diagnosis of Chiari III and a solitary encephalocele.[13] In this case, the possible underlying factors of encephalocele may be due to inadequate folic acid supplementation. The mother started taking folic acid supplementation at 17 weeks of gestation, which should be started before and during early pregnancy. This is because the early development of the fetal nervous system, including the neural tube, occurs in the first weeks of pregnancy, often before a woman realizes she is pregnant.

An encephalocele may be detected when a mass adjacent to the skull displays a pattern of folds similar to the brain, and a structural abnormality in the cranial bones is observed during sonography. Encephaloceles disrupt the regular flow of cerebrospinal fluid (CSF) and are frequently linked to hydrocephalus. Significant encephaloceles can be linked to microcephaly. The ability to visualize the defect with ultrasound may be limited by factors such as the size of the sac, inadequate positioning of the fetal head, or the location of the defect.[14] Nevertheless, sonography has been approximated to identify around 80% of all encephaloceles.[6] Most sonographic diagnoses are made during the second trimester, although in some instances, diagnoses can be made as early as 11 weeks.[14] In this case, the diagnosis was made during the third trimester based on ultrasonographic findings of a protruding mass from a defect in the occipital area measuring 5cm x 5cm x 4cm. Encephalocele is typically diagnosed postnatally. The prenatal diagnosis of encephalocele is conducted through ultrasound and maternal screening for serum and foetoprotein levels. The diagnosis is simply and confidently determined by the ultrasound results during the second trimester and can also be made during the first trimester. Moreover, Mhamdi et al. reported occipital encephalocele diagnosed during the third trimester (34 weeks of gestation) based on ultrasound results.[15] In 45% of the cases, the malformation was larger than 10 cm, and the average measurement was 4.75 cm.[16]

When assessing encephaloceles with sonography, it is crucial to ascertain the precise position of the skull defect, identify the internal components, and observe for any associated symptoms such as hydrocephalus. It is essential to show an intact fetal skull between the end of the first trimester and the beginning of the second trimester since this can help exclude the existence of encephaloceles. The hyperechoic appearance of the cranial bones is no longer visible in axial head imaging, indicating the presence of a bony defect known as an encephalocele. Using sagittal imaging planes of the fetal head can also assist in the diagnosis.[17] 3D sonography can enhance the diagnostic process and offer patients supplementary details, enhancing the quality of patient care.[6]

Fetal MRI is highly esteemed and

acknowledged as superior in detecting central nervous system problems found by ultrasonography. Fetal MRI has enhanced contrast resolution compared to ultrasonography and can produce high-quality images in scenarios where sonography may be limited, such as cases involving decreased amniotic fluid or artifacts caused by ossified bones. Fetal MRI is limited by the mobility of the fetus and its small dimensions hence it is typically not performed until 22 weeks of gestation. Fetal MRI provides high-resolution imaging of encephalocele contents and position, hence improving the precision of delivery planning for maximum safety.[18]

Several parameters are important for assessing the outcome in a patient with OE, including the extent of the malformation, the quantity and nature of the damaged brain components within the sac (such as the brainstem, occipital lobes, and existence of dural sinus), and the presence of hydrocephalus.[8] The patient's results are negatively associated with the quantity of brain parenchyma within the sac. Infants with abnormally large occipital encephaloceles with a substantial amount of brain matter contained within a sac that cannot be surgically corrected without the potential for causing neurological impairment have a bleak outlook, and there is limited certainty in predicting the child's lifespan.[10] Despite the utilization of contemporary surgical procedures, patients with OE continue to have elevated levels of morbidity and mortality. The rates have varied between 55% and 83%.[2]

The optimal counseling of this patient would have been possible if this defect had been detected prior to 20 weeks when the option of medical termination of pregnancy was available. Encephalocele can be diagnosed at 14 weeks using advanced imaging technology and skilled sonographers. In industrialized countries, such as those with facilities comparable to ours, in Indonesia, particularly in rural areas such as Garut residences, it is recommended to detect pregnancies up to 18 weeks, as the option of Medical Termination of Pregnancy (MTP) is readily accessible. Diagnosing during the first trimester is challenging because of the initiation of skull ossification at 10 weeks. Nevertheless, utilizing sophisticated technology like ultrasound equipment and highly competent sinologists, a conclusive diagnosis can be established before the 20th week, enabling the option of medical termination of pregnancy with the patient's consent.[19]

The shortcoming of this case report is that we did not have a history of the ultrasound examination performed by the general practitioner at an earlier antenatal visit. Ultrasound results at earlier antenatal visits may provide clues as to whether encephalocele is seen in second—and even first-trimester ultrasound results.

Conclusion

This case report underscores the complexities associated with the diagnosis and management of encephalocele during pregnancy, highlighting the critical need for early and accurate detection. The case demonstrates the significance of comprehensive prenatal screening and the role of advanced imaging techniques in identifying neural tube defects. Furthermore, it emphasizes the importance of understanding potential risk factors, including genetic predispositions and environmental influences, to better counsel patients on preventive strategies. The challenges encountered in the diagnosis and the subsequent decisions surrounding maternal and fetal health underscore the necessity of a multidisciplinary approach involving obstetricians, radiologists, and pediatric neurologists. This collaboration is essential to optimize outcomes, as it allows for tailored management plans that address maternal well-being and fetal prognosis.

Conflict of Interest

The authors declared there was no conflict of interest in this study.

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