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Case Report

Anencephaly: a case report on the rare neural tube defect

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ABSTRACT

Anencephaly is a severe, lethal, neural tube defect characterized by the partial or total absence of the brain and calvarium, with a 100% mortality rate either in utero or shortly after birth. It occurs when anterior neuropore fails to close between 23-26 days post-conception, affecting approximately 1 per 1,000 births and is associated with genetic and environmental risk factors such as maternal diabetes, obesity, toxin exposure, and folate deficiency. The condition can be diagnosed early in pregnancy, usually between the 11th and 14th weeks of pregnancy, by USG identifying the lack of cranial ossification and failure to measure the biparietal diameter and also detection through maternal serum alpha-fetoprotein levels. Early diagnosis is essential for effective management and counselling, particularly because of the high likelihood of pregnancy termination due to the fatal prognosis.

Keywords: Anencephaly, Acrania, Maternal AFP, Neurulation, Pregnancy termination

INTRODUCTION

Anencephaly is a rare lethal congenital malformation of the foetal nervous system caused by a defect in neural tube closure, leading to the absence of significant portions of the brain, including the cerebrum, cerebellum, and brain stem and calvarium. This condition has a prevalence of approximately 1 in 1,000 from multifactorial causes including geographical, toxins, metabolic, rare chromosomal factors and nutritional as folic acid deficiency.[1] Anencephaly affects the foetus during the first month of pregnancy, stemming from neural tube closure disruption at 23 to 26 days of post conception.[2] It can be diagnosed as early as the first trimester at 11 to 14 weeks of gestation through ultrasound.[3] Since the 1990s, therapeutic termination of pregnancy has been recommended in many developed countries, where the birth rate of anencephaly is now almost zero.[4] This approach helps parents avoid the difficult decision of giving birth to a severely disabled child, as anencephalic newborns typically survive only a few hours after birth and are often blind, deaf, unconscious, and incapable of feeling pain.[2,3]

CASE REPORT

Mrs X, 30 years old**,** homemaker, G3P1L1A1, B+ve, belonging to middle class, presented with an anomaly scan report from a private hospital, performed at 19 weeks and 5 days of gestation, one day before, revealing an anencephalic foetus for further management.

Past obstetric history of a previous lower segment caesarean section (LSCS), due to severe oligohydramnios 4 years back and a history of spontaneous miscarriage of 6 weeks duration one year back for which no D & C was done.

Diagnosed with gestational diabetes mellitus (GDM) at 8 weeks of the current pregnancy at 1st visit to GH, she has been advised for insulin actrapid 4 units BD for the past three months, reports are not available.

There was no dating or nuchal translucency (NT) scan done, and folic acid supplementation began after 8 weeks.

On examination, the patient has an average build, is well-nourished, and has no other co-morbidities. Routine investigations were normal, except for mildly elevated fasting blood sugar (FBS) at 128 mg/dL and postprandial blood sugar (PPBS) at 156 mg/dL, which was managed with oral metformin 500 mg BID and MNT.

After obtaining informed consent and completing all necessary documentations, medical termination of pregnancy (MTP) was performed by oral mifepristone 200 mg, followed by 200 mcg misoprostol, 6 hourly intravaginally. She tolerated the regimen well without any untoward effect. Foetus was extracted in breech presentation. Post abortal period was uneventful and patient was discharged after 24 hours of observation.

Foetuses with anencephaly can be diagnosed as early as 11-13 weeks of pregnancy. USG findings typically appear normal until ossification fails. A first-trimester scan can provide a dependable diagnosis and enable early treatment of anencephaly. Upon observation, the fetus exhibited the absence of cranium, a very short neck, low-set ears, and protruding eyeballs (Figure 1 B). There was a small amount of brain tissue present, with a normal spinal cord.

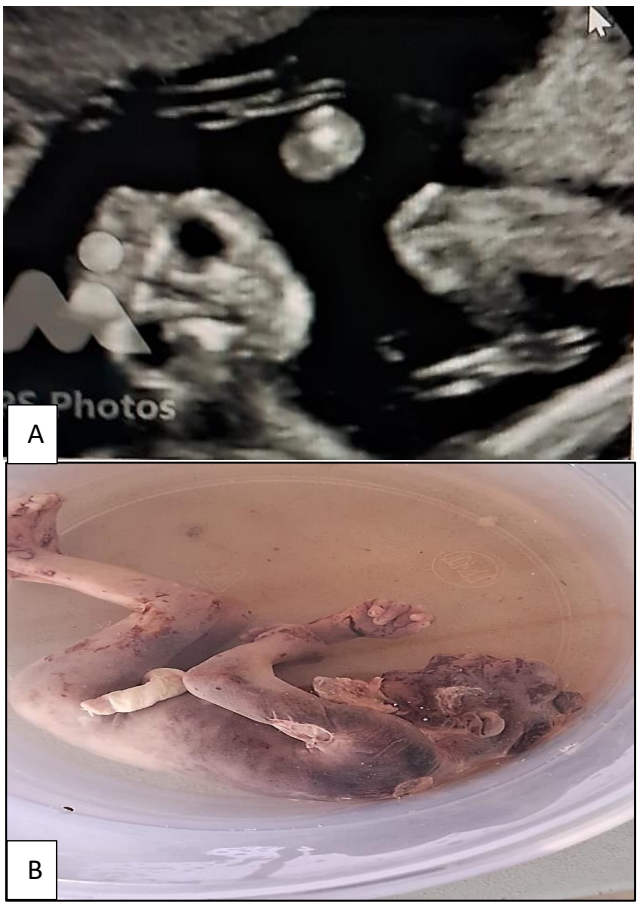

Figure 1 (A and B): Anencephaly USG image and anencephaly in foetus.

DISCUSSION

Anencephaly is a neural tube defect that results from improper closure of the neural tube during fetal development. Affected newborns are typically blind and deaf, and this condition ranks as one of the most prevalent neural tube defects, following spina bifida, with an incidence of approximately 1 in 1,000 pregnancies. Diagnosis can be made during the first trimester, between the 11th and 14th week of gestation, through ultrasound. Primary prevention of neural tube defects, including anencephaly, through folic acid supplementation during the periconceptional period has been shown to be effective Moussaoui et al.[5]

Neural tube defects, which encompass conditions such as anencephaly, spina bifida, and encephalocele, are among the most common congenital malformations, second only to heart and orofacial malformations.[5] Women who have had a previous pregnancy affected by anencephaly are at higher risk of recurrence in subsequent pregnancies Panduranga et al.[6]

In various studies, the average gestational age at diagnosis of anencephaly ranges from 18 to 22 weeks. For instance, Obeidi et al reported a mean diagnostic age of 21+4 weeks, with diagnoses occurring as early as 13+4 weeks and as late as 32+4 weeks.[7] Neural tube defects are more common among mothers with insulin-dependent diabetes, with the risk increasing from 2% at an HbA1c level of 5.5% to 6% at an HbA1c of 9%.[8]

Hydramnios, defined as an abnormal increase in amniotic fluid, occurs in 30-50% of anencephalic pregnancies and is believed to result from factors such as excess cerebrospinal fluid secretion, impaired swallowing, and abnormal amniotic fluid absorption. Anencephaly is a uniformly lethal condition, and its multifactorial origins emphasize the importance of folic acid supplementation for prevention. Canadian, British, and American health organizations recommend that women of childbearing age should consume 0.4 to 0.8 mg/day of folic acid, with a higher dose (0.8 to 4 mg) advised for those with a history of affected pregnancies.[8]

In this case report, the patient may have had undiagnosed diabetes mellitus, with a strong family history of the condition, which could have contributed to the development of the NTD. She began folic acid supplementation late, which is a known preventive measure.

CONCLUSION

Anencephaly can be diagnosed as early as 11weeks with transvaginal sonography and always associated with raised maternal serum AFP levels. Since anencephaly is rarely associated with aneuploidy, so amniocentesis for karyotyping is not necessary. Overall recurrence rate is 2-

5% in future pregnancies, but folic acid supplementation preconceptionally can reduce the risk by 70%.

Therefore, raising awareness about preventable causes like nutritional deficiencies and teratogen exposure is crucial for early diagnosis and the option of pregnancy termination to reduce recurrence and enhance early management.

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