

CASE REPORT

Anencephaly: A case report and the crucial role of folic acid supplementation and early diagnosis

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Received: February 23, 2024;

Accepted: April 22, 2024;

Published: April 26, 2024.

Citation: Mutumbua GK, Numbi BL, Kiakuvue YN, et al. (2024). Anencephaly: A case report and the crucial role of folic acid supplementation and early diagnosis. *Theory and Clinical Practice in Pediatrics*, 5: 121-124. <https://doi.org/10.25082/TCPP.2024.01.002>

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Abstract: Anencephaly is a congenital malformation caused by neural tube failure, which can be managed through folic acid intake during pregnancy. Diagnosis is typically prenatal, and termination of pregnancy is often recommended. A newborn born at Kolwezi paediatric clinic was born with a visible congenital malformation. The mother had a low socio-economic background and a 41-year-old. An ultrasound scan revealed a progressing pregnancy at 30 weeks' gestation with polyhydramnios. No antenatal care monitoring or folic acid prophylaxis were implemented during the periconceptional period and throughout the pregnancy. Anencephaly diagnosis was confirmed, and the newborn died five minutes post-birth. Preventive folic acid treatment has shown a decrease in neural tube defects, and first-trimester ultrasound scans can help identify life-incompatible congenital malformations. Advocating for legislative measures and genetic counseling is crucial to educate parents on recurrence risks and the advantages of folic acid supplementation before subsequent pregnancies.

Keywords: anencephaly, folic acid supplementation, antenatal diagnosis, Kolwezi

1 Introduction

Anencephaly is a congenital malformation of the fetal nervous system, resulting from a defect in the closure of the neural tube. The fetus is left with a portion of the encephalon (brain, cerebellum, and brain stem) missing. The prevalence ranges from 1 in 1000 to 1 in 2000, with variations influenced by different genetic backgrounds in various populations and dietary factors, including folic acid deficiency [1]. The condition typically affects the fetus between the 3rd and 4th week of amenorrhea, stemming from a disruption in neurulation [2].

First-trimester ultrasound can diagnose almost all cases of anencephaly and should be performed by a specialist or staff with appropriate training [3]. In developed countries, medical termination of pregnancy has been routine for all congenital malformations incompatible with life, including anencephaly, since the 1990s, leading to a reduction in the mortality rate to zero in these regions [4]. It is essential to note that newborns with anencephaly generally have a life expectancy of a few hours to a few days after birth and often present with disorders such as deafness, blindness, insensitivity to pain, and unconsciousness. Preventing congenital malformations of the fetal nervous system by supplementing folic acid before conception and during the first three months of pregnancy could enable couples to avoid the tragedies mentioned above [2, 3, 5].

In the Democratic Republic of the Congo (DRC), a strategy has been developed to prevent this type of malformation. It involves systematically administering folic acid to all pregnant women during their antenatal visits and conducting at least four ultrasounds, including one in the first trimester, throughout their gestation.

The aim of this study is to demonstrate the importance of folic acid in the prevention of neural tube defects, including anencephaly, and the role of first and second trimester obstetric ultrasound in the early diagnosis of the disease.

2 Case report

A newborn baby, female, was born in the maternity ward of the Kolwezi pediatric clinic with a visible congenital malformation. In the maternal history, we noted a low socio-economic level,

maternal age of 41 years, P6G7, with no consanguinity. The mother had a single antenatal visit in the 3rd trimester, where an ultrasound scan revealed a progressive singleton pregnancy of 30 weeks' amenorrhea with polyhydramnios. There was no history of folic acid intake. However, she did not return for the recommended follow-up appointment. She presented 4 weeks later with intense lumbo-hypogastric pain, 7 cm cervical dilation, and 90% effacement. The premature delivery at 34 weeks' amenorrhea resulted in the birth of a malformed live infant weighing 1900g. The infant exhibited hypothermia at 35 degrees Celsius, bradycardia at 61 beats per minute, altered consciousness, absence of archaic reflexes, axial and peripheral hypotonia, and respiratory distress with desaturation at 65%. Morphologically, there was a partial absence of the cranial vault with brain matter protrusion, upward-facing orbits, prominent superciliary arches, bulging eyes, a short neck, and low-set ears resembling mongolism (Figure 1). The diagnosis of anencephaly was confirmed without associated malformations. Despite resuscitation attempts with suction and oxygen therapy, no blood count was obtained, and the newborn unfortunately did not survive more than 5 minutes after birth.



Figure 1 Anencephaly with partial absence of the cranial vault

3 Discussion

The term “neural tube defects” (NTDs) refers to all visible malformations resulting from a neural tube closure defect of variable extent during the 4th week of embryonic development. They are the most common central nervous system malformations and the most frequent congenital malformations, followed by cardiac malformations [6]. Anencephaly is one of these and the most severe of the neural tube closure disorders, uniformly unilateral in the neonatal period, and is one of the most lethal of the congenital malformations [6, 7]. Epidemiological studies of NTD have led to suspicions of certain etiologies. Geographically, these are well known but poorly understood, as the incidence of NTD varies greatly from one region to another. For example, the highest incidence in Ireland and Wales is 40 times higher than the lowest incidence in Colombia. The prevalence in Ireland is 7 to 8 per 1,000 births, while it is 1 to 2 per 1,000 in the United States and continental Europe. In the United States, prevalence decreases from north to south. These variations cannot be explained by racial factors alone, as rates among immigrants are intermediate between those in their country of origin and those in the host country [7, 8]. In a systematic review and meta-analysis encompassed twenty-four studies conducted by Oumer et al. [9], comprising a total of 4,963,266 births. The combined birth prevalence of anencephaly in Africa was calculated at 0.14% (95% CI: 0.12, 0.15%). Notably, Ethiopia (0.37%, CI: 0.15, 0.58%), Algeria (0.24%, CI: 0.24, 0.25%), and Eritrea (0.19%, CI: 0.19, 0.19%) exhibited a higher burden of anencephaly. Studies encompassing both live births and stillbirths (0.16%) and those conducted after 2010 (0.25%) demonstrated a higher pooled prevalence of anencephaly. Conversely, countries with mandatory folic acid fortification showed a lower burden (0.05%) of anencephaly [9].

In Lubumbashi (DRC), a study of congenital malformations and their prevention revealed that among all congenital malformations, nervous system malformations were the most prevalent, with an incidence of 2.029 per 1,000 births, and their prevention was associated with folic acid intake [10]. In the United States, the highest incidence was noted in Caucasians, with a rate of 2.8 per 1000 compared to 2.1 per 1000 for other races [11]. The newborn described in our case is of African descent.

According to numerous studies, anencephaly affects more female than male children, with no

clear explanation for this phenomenon. Furthermore, the prevalence of anencephaly is higher among spontaneously aborted fetuses than among those born at term, suggesting in utero selection against fetuses with this malformation [10]. In our case, the newborn was female. Maternal age and parity are also risk factors; studies on the influence of maternal age on the occurrence of NTD are contradictory. Multiparity is also a risk factor [12]; in our case, the mother was multiparous. Anglo-Saxon studies have shown a high frequency of NTD in lower socio-economic backgrounds, which could be linked to nutritional deficiencies, particularly in folic acid, thus favoring the occurrence of NTD [10]. In our case, the woman came from a low socio-economic background, and was therefore at risk of folic acid deficiency. This risk was compounded by the fact that she did not attend antenatal clinics, which could have compensated for these deficiencies by the systematic administration of folic acid.

In Bangui (in Central African Republic), an anencephalic newborn was observed, a case that had not been diagnosed prenatally despite ultrasound scans being performed in the 1st, 2nd, and 3rd trimesters [13]. In the case studied, an ultrasound scan was performed in the 3^e trimester but unfortunately did not reveal anencephaly. This underscores the importance of proper pregnancy monitoring, with morphological ultrasound scans conducted by qualified professionals to ensure precise prenatal diagnosis of congenital malformations, thereby reducing postnatal discoveries of these malformations that are typically identifiable antenatally. Such prenatal diagnosis would have facilitated better organization of care, including potential medical termination of the pregnancy, as could have been the case for our patient. The lack of diagnosis may also be attributed to the absence of alpha-fetoprotein testing in maternal serum and the failure to follow up on antenatal consultations, as seen in our patient's case.

Anencephaly is associated in 50 percent of cases with rachischisis and in 30 percent of cases with a visceral malformation [7]. This hypothesis is challenged by an observation from Central Africa, where no associated pathology has been found in cases of anencephaly [13]. In the newborn described in our observation, no associated malformations were found. On the other hand, the presence of polyhydramnios in our case, observed on ultrasound in the 3rd trimester, is justified by the fact that, at the end of pregnancy, there is a swallowing problem in fetuses with anencephaly [7]. The treatment of anencephaly is preventive and is now based on therapeutic termination of pregnancy. It seems that this option has become commonplace since the 1990s in many developed countries, bringing the anencephaly mortality rate down to zero [4]. In the DRC, no steps have been taken in this direction. Regarding the prognosis for anencephaly, survival at birth is only a few minutes to a few days. Jaquier et al. [5] in a 2006 study found that 6 out of 211 cases survived a maximum of 6-28 days. In our case, the newborn lived only 5 minutes after birth.

The primary prevention of neural tube closure anomalies (anencephaly being one of the most severe forms) by taking folic acid during the periconceptional period has proved effective not only in preventing a possible recurrence after the birth of a first affected child or fetus but also in reducing the risk of these malformations occurring in the general population [4]. In our case, the mother of the newborn was not taking folic acid prophylaxis. As per recommendations, we would like to emphasize that neural tube closure is an early event in embryogenesis; anterior closure occurs at around 26-27 days [2]. It is crucial to acknowledge that if closure does not occur within the normal timeframe, it is impossible to repair this anomaly at a later stage. Therefore, it is necessary to have an adequate intake of folic acid at the very start of pregnancy, a time when patients often do not yet know they are pregnant. Beyond this critical period, folic acid no longer impacts the occurrence of NTDs. Hence, it is advisable to suggest supplementation before pregnancy to ensure a sufficient folate supply and correct intakes at the beginning of pregnancy, before it is recognized. Supplementation is no longer necessary to prevent NTDs beyond the first trimester of pregnancy [12, 14]. Practically, at-risk patients should take 4 mg of folic acid per day, and patients with no specific history should take 0.4 mg per day, ideally before becoming pregnant, to ensure adequate intake at the start of pregnancy.

4 Conclusion

Folic acid plays a crucial role in preventing the occurrence of NTDs. It is essential to raise awareness among practitioners and women of childbearing age about the importance of folic acid supplementation before conception and throughout the first trimester of pregnancy to prevent recurrences or the development of anencephaly. Early antenatal diagnosis of anencephaly provides parents with valuable information and allows for the consideration of medical termination of pregnancy. Legislation on medical termination of pregnancy is necessary to prevent psychological distress for patients who may be facing the birth of severely malformed infants.

Conflicts of interest

The authors declare that they have no conflict of interest.

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