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A Case of Anencephaly with Concomitant Maternal Overweight: Clinical Presentation and Outcome

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ABSTRACT
Anencephaly, a lethal neural tube defect (NTD), results from failed cranial neural tube closure during early embryogenesis. Maternal risk factors, including obesity and inadequate periconceptional folic acid intake, are well-established contributors. This report describes a case involving a 29-year-old primigravida with a Body Mass Index (BMI) of 28.03 kg/m² (overweight) who was referred at 39 weeks gestation for delivery following a prenatal diagnosis of anencephaly. The patient had no personal history of chronic illness but had not taken folic acid supplements prior to conception, initiating prenatal vitamins only after pregnancy confirmation. A cesarean section delivered a male infant with classic features of anencephaly. Despite resuscitation, the newborn died shortly after birth. The development of anencephaly in this instance is associated with two key modifiable risk factors: maternal overweight status and the absence of preconceptional folic acid supplementation. Neural tube closure occurs within the first month of pregnancy, often before pregnancy recognition, making folic acid initiation after confirmation too late to prevent NTDs. Maternal overweight or obesity is an independent risk factor that can compromise the fetal environment and potentially alter folate metabolism. This case underscores the critical importance of preconception health counseling, emphasizing the necessity of folic acid supplementation for all women of reproductive age, commencing before conception. This public health measure is particularly crucial for women with elevated BMI.

Keywords: anencephaly, maternal body mass index, clinical outcome

INTRODUCTION

Anencephaly is one of the most severe congenital anomalies of the central nervous system, resulting from the failure of neural tube closure during the third to fourth week of embryonic development (Andonotopo et al., 2025; Sen & Jangra, 2023). This condition leads to the absence of most brain structures and skull bones. Anencephaly is characterized by the underdevelopment of the major portions of the skull and essential brain tissue; infants born with this condition typically have an extremely poor prognosis, with a mortality rate approaching 100% within the first year of life (Lwin et al., 2022; Dickman et al., 2016).

The global prevalence of anencephaly varies, with an average reported incidence of approximately 1 to 3 cases per 10,000 live births (Ekmekçi & Gençdal, 2019; Dickman et al., 2016). In some regions, such as Turkey, the prevalence can be as high as 2.1 per 10,000 live births (Ekmekçi & Gençdal, 2019). Research indicates that demographic, environmental, and genetic factors, as well as the availability of adequate healthcare services for early detection and management of the anomaly, influence its prevalence (Alabbad et al., 2023; Munteanu et al., 2020).

Various risk factors have been identified in the occurrence of anencephaly (Abebe et al., 2021). One of the most significant causes is folic acid deficiency during the preconceptional

period and early pregnancy; studies show that inadequate folic acid intake can increase the likelihood of neural tube defects, including anencephaly (Iglesias-Vázquez et al., 2022). Furthermore, maternal medical conditions such as body mass index (BMI), diabetes, and obesity are also associated with an increased risk of anencephaly in offspring (Razaz et al., 2017; Zhang et al., 2021). This case highlights critical aspects concerning maternal health, prenatal care, and the implications of congenital anomalies such as anencephaly, particularly in the context of the mother's BMI and nutritional practices (Brunner et al., 2025; Koletzko et al., 2019).

A well-established modifiable risk factor for neural tube defects (NTDs) is inadequate maternal folate status before and during early pregnancy (Bhide, 2021; Martinez et al., 2023). Folate is essential for DNA synthesis and cellular division, and its deficiency during neural tube development disrupts proper closure (Wang et al., 2023). However, another critical and increasingly prevalent risk factor is maternal overweight and obesity (Portela et al., 2015). In Southeast Asia, including Indonesia, the rising prevalence of overweight among women of reproductive age poses an additional challenge to prenatal health. Maternal obesity (BMI $\geq$ 30 kg/m^2) and overweight (BMI 25–29.9 kg/m^2) are associated with a 1.5- to 2-fold increased risk of NTDs, independent of folate intake.

The underlying mechanisms linking high BMI to NTD risk are multifactorial. Obesity is associated with a chronic inflammatory state, insulin resistance, and altered levels of adipokines, which may interfere with embryonic development (Gasmi et al., 2021). Additionally, obese women often have lower circulating folate levels due to reduced absorption or increased metabolic demand, creating a functional folate deficiency despite adequate intake. This "folate resistance" hypothesis suggests that higher doses of folic acid may be required to achieve protective serum levels in overweight and obese women.

Despite these known risks, public health messaging in many regions—including Indonesia—often does not sufficiently emphasize the combined impact of BMI and folate timing. Many women begin folic acid supplementation only after confirming pregnancy, which is typically after neural tube closure has occurred. This gap is especially critical in overweight women, who may need targeted preconception counseling.

This case report highlights the confluence of maternal overweight and absent preconception folic acid supplementation in a patient with an anencephalic pregnancy. It underscores the need for regionally aware, proactive preconception care—particularly in settings where nutritional and weight-related risk factors are increasingly common. By detailing this case, we aim to reinforce the importance of tailored public health strategies to reduce the incidence of preventable NTDs in susceptible populations.

Method

Case Report

A 29-year-old female patient, Mrs. IR, G1P0A0 at 39/40 weeks of gestation, was referred from the obstetrics polyclinic to the delivery room for a planned termination of pregnancy due to a congenital anomaly, Anencephaly. The patient became aware of the congenital anomaly in this pregnancy at a gestational age of 3 months and 2 days. Termination of the pregnancy was initially planned at 20/21 weeks of gestation; however, the patient wished to continue the pregnancy. Upon arrival at the obstetrics polyclinic, the patient denied any complaints.

The patient has no personal history of diabetes mellitus or hypertension. However, her family history is significant for both diabetes mellitus and hypertension in her mother. There is no family history of a similar congenital anomaly. During the pregnancy, the patient denied taking any medication other than prenatal vitamins prescribed by her obstetrician. Prior to conception, she had not taken any supplements, including iron or folic acid. Upon confirmation of her pregnancy, she began a regular regimen of prenatal supplements, including folic acid, calcium, and Viliron (a multivitamin supplementation). This is her first pregnancy with no previous history of miscarriage.

Physical examination revealed a good general condition with a clear consciousness (compos mentis). Anthropometric status showed a weight of 70.8 kg, a height of 159 cm, and a Body Mass Index (BMI) of 28.03 kg/m². Vital signs were as follows: blood pressure 128/85 mmHg, pulse 98 beats per minute, respiratory rate 22 breaths per minute, and temperature 36.8 °C. The general physical examination was within normal limits.

Based on the obstetric examination, the uterine fundal height was 28 cm, located midway between the xiphoid process and the umbilicus. Leopold maneuvers revealed the following: L1 suggested the fetal breech at the fundus, L2 palpated a large, firm mass consistent with the fetal back on the mother's left side, and L3 identified a soft, ballottable mass in the lower uterus, consistent with the fetal head. L4 confirmed that the head was not yet engaged in the pelvic inlet. The fetal heart rate was 135 beats per minute, and no uterine contractions were present. An obstetric check revealed no cervical dilation. Laboratory findings were within normal limits, showing a white blood cell count of 8.35 x 10^3/µL, red blood cell count of 3.86 x 10^6/µL, hemoglobin of 12.40 g/dL, and platelets of 205 x 10^3/µL, with a random blood glucose level of 121 mg/dL.

A supporting ultrasound investigation was performed, revealing a single live fetus with biometry corresponding to a gestational age of 34/35 weeks and an estimated fetal weight (EFW) of 2000 grams; the fetus was diagnosed with Anencephaly, characterized by incomplete formation of the brain parenchyma, along with a posterior, grade 3 placenta and adequate amniotic fluid volume. Based on the history, physical examination, and these investigational findings, the working diagnosis for this case was a primigravida (G1P0A0) at 39/40 weeks of gestation with a Term Pregnancy complicated by a Congenital Anomaly (Anencephaly), where the last ultrasound estimated a fetal weight of 2210 grams. Consequently, the pregnancy was terminated via cesarean delivery.

The infant was delivered via cesarean section at a gestational age of 39/40 weeks (term), in face presentation, and was male. His birth length was 40 cm with a weight of 2210 grams. Vital signs at birth included a heart rate of 92 beats per minute, oxygen saturation of 65%, and an APGAR score of 4 at 5 minutes. Resuscitation was initiated, including the administration of an adrenaline injection, which resulted in an improvement of the heart rate to 135 beats per minute and oxygen saturation to 90-91%. The infant's care was continued in the Neonatal Intensive Care Unit (NICU), where an orogastric tube (OGT) was placed (**Figure 1.**). The parents were educated regarding the infant's high-risk condition and acknowledged the possibility of the worst outcome. After approximately one hour, the infant experienced desaturation and a drop in heart rate, leading to death.

Figure 1. Anencephaly infant in NICU
Source: Author's Medical Documentation, 2025

Result and Discussion

Anencephaly is recognized as one of the most severe and lethal forms of central nervous system malformations, with prognosis for survival beyond the neonatal period being extremely limited. Prenatal detection of anencephaly typically occurs via ultrasound, enabling informed decision-making regarding pregnancy management (Dickman et al., 2016).

Initial recommendations for termination at 20 to 21 weeks were consistent with clinical guidelines; however, the patient's decision to continue the pregnancy illustrates the complex emotions and ethical dilemmas that patients face when confronted with such diagnoses. Studies indicate that the rate of termination for diagnosed anencephaly can exceed 80% in certain populations, though the precise figures can vary depending on regional attitudes towards termination (Aggarwal; Zielinski).

The patient's BMI is assessed at 28.03 kg/m², categorizing her as overweight, which may contribute to various obstetric complications. Maternal obesity is associated with an increased risk of neural tube defects (NTDs), including anencephaly, due to the potential effects of obesity on maternal metabolism and the fetal environment. Specifically, obesity can influence the availability of nutrients critical for fetal development, while excessive maternal weight may diminish the efficacy of prenatal vitamin intake, particularly folic acid (Youssouf et al., 2022; , Endalifer & Gedefaw, 2020).

Folic acid supplementation is notably important, as it has been evidenced to significantly reduce the risk of NTDs when taken preconceptionally (Berhane & Belachew, 2022; Petersen et al., 2019). In this case, Mrs. IR did not begin taking folic acid until confirmation of her pregnancy and had not supplemented before conception, which is critical given that the neural tube closes within the first month of pregnancy (Andonotopo et al., 2025). This delay in intervention may have played a role in the development of the congenital anomaly observed in her fetus. Research indicates that failure to use folic acid preconceptionally is associated with a higher incidence of NTDs (Petersen et al., 2019; Manimekalai et al., 2024).

Moreover, while the mother did incorporate prenatal vitamins into her regimen during gestation, the absence of folate prior to conception poses significant risks. Evidence suggests that the timing and adequacy of folic acid intake are crucial; all women of childbearing age are encouraged to consume at least 400 µg of folic acid daily, but those at higher risk may benefit from increased dosages (Petersen et al., 2019; Malik & Malik, 2025).

Additionally, while this case notes no family history of congenital anomalies, genetic

factors also contribute significantly to the incidence of NTDs. Environmental influences such as maternal health behaviors, including nutrient intake and overall wellness, further illustrate the multifactorial nature of such defects. A systematic review delineates that various determinants—including maternal obesity, nutritional status, and genetic predisposition—are interconnected with the outcomes of pregnancies complicated by NTDs like anencephaly (Manimekalai et al., 2024).

CONCLUSION

The interplay between BMI, nutritional habits, and genetic and environmental factors critically influences fetal health outcomes. In the case of Mrs. IR, her preconception overweight status combined with inadequate folic acid intake likely contributed to the diagnosis of anencephaly. This highlights the urgent need for comprehensive prenatal care that prioritizes preconception health, particularly focusing on weight management and timely folic acid supplementation to reduce the risk of congenital anomalies. Future research should explore effective region-specific interventions and strategies to improve preconception nutritional education and weight control among women of reproductive age to prevent neural tube defects more effectively.

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