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Case Series

Neural tube defects in prenatal diagnosis: A case series

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Abstract

Neural tube defects (NTDs) are among the most common congenital anomalies, arising from the incomplete closure of the neural tube during embryogenesis. These defects can lead to severe conditions such as anencephaly, spina bifida, and encephalocele, significantly impacting fetal morbidity and mortality. This case series presents four distinct instances of NTDs diagnosed during the second trimester of pregnancy: occipito meningoencephalocele, fetal acrania, Arnold-Chiari malformation type II, and semilobar prosencephaly. The series highlights the complexities of managing pregnancies complicated by NTDs, where medical termination of pregnancy (MTP) was the outcome in each case due to the poor prognosis associated with these conditions. Despite the well-established preventive benefits of folic acid supplementation, NTDs continue to occur, warranting the need for ongoing public health efforts to promote folic acid intake and improve prenatal care. These cases demonstrate that, while early diagnosis allows for informed decision-making, further research is needed to enhance preventive strategies and improve outcomes for those affected by NTDs.

Keywords: Neural tube defects, Prenatal diagnosis, Occipito meningoencephalocele, Fetal acrania arnold-chiari malformation, Semilobar prosencephaly.

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1. Introduction

Neural tube defects (NTDs) represent a significant class of congenital malformations resulting from the improper closure of the neural tube during embryogenesis. These defects can involve the brain, spine, or spinal cord and lead to conditions such as anencephaly, spina bifida, encephalocele, and others. NTDs are among the most common congenital anomalies, with a global incidence of approximately 1 in 1,000 pregnancies, though the incidence can vary depending on geographic, genetic, and environmental factors.[1]

The etiology of NTDs is multifactorial, involving a combination of genetic predispositions and environmental influences. Folate deficiency, for example, is a well-documented risk factor, and the introduction of folic acid supplementation has been shown to reduce the incidence of these defects.[2]

Prenatal diagnosis of NTDs has evolved significantly with advances in ultrasonography and other imaging modalities. These technologies allow for early detection, typically during the first or second trimester, enabling timely decision-making regarding the management of the pregnancy. In many cases, severe forms of NTDs are incompatible with life or are associated with significant neurological impairment, leading parents and healthcare providers to consider medical termination of pregnancy (MTP).[3]

This case series explores four instances of NTDs, each presenting with different forms of the defect. These cases include occipito meningoencephalocele, fetal acrania, Arnold-Chiari malformation type II, and semilobar prosencephaly. Each case illustrates the diversity of NTD presentations and the critical role of prenatal imaging in diagnosis.[4] The series also highlights the decision-making

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process regarding MTP, reflecting the severe prognosis associated with these defects and the necessity for comprehensive prenatal counseling.[5]

Through this series, we aim to create awareness of the importance of early and accurate prenatal diagnosis of NTDs, discuss the implications for clinical practice, and contribute to the broader understanding of these complex congenital conditions.

2. Case Presentations

2.1. Case 1: Occipito meningoencephalocele

A 23-year-old gravida 2, para 1, living 0 (G2P1L0), presented at 19 weeks and 6 days of gestation with an anomaly scan report indicating an anomalous fetus with occipito meningoencephalocele. The anomaly was first suspected during an NT scan, which suggested an encephalocele. A detailed anomaly scan confirmed the presence of a defect in the occipital region, measuring 1.5 cm, with herniation of the cerebellum and meninges through the defect, forming a hernial sac measuring approximately 4x4 cm with internal septations.

The patient had a non-consanguineous marriage of 1.5 years and no significant medical history, except for a past history of ileocecal tuberculosis treated the previous year. Given the poor prognosis associated with the defect, the decision was made to proceed with medical termination of pregnancy (MTP). The patient was administered 200 mg of mifepristone orally, followed by 400 mcg of misoprostol vaginally. The fetus was expelled with a birth weight of 332 grams, and was identified as female. (**Figure 1** and **Figure 2**) Examination revealed an occipital myelomeningocele, while the neck, trunk, and limbs were grossly normal.

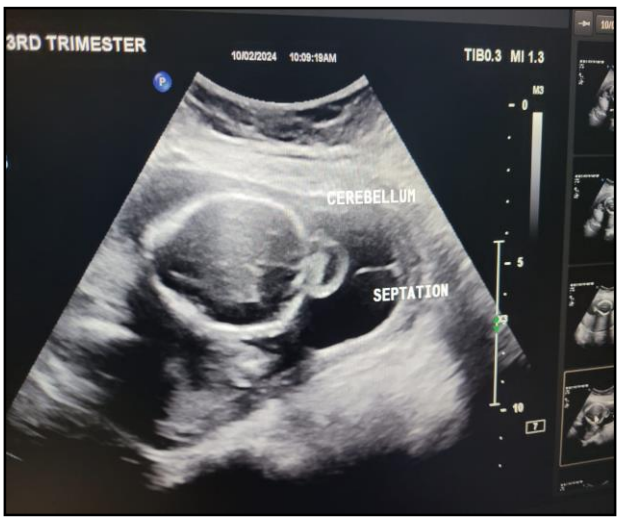

Figure 1: (USG-occipito meningoencephalocele)

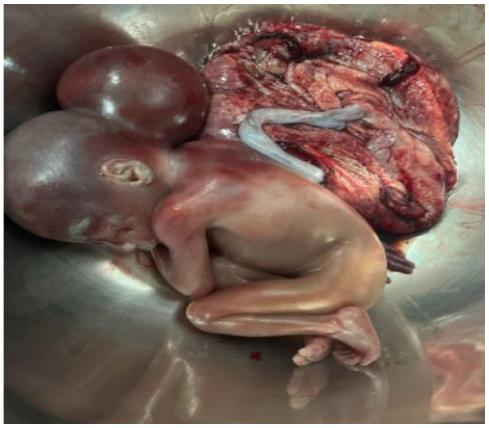

Figure 2: Occipito meningoencephalocele

2.2. Case 2: Fetal acrania

A 27-year-old (G2P1L1), at 14 weeks and 2 days of gestation presented for review with an NT scan showing features suggestive of fetal acrania. The scan at 11 weeks and 6 days indicated a crown-rump length (CRL) of 46 mm and a nuchal translucency (NT) of 2.7 mm, with diffusely reduced echogenicity of the calvarial bones, and the skull vault filled with fluid, raising a differential diagnosis of acrania versus hydranencephaly.

A follow-up scan at 14 weeks confirmed the diagnosis of fetal acrania. The patient opted for MTP, which was performed using 200 mg of mifepristone orally, followed by 400 mcg of misoprostol vaginally. The fetus, weighing 26 grams, expelled and was identified as male. The fetus exhibited the typical "frog-eye" appearance associated with acrania. (**Figure 3** and **Figure 4**).

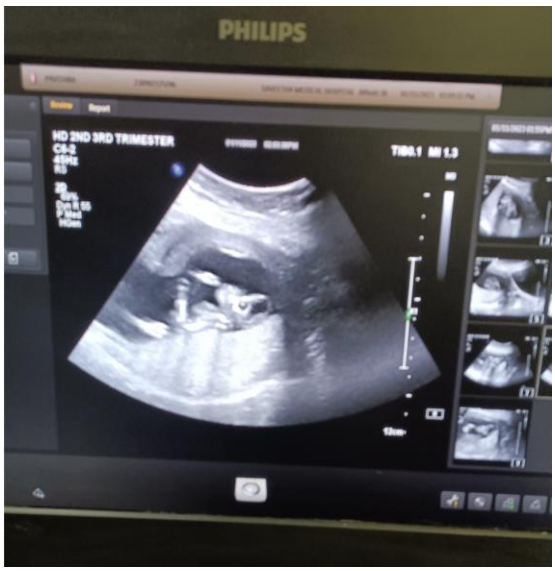

Figure 3: USG-Fetal acrania

Figure 4: Fetal acrania

2.3. Case 3: Arnold-chiari malformation type II

A 30-year-old (G2P1L1), with a history of a previous lower segment cesarean section (LSCS) presented at 19 weeks and 5 days of gestation with an anomaly scan revealing Arnold-Chiari malformation type II. The NT scan, performed earlier in the pregnancy, showed a measurement of 0.6 mm.

The anomaly scan at 18 weeks and 4 days revealed significant findings, including defects in the posterior elements of the spine with protrusion of neural contents within a cystic swelling, consistent with myelomeningocele. The scan also noted the "banana sign" in the cerebellum and a "lemon-shaped" skull, characteristic of this malformation. The ventricles appeared prominent with a dangling choroid sign, further supporting the diagnosis.

Given the severe nature of the malformation, the patient underwent MTP with 200 mg of mifepristone followed by 400 mcg of misoprostol vaginally. The fetus was expelled en sac with a birth weight of 252 grams. Examination of the fetus revealed a grossly normal head and myelomeningocele at the lumbosacral region.(**Figure 5**)

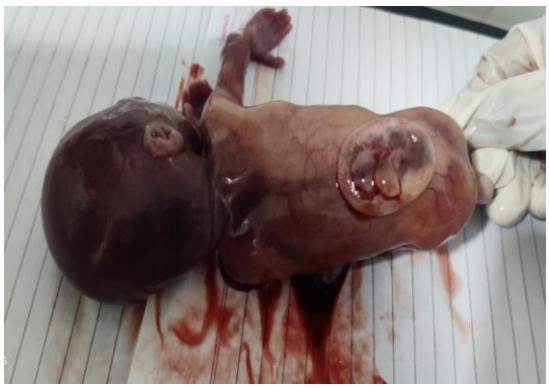

Figure 5: Arnold-chiari malformation type II

2.4. Case 4: Semilobar prosencephaly

A 32-year-old (G3P1L1A1), presented at 22 weeks and 4 days of gestation with an anomaly scan showing semilobar prosencephaly. The scan indicated the absence of the cavum septum pellucidum, along with microphthalmia, and other facial dysmorphisms, including a single nostril and unilateral anophthalmia. The anomaly scan provided a comprehensive assessment, confirming the diagnosis of semilobar prosencephaly with associated facial abnormalities. Given the poor prognosis and potential for severe neurodevelopmental impairment, the patient elected for MTP. She was administered 200 mg of mifepristone orally, followed by 400 mcg of misoprostol vaginally. The fetus, a male weighing 480 grams, was expelled (**Figure 6**, **Figure 7**).

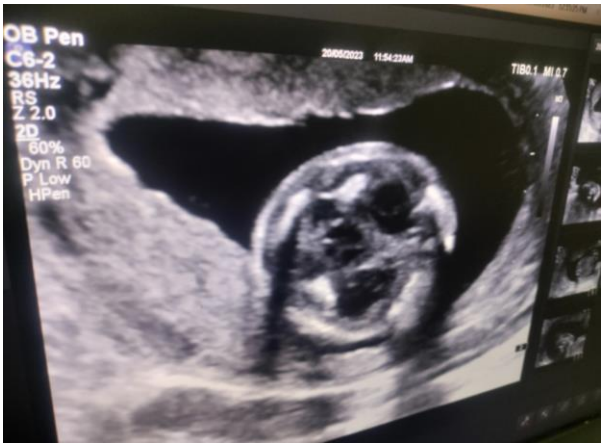

Figure 6: USG- semilobar prosencephaly

Figure 7: Semilobar prosencephaly

3. Discussion

This case series highlights the diversity and complexity of neural tube defects (NTDs) and their impact on prenatal care. Each case presents a unique manifestation of NTDs, demonstrating the importance of early and accurate prenatal diagnosis for effective management. The decision to proceed with MTP in each case tells the severe prognosis associated with these conditions and the need for timely and compassionate counseling.

Neural tube defects, such as those presented in this series, often carry significant morbidity and mortality. This highlights the ongoing need for research into additional preventive measures and early diagnostic techniques.[1]

The management of pregnancies complicated by NTDs requires a multidisciplinary approach involving obstetricians, genetic counselors, and pediatric specialists to provide comprehensive care and support to the affected families.[3] The outcomes of these cases further emphasize the importance of individualized care plans based on the specific type and severity of the defect, as well as the wishes of the patient and family.[5]

In all four cases, the diagnoses were made during the second trimester, allowing for informed decision-making regarding the continuation of the pregnancies. The use of high-resolution ultrasonography and the expertise of the healthcare team were instrumental in identifying these complex conditions early.[6]

A case series by Alwahab et al. documented several cases of occipito meningoencephalocele diagnosed in the second trimester via ultrasonography. The study highlighted the high mortality rate associated with this condition, especially when the herniated brain tissue is extensive. In most cases, pregnancies were terminated due to the poor prognosis. The study emphasized the importance of early detection through high-resolution ultrasound and MRI to assess the extent of the encephalocele and to guide clinical decisions.

Another study by R A Hamilton reported on the prenatal diagnosis of encephalocele and the subsequent management options, depiciting the role of multidisciplinary teams in counseling and managing these pregnancies. The findings supported early termination as the most viable option in cases with significant brain tissue herniation.[8]

Frey and Hauser conducted a study on the prenatal diagnosis of fetal acrania, where all cases were identified during the first or early second trimester using NT and anomaly scans. The study found that acrania is invariably fatal, and most cases resulted in spontaneous miscarriage or were medically terminated after diagnosis. The importance of early detection to prevent unnecessary maternal risk and emotional distress was highlighted.[9]

A study by Stoll et al. analyzed the outcomes of pregnancies diagnosed with fetal acrania, finding that early prenatal diagnosis allows for informed decision-making, with most parents opting for termination due to the lethal nature of the condition. The study also noted the potential for acrania to be associated with other anomalies, which should be evaluated through detailed imaging.[10]

A study by Gupta et al. focused on the prenatal diagnosis of Arnold-Chiari malformation type II and the associated neural tube defects, such as myelomeningocele. The findings indicated that most cases were diagnosed during the second trimester through detailed anomaly scans. The study showed that the prognosis for these fetuses was poor, with many developing severe hydrocephalus and requiring surgical intervention postnatally if the pregnancy was continued. However, in most cases, parents opted for termination due to the anticipated poor quality of life for the child.[11]

Another study by Gupta et al. reported on the outcomes of fetuses diagnosed with Arnold-Chiari malformation, emphasizing the role of fetal surgery in selected cases. However, the study found that even with in utero repair, many children still experienced significant neurological deficits, leading to the recommendation of termination in severe cases.[11]

A case series by Catherine et al. on semilobar prosencephaly highlighted the severe neurodevelopmental impairment associated with the condition. The study found that most cases were detected in the second trimester, with detailed ultrasound showing the characteristic midline defects. The prognosis was universally poor, with most affected fetuses either resulting in termination or neonatal death. The study reinforced the importance of early prenatal counseling to help parents understand the implications of the diagnosis.[12]

A study by Hahn et al. described the prenatal diagnosis and outcomes of semilobar prosencephaly, noting that the condition is often associated with facial dysmorphisms and other systemic anomalies. The research found that even with postnatal care, the survival rate was low, and most children had profound neurological impairments. The study supported early termination as a common outcome following diagnosis.[13]

The prevention of neural tube defects (NTDs) primarily hinges on adequate folic acid intake, which is critical during the early stages of pregnancy when the neural tube is forming. Folic acid, a synthetic form of vitamin B9, is essential for DNA synthesis, repair, and methylation, all of which are vital processes during embryogenesis. Numerous studies have demonstrated that periconceptional folic acid supplementation can reduce the risk of NTDs by up to 70%, making it the most effective preventive strategy available. Public health guidelines typically recommend that all women of childbearing age consume 400 to 800 micrograms of folic acid daily, starting at least one month before conception and continuing through the first trimester. In addition to supplementation, many countries have introduced mandatory folic acid fortification of staple foods, such as flour, to increase folate intake across the population and reduce the incidence of NTDs. Despite these measures, NTDs continue to occur, highlighting the need for ongoing public health efforts to promote awareness and adherence to folic acid guidelines.

Autopsy and genetic evaluation:

1. Case 1 (Occipito meningo encephalocele), the autopsy confirmed a defect in the occipital bone with herniation of the cerebellum and meninges. There

were no other visible abnormalities, which reassured the family that the condition was isolated.

2. Case 2 (Fetal acrania) showed the expected absence of skull bones with malformed brain tissue, consistent with the diagnosis. No other malformations were detected.
3. Case 3 (Arnold-Chiari type II malformation) confirmed spinal cord herniation through a lumbosacral defect and characteristic changes in brain shape. Again, no other systemic anomalies were found.
4. Case 4 (Semilobar prosencephaly), the autopsy revealed a midline brain fusion, a single brain ventricle, and characteristic facial features like a single nostril and absence of one eye. These findings explained the ultrasound features seen earlier.

These examinations gave families clarity and helped the clinical team confirm the prenatal diagnoses, which can be especially important when preparing for future pregnancies.

To further understand why these defects occurred, genetic testing was recommended. All four fetuses underwent karyotyping and chromosomal microarray analysis (CMA): The karyotypes were normal in all cases.

Through these steps, families received not only medical explanations but also emotional closure, and were offered genetic counselling to guide them in future pregnancies with reassurance and confidence.

4. Conclusion

This case series emphasizes the critical role of early prenatal diagnosis in the management of neural tube defects (NTDs). Through detailed imaging and timely detection, healthcare providers can offer comprehensive counseling and guide families through difficult decisions, often leading to medical termination of pregnancy in cases with poor prognosis. Despite advances in prenatal care and the implementation of folic acid supplementation and food fortification programs, NTDs remain a significant cause of fetal morbidity and mortality. The diversity in presentation, as demonstrated in these cases, highlights the complexity of NTDs and the need for continued research and public health efforts to further reduce their incidence. This series also reinforces the importance of a multidisciplinary approach to managing pregnancies affected by NTDs, ensuring that families receive the support and care needed during such challenging times. As prenatal screening technologies continue to advance, early and accurate diagnosis will remain pivotal in improving outcomes for affected families.

5. Authors Contributions

Dr. Roselin Sheela J and Dr. Rajalekshmi Murugan**:** All authors contributed to the preparation of the manuscript, approved the final version, and agree to be accountable for all aspects of the work.

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None.

7. Conflict of Interest

None.

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