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**INCIDENTAL PRENATAL DIAGNOSIS OF ANENCEPHALY IN A FETUS OF A 32-YEAR-
OLD WOMAN AT 32 WEEKS GESTATION**

INCIDENTAL PRENATAL DIAGNOSIS OF ANENCEPHALY IN A FETUS OF A 32-YEAR-OLD WOMAN AT 32 WEEKS GESTATION

Eretan T¹, Eretan O¹, Akinola R², Balogun B²

¹Department of Radiology, University of Medical Sciences Teaching Hospital, Laje, Ondo, Nigeria. ²Department of Radiology, Lagos State University Teaching Hospital, Ikeja, Lagos, Nigeria.

Correspondences to: Eretan T, Department of Radiology, University of Medical Sciences Teaching Hospital, Laje, Ondo, Nigeria. Telephone: +234-8033559098; E-mail: eretanhelma@gmail.com

ABSTRACT

Introduction: Anencephaly is a congenital central nervous system anomaly associated with absence of the skull vault and normal brain tissue with exposure of intracranial structures to the amniotic fluid. It is not compatible with life, with death occurring in most cases before birth. Genetic mutations, medications and environmental factors are some of the causative factors.

Case Report: Incidental prenatal diagnosis of anencephaly in a fetus of a 32-year-old multiparous woman at 32 weeks gestation is reported. The prenatal diagnosis, mode of delivery and the clinical findings on the fetus are discussed.

Conclusion: The index case highlights the importance of routine antenatal drug compliance especially folate in prevention of neural tube defects and early prenatal ultrasound screening in detection of congenital abnormalities of the central nervous system.

Keywords: Anencephaly, Congenital anomaly, Central nervous system

INTRODUCTION

Anencephaly is a congenital central nervous system anomaly associated with absence of the skull vault and normal brain tissue with exposure of intracranial structures to the

amniotic fluid. It is not compatible with life, with death occurring in most cases before birth. Genetic mutations, medications and environmental factors are some of the causative factors.

Neural tube defects can be prevented by dietary consumption of folates,^{1,2} as folic acid supplementation has been shown to be beneficial in reducing NTDs by 50 – 70%.³

Anencephaly is a relatively rare central nervous system embryological abnormality. It is uncommon in this locality. Imaging such as ultrasound in early pregnancy is useful in making a diagnosis.

CASE REPORT

A.O is a 32-year-old Gravida 2 para1 + 0 housewife, who presented at a private diagnostic facility for a routine antenatal ultrasound scan, on account of a large for date uterus at 32 weeks gestational age. Up until then the pregnancy had been uneventful.

She did not take folates in the periconceptual period. There was no history of trauma, smoking, ingestion of herbal mixtures, alcohol consumption or febrile illness in the index pregnancy.

Ultrasound scan performed using a 3.5 MHz transducer with coupling gel revealed a live active viable fetus with good cardiac activity and limb motion with unstable lie. Placenta was anterofundal, while the amniotic fluid

volume was greater than 12cm (single largest pocket). Total liquor volume was 49.48cm, indicative of polyhydramnios (Figure 1). The fetal calvarium was absent with protrusion of the orbits (Figure 2).

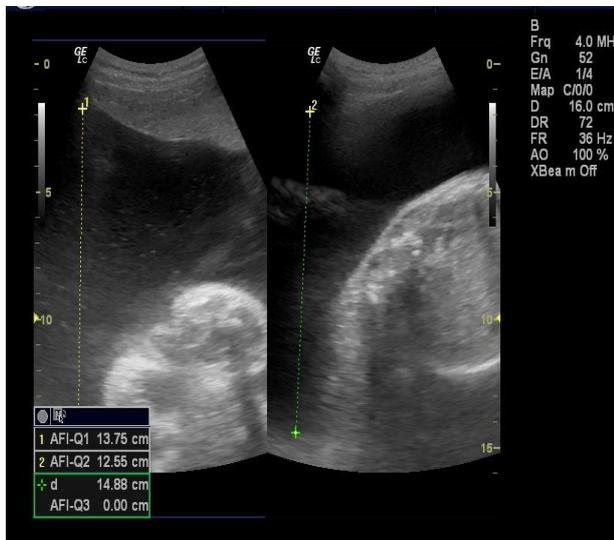


Figure 1: Grayscale ultrasound image of two single anteroposterior amniotic fluid pockets >8cm showing polyhydramnios

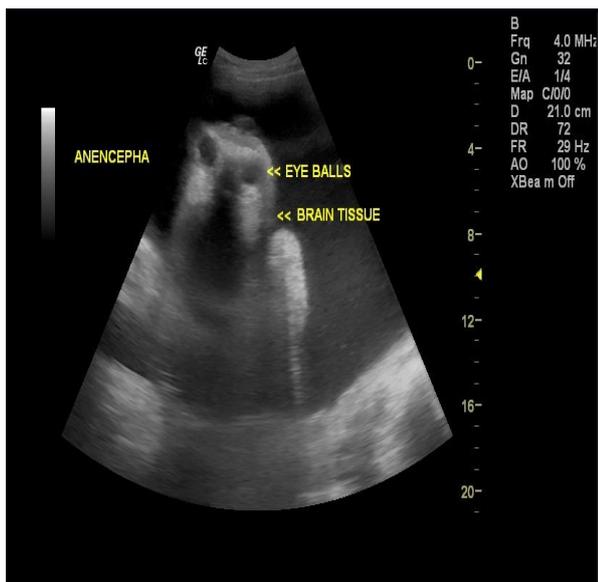


Figure 2: Grayscale coronal ultrasound image of fetal head showing absent fetal calvarium and dysmorphic brain tissue.

Additional parameters on scanning include; Abdominal circumference (AC) – 27.13cm – Av 31w2dy (31w2d -34w1d); Femur length (FL) – 6.75cm – Av 34w5d (31w5d – 37w5d); Estimated fetal weight (EFW) – 2045g ±

327.29g; Amniotic Fluid Index (AFI) – 13.75, 12.55, 10.27, 12.92 (49.48cm); Heart rate – 133bpm; Cummulative Gestational Age (CUA) 33w ± 1w1d.

A female neonate with anencephaly, short neck and malformation of the auricles was delivered by elective termination of pregnancy via caesarian section at 34 weeks gestation. Apgar score was 3 at 5 minutes; heart rate and respiratory rate were 90/minutes and 28/minutes respectively. Fetus died about 37 minutes after delivery (Figure 3). Delivery of the fetus was not attended by a paediatrician and internally related malformations of the organ systems could not be excluded as parents did not consent to an autopsy. Resuscitation of the newborn was not performed due to the already existing brain malformations. The parents were counselled and emotional support was given after the delivery.



Figure 3: Female fetus with anencephaly, short neck and malformation of the auricles. There is dysmorphic brain tissue and absent skull vault, with protrusion of the orbits giving a frog-like cranial appearance.

DISCUSSION

Maternal screening for elevated alpha-fetoprotein levels in the second trimester of pregnancy can identify over two-thirds of fetuses with open neural tube defects,

including almost all fetuses with anencephaly.⁴ The actual cause of this neural tube defect is not known, although pre-conceptional intake of dietary folates (up to 6 months) have been known to reduce its incidence. The mother admitted to non-compliance with hematinics as she did not like to take medications.

Risk factors for NTDs include genetic factors (genes associated with folate metabolism),⁵ gestational diabetes, maternal obesity and inadequate intake of nutrients such as folate and B12 which vary by ethnicity and race.⁶ Other known risk factors include passive smoking, maternal miscarriage history, twin and triplet pregnancies,⁷ young maternal age, older maternal age (between 18 – 42 years), (there is no age that is not at risk for NTDs, however risk increases with maternal age). About half of anencephalic fetuses are born to primigravida mothers; however the mother in this case was 32 years old and had a normal first child.

Mean survival times have been put at 55 minutes to 8 days.⁸ The fetus died within 31 minutes of delivery. The fetus was delivered via caesarian section on account of previous caesarian section in the mother.

Several malformations with various frequency distributions associated with this lethal anomaly include spina bifida with or without meningocele,⁹ facial defects (cleft lip/palate), kidney and urinary tract (renal agenesis, obstructive uropathy, dysplastic kidneys),¹⁰ limb and heart defects, genital defects (penis hypoplasia, cryptorchism, scrotum hypoplasia, ambiguous genitalia, hemiuterus), gastrointestinal tract defects (intestinal obstruction, imperforate anus),¹⁰ pulmonary defects (pulmonary agenesis, cystic adenomatoid malformation of the lung) and others such as single umbilical artery, omphalocele, congenital diaphragmatic hernia and gastroquiasis.^{9,10}

Alfa-fetoprotein levels are valuable as a marker for NTDs, as it is elevated in pregnancies of fetuses with NTDs.

CONCLUSION

The index case highlights the importance of routine antenatal drug compliance especially folate in prevention of neural tube defects and early prenatal ultrasound screening in detection of congenital abnormalities of the central nervous system. There is a need for further studies on evaluation of risk factors especially as relates to genetic metabolism of folates and the presence/absence of teratogenic agents.

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