

A Fatal Antenatal Diagnosis: Case Report of Anencephaly

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Cite this paper as: Dr Aparna Sarwade, Dr Radhika Dhedia, (2025). A Fatal Antenatal Diagnosis: Case Report of Anencephaly. *Journal of Neonatal Surgery*, 14 (21s), 429-431.

ABSTRACT

Anencephaly is a fatal neural tube defect characterized by the absence of the calvarium and major portions of the brain, resulting from failed closure of the cranial end of the neural tube during early embryogenesis. Despite declining prevalence rates in recent decades due to folic acid supplementation and food fortification programs, it remains a significant cause of perinatal mortality. We report a case of anencephaly diagnosed in a 21-year-old primigravida at 20 weeks of gestation. The pregnancy was the result of a second-degree consanguineous union, with no significant personal or familial history of congenital anomalies. Ultrasonographic evaluation revealed absence of the calvarial vault, disorganized brain tissue, and spinal anomalies, consistent with acrania-exencephaly sequence and cervical spina bifida. After appropriate counselling and informed consent, medical termination of pregnancy was carried out using mifepristone and misoprostol. Post-delivery, the foetal specimen was sent for karyotyping, which revealed a normal chromosomal profile. This case underscores the importance of early prenatal screening, particularly in populations with genetic risk factors. Anencephaly, being incompatible with life, necessitates timely diagnosis and sensitive, multidisciplinary counselling to aid parental decision-making. Public health measures, especially periconceptional folic acid supplementation, have proven highly effective in reducing the incidence of neural tube defects. Preventive strategies, including folic acid supplementation and genetic counselling, are vital in reducing the burden of anencephaly. Early detection through routine antenatal care allows for informed clinical decisions and better psychological support for affected families

Keyword: *Acrania, Karyotyping, Folic acid supplementation*

1. INTRODUCTION

Anencephaly is a severe congenital malformation characterized by the absence of the calvarium and the majority of cerebral structures, resulting from the failure of closure of the rostral (cranial) end of the neural tube during embryogenesis. This condition is part of a broader category known as neural tube defects (NTDs), which also includes spina bifida, arising from the failure of closure at the caudal (lower) end of the neural tube.

The prevalence of anencephaly has shown a declining trend in the United States. According to data from the National Centre for Health Statistics, the prevalence in 2001 was reported at 9.40 per 100,000 live births. [1] Between 1996 and 2001, there was a 22% decline in anencephaly cases, coinciding with public health initiatives promoting folic acid supplementation and fortification of food products. (2)

Folic acid plays a critical role in the prevention of NTDs. Studies have demonstrated that periconceptional supplementation with folic acid can reduce the incidence of NTDs by up to 72%. In response to such evidence, mandatory folic acid fortification of certain food products was implemented in the United States in 1998, leading to significant reductions in NTD prevalence. (3)

Despite these advancements, anencephaly remains a condition with a poor prognosis. Affected fetuses are often stillborn or die shortly after birth. Prenatal diagnosis through ultrasonography allows for early detection, enabling healthcare

providers to offer appropriate counselling and management options to expectant parents.



Ongoing public health efforts aim to further reduce the incidence of NTDs through education on the importance of folic acid supplementation before and during pregnancy, as well as continued monitoring of fortification programs' effectiveness

Case Report

A 21-year-old primigravida, married in a second-degree consanguineous union, presented at 20 weeks of gestation based on last menstrual period, with a first trimester ultrasound dating the pregnancy at 19 weeks and 3 days. She was referred following an anomaly scan which raised suspicion of a foetal malformation. The patient had no significant past medical, surgical, or familial history of congenital anomalies or genetic disorders.

On general examination, the patient was found to be vitally stable. Per abdominal examination revealed a uterine fundal height corresponding to 20 weeks of gestation. The foetal lie was longitudinal, with the foetal back oriented to the maternal left side, and the foetal heart rate was auscultated at 120 beats per minute, which is within normal limits.

Per vaginal examination showed a closed and uneffaced cervical os, indicating no signs of labour.

A detailed ultrasonographic evaluation revealed a single live intrauterine foetus with significant structural anomalies. The most notable findings included the absence of an ossified calvarial vault with herniation of disorganized brain tissue into the amniotic cavity, consistent with acrania–exencephaly sequence, which is recognized as a precursor to anencephaly, a severe neural tube defect. Additionally, the ultrasound suggested spinal deformities, specifically spinal dysraphism, most likely cervical spina bifida, further supporting the diagnosis of a neural tube defect. There was also a suspicion of associated congenital heart anomalies.

Given the severity and non-viability associated with these foetal anomalies, and following appropriate counselling and obtaining informed consent, a decision was made to proceed with medical termination of pregnancy. Termination was initiated using misoprostol and mifepristone induction protocol as per institutional guidelines.

The foetal specimen was collected post-delivery (figure 1 and figure 2) and sent for karyotyping to identify any underlying chromosomal abnormalities that could provide insight into the aetiology and guide future reproductive counselling and was found to be normal.



Figure 1 Anencephalic fetus from ventral aspect



Figure 2 Anencephalic fetus from Dorsal aspect

2. DISCUSSION

Anencephaly is a critical congenital condition that poses significant challenges for both the patient and healthcare providers. The diagnosis is typically made through prenatal ultrasonography, which reveals the absence of the cranial vault and brain tissue. This condition is incompatible with life, and affected fetuses are either stillborn or die shortly after birth.

Aetiology and Risk Factors: The exact cause of anencephaly is not fully understood, but it is believed to result from a combination of genetic and environmental factors. Risk factors include:

- **Genetic predisposition:** A family history of neural tube defects increases the risk.

- **Nutritional deficiencies:** Lack of folic acid before and during early pregnancy is a significant risk factor.
- **Environmental exposures:** Certain medications, uncontrolled diabetes, and exposure to high temperatures in early pregnancy can increase the risk.

Prevention: Folic acid supplementation has been shown to significantly reduce the incidence of neural tube defects, including anencephaly. It is recommended that all women of childbearing age take 400 micrograms of folic acid daily, starting at least one month before conception and continuing through the first trimester [\[4\]](#) [\[5\]](#).

Genetic Counselling: Genetic counselling is crucial for parents who have had a child with anencephaly. It provides information about the risk of recurrence in future pregnancies and helps parents make informed decisions. The recurrence risk for neural tube defects in subsequent pregnancies is approximately 2-5% [\[6\]](#) [\[7\]](#).

Management and Ethical Considerations: The management of anencephaly involves a multidisciplinary approach, including obstetricians, genetic counsellors, and neonatologists. Ethical considerations play a significant role in decision-making, particularly regarding the continuation or termination of the pregnancy. In this case, the decision for medical termination was made after thorough counselling and informed consent.

3. CONCLUSION

Anencephaly is a severe and often fatal neural tube defect that occurs during embryonic development. Research has shown that the incidence of anencephaly can be significantly reduced through preventive measures, particularly folic acid supplementation. It is recommended that all women of childbearing age, especially those planning to conceive, take a daily supplement of folic acid. This simple yet effective measure can greatly reduce the risk of anencephaly and other neural tube defects.

Regular follow-up with healthcare providers is crucial to ensure adherence to these preventive measures. Through these efforts, we can hope to decrease the incidence of anencephaly and ensure healthier outcomes for both mothers and infants.

Genetic counselling provides expectant parents with information about the risk of recurrence in a future pregnancy. This is particularly important for parents who have had a child with anencephaly, as they may be at a higher risk of having another child with a neural tube defect.

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