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Anencephaly: A Case Report of a Preventable Congenital Neural Tube Defect (NTD)

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Abstract

Anencephaly is a congenital malformation of the central nervous system involving the neural tube. It is a lethal condition with a 100% mortality rate. The condition can be diagnosed antenatally, usually in the first trimester, by ultrasound. This case report is about a 34-year-old woman with a long history of alcohol consumption, who never attended antenatal care and ended up delivering a stillborn female baby with anencephaly. It focuses on the need for early antenatal care booking, preconception and antenatal folic acid supplementation, avoidance of alcohol during pregnancy and preconception diabetic control, and the importance of first-trimester anomaly ultrasound.

Keywords

NTD, Anencephaly, Antenatal, Neural Tube, Folic Acid, Ultrasound, Alcohol, Preconception, Congenital Malformation, Diabetes

1. Introduction

Anencephaly is a severe congenital malformation of the central nervous system resulting from the failure of closure of the cranial (rostral) end of the embryologic neural tube. It is a rare malformation representing approximately 40% of all neural tube defects (NTDs). Lower prevalence of this condition is observed in countries with mandatory folic acid fortification. The etiology of anencephaly has a multifactorial origin that includes nutritional deficiencies (*i.e.*, folate deficiency), chromosomal abnormalities, environmental teratogens and certain medications, and toxins [1]. The anomaly is lethal, and pregnancy outcomes are either miscarriage, stillbirth or early neonatal death. Since there is no curative treatment, health

care is mainly focused on prenatal diagnosis, counselling, supportive care, and ethical decision-making. The anomaly can be diagnosed in the first trimester by using an ultrasound. Once the diagnosis has been confirmed, appropriate management, once agreed upon by the managing obstetrician and the parents, can be instituted.

2. Case Summary

A 34-year-old G5P4 woman presented to our hospital labour ward with a history of on and off abdominal pains for the past 6 hours. On interrogation, it was noted that she did not know her last menstrual period and she had not attended any antenatal care. Therefore, she never received any preconception or antenatal folic acid supplementation and had not had any obstetric ultrasound.

She also admitted to having been drinking alcohol before and during pregnancy. She started drinking alcohol when she was an adolescent, but she could not specify the exact age. She admitted to having been drinking almost on a daily basis during pregnancy. She had no known comorbidities and her hemoglobin test (HGT) and glycated hemoglobin ($HBA1_C$) were normal and no one in her family or close relatives has ever delivered a child with anencephaly. On evaluation, she was found to be in the second stage of labour but a foetal heart could not be picked up on cardiotocograph (CTG) tracing. The second stage of labour was complete within minutes and she delivered a female fresh stillborn weighing 2200 grammes, shown in Figure 1. Characteristically, the baby had no cranium and brian tisue, and had a "frog-like" face.

Figure 1. Photographs of a female baby, born lifeless with anencephaly, weighing 2200 grams.

3. Discussion

Anencephaly is a variety of neural tube defects (NTD) that usually occurs in the first trimester. In this condition the cerebrum and cerebellum are absent and the affected fetuses are either miscarried, born still, or die soon after delivery. It is a rare form of congenital neural tube malformation, only second to spina bifida. The global prevalence of this malformation is about 1 in 1000 pregnancies [2], and

it is more common in females than in males. The prevalence of anencephaly in Africa is reported to be 0.014 per 1000 births [3], with Ethiopia having the highest burden of the condition, with an incidence of 23.70 per 10,000. Other African countries with high prevalences of anencephaly are Algeria, and Eritrea [4].

Nutritional lack of folic acid or conditions that lead to reduced folic acid at the time of conception and through the first trimester have been found to be risk factors for anencephaly. Although studies have shown that immediate family members are more at risk suggesting a genetic inheritance, in the index case, there was no history of anencephaly in the immediate family members or close relatives. Uncontrolled diabetes, maternal obesity, alcohol consumption during pregnancy, and some medications, especially the folate antimetabolites like sodium valproate (an antiepileptic drug), have also been linked to the development of anencephaly [5]. Uncontrolled diabetes (hyperglycemia) interferes with the expression of essential genes, such as Pax3, leading to increased cell death (apoptosis) in the neural folds, preventing proper closure of the skull and forebrain. There is a threefold ratio of incidence of anencephaly in infants of diabetic mothers compared with those of non-diabetic ones [6]. Alcohol consumption during pregnancy, especially during the first trimester, is also associated with anencephaly through a different pathway. Alcohol reduces the production of retinoic acid, a compound derived from vitamin A, that is essential for the development of facial and central nervous system structures, thus disrupting the gene regulation that controls the timing and sequencing of these developmental steps [7]. In this index case, it was found that the mother was a chronic alcohol drinker, she drank alcohol during pregnancy and never attended antenatal care, hence, she never received any folic acid supplementation, which is usually offered to pregnant women in antenatal clinics.

NTDs may first be suspected from an elevated maternal serum alpha-fetoprotein (MSAFP) screening test. MSAFP screening during the second trimester of pregnancy is an effective screening tool for identifying the vast majority of cases of anencephaly in women with or without a positive family history or other risk factors for neural tube defects [8]. Confirmation of the diagnosis is then done by using ultrasound, although in most cases ultrasound is the first intervention due to the routine ultrasound examinations in modern obstetrics. High-quality ultrasound detection of anencephaly, however, depends on the professional level of the technician or physician performing the ultrasound, the position of the fetus and the gestational age, etc.

Sonographic diagnosis of anencephaly: The vault of the skull is composed of flat bones (frontal, parietal, occipital), sutures (coronal, sagittal, lambdoid) and fontanelles (anterior and posterior). The main function of the vault is to protect important structures such as the cerebral hemisphere and cerebral blood vessels [9]. The ossification of the fetal skull is normally completed after 11 weeks of gestation. Ultrasonographic diagnosis of anencephaly, therefore, is only possible from the 11^{th} - 12^{th} week of pregnancy. Early prenatal ultrasound diagnosis is essential to initiate counselling of the mother/couple in favor of termination of a

pregnancy with an anencephalic fetus as the condition is not compatible with life [2]. However, although women are advised on early antenatal booking, most of them, especially in Low-income countries, either start their antenatal care late or not at all. Important ultrasound findings in anencephaly include:

- Absent calvarium (cranial vault) above the orbits: it is an obvious sign seen in both sagittal and axial ultrasound views, where the skull bones covering the brain are absent (no echogenic skull bones)
- Presence of exposed or disorganized neural tissue (area cerebrovasculosa): the little remaining brain tissue appears eroded, irregular, and dissolved, producing the classic sign of prominent orbits known as "Frogeye" or "Mickey Mouse" appearance [10]. There is a complete absence of the normal head contour.
- Polyhydramnios is observed mainly in advanced gestations due to deficient swallowing by the fetus.

4. Prevention

Preconception as well as antenatal folic acid supplementation is important at reducing the incidence of anencephaly and other NTDs. This can be achieved through food fortification with folic acid. Similarly, women who are diabetic should make sure that they are well optimized before they attempt to become pregnant. Prevention requires preconception counseling and strict glycemic control (HbA1c < 6.5%). This needs collaboration among interprofessional healthcare teams in delivering coordinated care for affected female patients in their reproductive years with diabetes as these patients are under the care of the internist before they conceive and are seen by the obstetrician, in most cases, only when they have become pregnant [11]. The internist should, at all medical reviews, ask diabetic women about their reproductive plans and, where necessary, refer them to the obstetrician for better evaluation, counselling and optimization. Finally, women should avoid alcohol consumption once they learn they are pregnant if they are to avoid this congenital malformation. As withdrawal from alcohol may be challenging, women who drink alcohol need psychosocial support from social workers to properly understand the danger of drinking alcohol while they are pregnant, and to help them abstain.

5. Conclusion

Anencephaly is the second most common type of neural tube defect. Unlike spina bifida, there is no effective treatment for children with anencephaly. Therefore, emphasis should be put on prevention by preconception and prenatal nutritional supplementation with folic acid; preconception diabetic control and avoidance of alcohol consumption during pregnancy especially in the first half of pregnancy. This requires a collaborative interprofessional primary health care team approach, with internal referral whenever necessary. Additionally, folic acid food fortification has proven to be beneficial at reducing the incidence of anencephaly in countries that have implemented it. Governments should therefore try to implement

this program as a primary health care intervention to reduce the incidence of anencephaly. Early prenatal screening and diagnosis with timely medical termination of affected pregnancies is also important in reducing the incidence of the condition.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

References

[1] Finnell, R.H., Caiaffa, C.D., Kim, S., Lei, Y., Steele, J., Cao, X., *et al.* (2021) Gene Environment Interactions in the Etiology of Neural Tube Defects. *Frontiers in Genetics*, **12**, Article 659612. https://doi.org/10.3389/fgene.2021.659612

[2] Kamal El, M., Sara El, B., Ibrahim, G., Aziz, B. and Aicha, K. (2021) Anencephaly: Case Report and Literature Review. *Journal of Gynecological Research and Obstetrics*, **7**, 5-7. https://doi.org/10.17352/jgro.000096

[3] Oumer, M., Kibret, A.A., Girma, A., Tazebew, A. and Silamsaw, M. (2021) Prevalence of Anencephaly in Africa: A Systematic Review and Meta-Analysis. *Scientific Reports*, **11**, 1-13. https://doi.org/10.1038/s41598-021-02966-w

[4] Tesfay, N., Hailu, G., Habtetsion, M. and Woldeyohannes, F. (2023) Birth Prevalence and Risk Factors of Neural Tube Defects in Ethiopia: A Systematic Review and Meta-Analysis. *BMJ Open*, **13**, e077685. https://doi.org/10.1136/bmjopen-2023-077685

[5] Munteanu, O., Cîrstoiu, M.M., Filipoiu, F.M., Neamțu, M.N., Stavarache, I., Georgescu, T.A., *et al.* (2020) The Etiopathogenic and Morphological Spectrum of Anencephaly: A Comprehensive Review of Literature. *Romanian Journal of Morphology and Embryology*, **61**, 335-343. https://doi.org/10.47162/rjme.61.2.03

[6] Chen, C.P. (2005) Congenital Malformations Associated with Maternal Diabetes. *Taiwanese Journal of Obstetrics and Gynecology*, **44**, 1-7. https://doi.org/10.1016/s1028-4559(09)60099-1

[7] Edri, T., Cohen, D., Shabtai, Y. and Fainsod, A. (2023) Alcohol Induces Neural Tube Defects by Reducing Retinoic Acid Signaling and Promoting Neural Plate Expansion. *Frontiers in Cell and Developmental Biology*, **11**, Article 1282273. https://doi.org/10.3389/fcell.2023.1282273

[8] Chen, Y., Ning, W., Wen, C., Gu, L. and Chu, X. (2025) Predictive Value of the Maternal Alpha-Fetoprotein Variant, L2, during the First Trimester of Pregnancy Screening for Fetal Trisomy 21, Trisomy 18, and Neural Tube Defects. *BMC Pregnancy and Childbirth*, **25**, Article No. 844. https://doi.org/10.1186/s12884-025-07893-6

[9] Moore, K.L., Persaud, T.V.N. and Torchia, M.G. (2020) Before We Are Born: Essentials of Embriology and Birth Defects. 10th Edition, Elsevier.

[10] Akagbue, V.N., Wekhe, C. and Ernest-Ekwe, C.L. (2024) Prenatal Diagnosis of Anencephaly: A Case Report and Review of Literature. *International Journal of Medical Science and Clinical Research Studies*, **4**, 1945-1949.

[11] Boardley, E., Shanks, A.L., Thada, P.K., *et al.* (2025) Diabetic Embryopathy. StatPearls Publishing. https://www.ncbi.nlm.nih.gov/books/NBK558974/