International Journal of Medical Science and Clinical Research Studies

ISSN(print): 2767-8326, ISSN(online): 2767-8342

Volume 04 Issue 11 November 2024

Page No: 1945-1949

DOI: https://doi.org/10.47191/ijmscrs/v4-i11-01, Impact Factor: 7.949

Prenatal Diagnosis of Anencephaly: A Case Report and Review of Literature

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ABSTRACT

Anencephaly is an inborn deformity of the central nervous system that results to the failure of closure of the cranial end of the embryologic neural tube, typically happens between the 23rd to 26th days after conception. It represents 40% of neural tube deformities, which is the second foremost cause of nervous system abnormalities after spina bifida. The occurrence rate is 1/1,000. Diagnosis is made by the 1st trimester ultrasound between the 11th and 14th week. Etiologies are multifactorial (toxic, metabolic, nutritional, iatrogenic and exceptionally chromosomal).

The importance of this case report is to highlight the role of imaging in the making prenatal diagnosis and thereby reiterating the usefulness of anomaly scan in pregnant women in our clime. Ultrasound is a non-ionizing imaging modality which is highly operator dependent and is repeatable.

Our case study is a 36-years-old woman Gravida 3 Para 1+2 (None Alive) at 24weeks 1day Gestation age (GA) who presented to the Radiology department for routine obstetric scan for the first time in index pregnancy. The obstetric ultrasound shows absence of the cranium in an active fetus with good cardiac activity (fetal heart rate =158bpm) with increase amniotic fluid in transverse lie. Frog eye sign was also appreciated. The placenta is sited anteriorly and not low lying. Femoral length (FL) measures 4.31cm with a GA of 24 weeks 1 day.

KEYWORDS: Anencephaly, Ultrasonography, Neural Tube Defect, Prenatal diagnosis.

ARTICLE DETAILS

Published On:
04 November 2024

Available on:
https://ijmscr.org/

INTRODUCTION:

Anencephaly is an embryological deformity of the central nervous system, consistently fatal, categorized by the nonappearance of the brain and cranial vault along with other defects of the cranial structures. From the embryological standpoint, anencephaly results from a failure in the neural tube closure. Quite a rare anomaly with incidence up to 1 in 20,000 live births [1].

Notably, the newborn may be blind, deaf, unconscious and usually dies during the birth process or within a few hours. Diagnosis can be made in-utero on ultrasound examination between 11th-12th week of pregnancy and by raised maternal serum levels of alpha fetoprotein (AFP) [2]. Polyhydramnios is a common association with anencephaly. Approximately 65% of the cases of anencephaly die in-utero, and some may be delivered prematurely. Interestingly, it has a multifocal pattern of transmission, with the interface of environmental and genetic factors. Report has it that folic acid supplementation reduces/diminishes the risk of anencephaly to a great degree [3]. Embryologically, neural crest cells originating in the neuroectoderm form the facial skeleton and most of the skull. Note that these cells are highly susceptible and as they leave the neuroectoderm, they are frequently a target for teratogens. Mothers exposed to teratogens also show increased risk of such defects [4]

Sonography is a diagnostic tool. The importance of this case report is to highlight the role of imaging in the making prenatal diagnosis and thereby reiterating the usefulness of anomaly scan in pregnant women in our clime. It is also to add to the body of already existing knowledge on the subject matter.

This case report shows that prenatal diagnosis of congenital anomalies is very much possible and the obstetricians should encourage expecting mothers to do anomaly scan early in the pregnancy.

AIM/OBJECTIVE

To report the occurrence of this rare congenital deformity in our health facility/environment as well as further contribute on the already existing knowledge of the use of ultrasonography in confirming and monitoring pregnancy at different stages as well as prompt detection of congenital malformations/other sinister issues.

CASE REPORT

Our case study is a 36-years-old woman Gravida 3 Para 1+2(None Alive) at 24weeks 1day Gestation age(GA) who presented to the Radiology department for routine obstetric scan for the first time in this index pregnancy. The obstetric ultrasound shows absence of cranium in an active fetus with good cardiac activity (Fetal heart rate =158bpm) with increase amniotic fluid in transverse lie. Frog eye sign was also appreciated. The placenta is sited anteriorly and not low lying. Femoral length (FL) measures 4.31cm with a GA of 24 weeks 1 day. Patient was counselled on the findings of the ultrasound, since it is a highly sort after pregnancy after several years of waiting. Patient represented for scan at 28 weeks with history of liquor drainage, similar findings was still documented, she later fell into labour and delivered an anencephalic fresh still birth male baby with zero APGAR scores at 1 and 5 minutes respectively. Birth weight was 1.3kg. Placenta weight was 0.4kg.

Images:

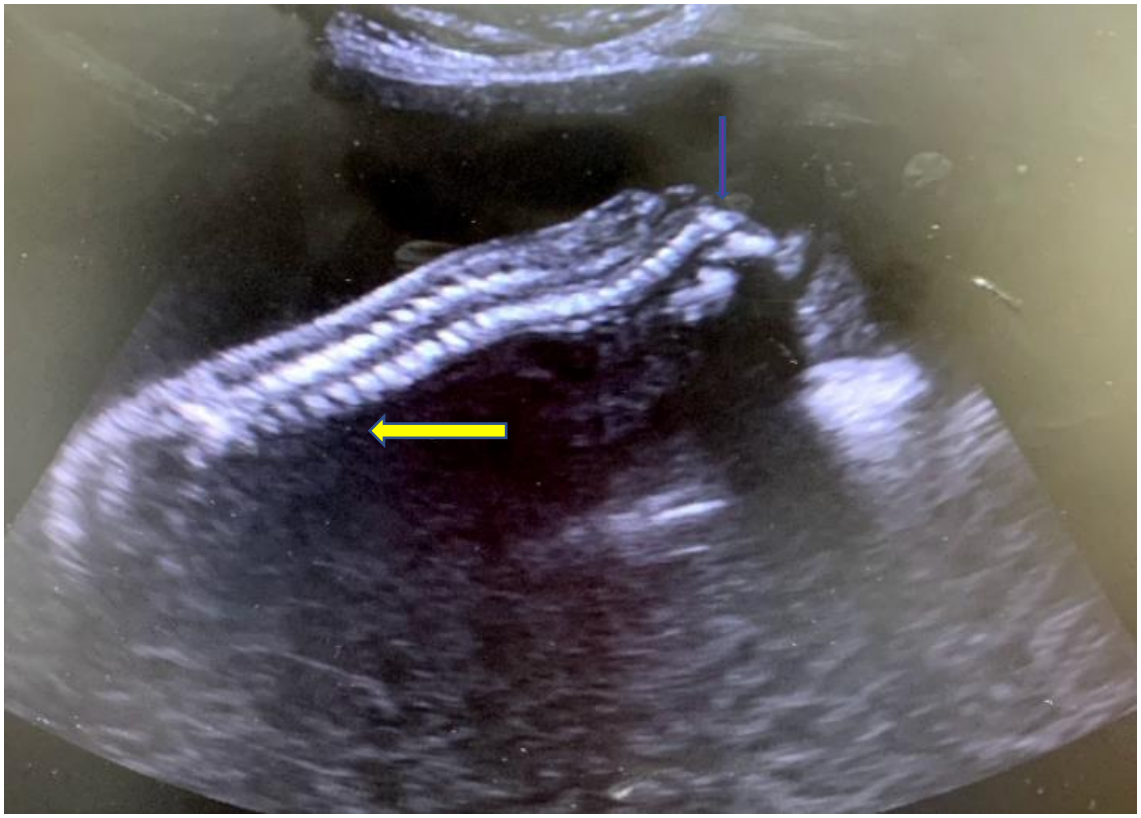
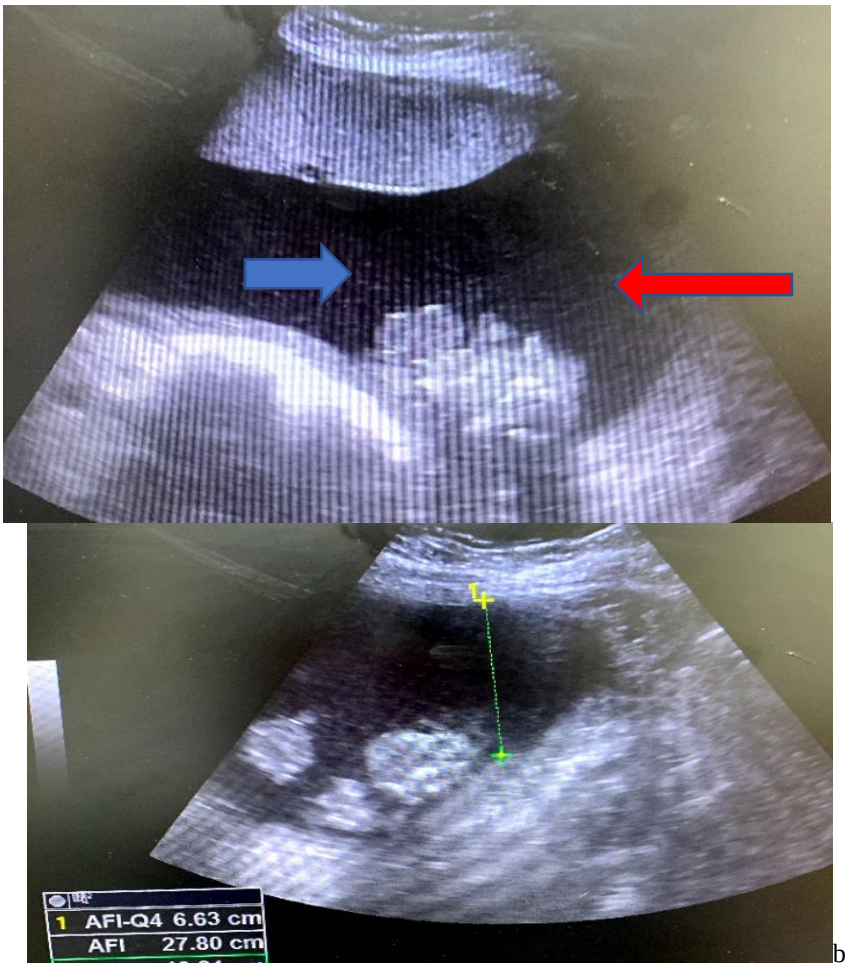


Figure 1 is a B-mode image of the fetus showing the absence of the cranial vault (blue slim arrow) and the spine(slimmed yellow arrow).

a



b

Figure 2a and b are B-mode images of the gravid uterus showing the placenta anteriorly placed(blue thick arrow), large pool of amniotic fluid(red arrow) and amniotic fluid index in b of 27.80cm. The finding is in keeping with polyhydramnios.

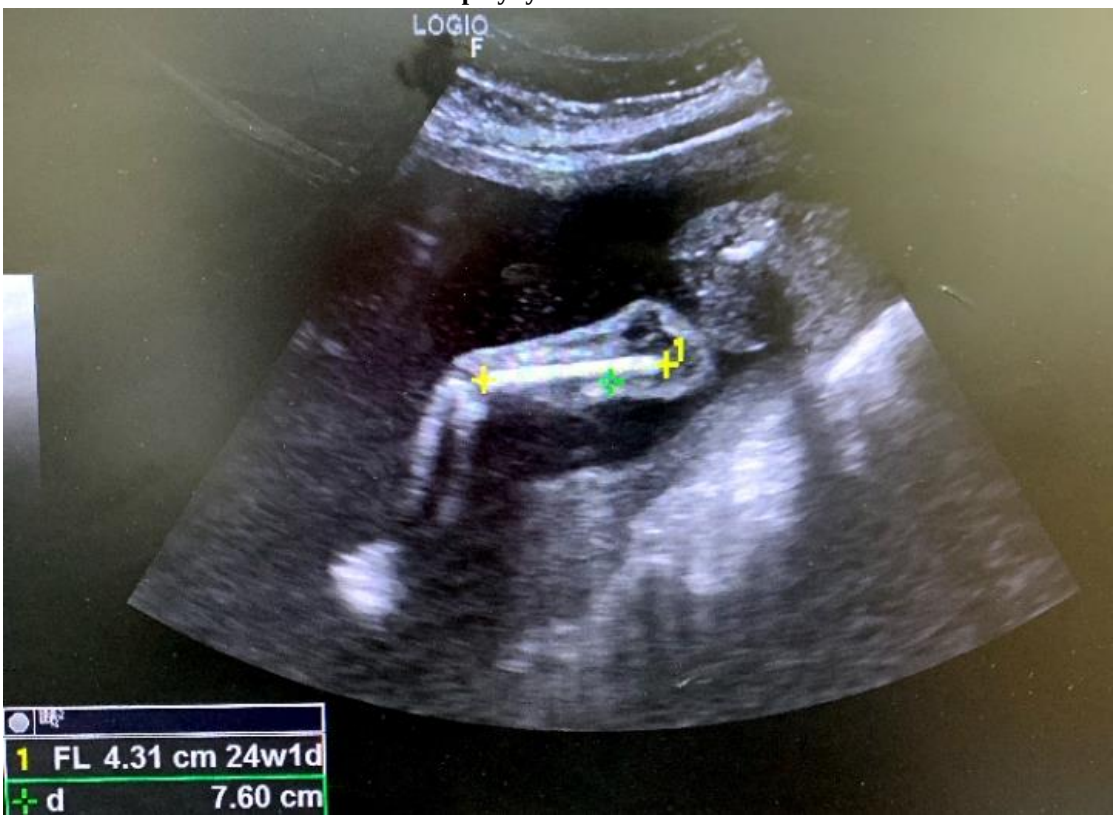


Figure 3 is a B-mode image of the fetus showing the femoral length for dating the pregnancy.

Figure 4 is a B-mode image of the fetus showing the a close up view of the characteristic ``frog eye appearance``.

DISCUSSION

A defect in the closure of the neural tube during fetal development is referred to as anencephaly. The striking features seen in a baby born with anencephaly is often deaf, unconscious, analgesia and blind. Additionally, it is one of the commonest forms of neural tube defect, after spina bifida, affecting nearly 1 in 1,000 pregnancies. It is worthy of note that diagnosis is made through first trimester ultrasound between 11th and 14th week of pregnancy by the demonstration of exencephaly which results in the non-visualization of the ossification of the cranial box and hence making the measurement of the biparietal diameter unattainable. Studies have shown that primary prevention of neural tube closure abnormalities by taking folic acid during the periconceptional period is effective. These are the examples of Neural Tube Defects (NTDs) in no particular order spina bifida, encephalocele and anencephaly. They are the commonest congenital deformities after heart and orofacial deformities [5]. Despite advancement in research their etiologies is considered multifactorial.

In Africa, precisely Morocco, the frequency is around 10 cases per 10,000 births each year. Previous studies have revealed that probably of having a second anencephalic child is raised in women who has had a previous child affection compared to the rest of the population [6]. Panduranga et al. [7] in their study, all cases were detected between the 16th and 34th week of pregnancy with a mean gestational age of 21 weeks. Another study by Shilpa et al. [8] documented an average age of diagnosis of 22 weeks, fluctuating between the 20th and 30th week of amenorrhea (WA). In a related study by Emre et al. [9] anencephaly was diagnosed in 28 fetuses between 11- and 36-weeks gestational age. The mean age of diagnosis was 18 weeks. Nidaa et al [10] documented 21 + 4 WA as the mean gestational age at diagnosis with an interval between 13 + 4 and 32 + 4 WA. Neural Tube Defects (NTDs) are more common Maternal insulin-dependent diabetes in the first trimester of pregnancy has been implicated as common risk factor for neural tube defects (NTDs). In comparison to the general population, the occurrence of these malformations varies/differs from 4% to 15% (2.1% in the general population). Modest hyperglycemia values subject also show increased risk for malformation. The risks increases as the level of HbA1c increases (2% for an HbA1c of 5.5%, to 6% for an HbA1c of 9%)[11].

In Morocco, the use of this plant fenugreek (Trigonellafoenumgraecum L.) is wide and in Arabic Helba, is one of the oldest medicinal and culinary plants. Its seeds are useful in the management of diabetics, for controlling glucose and cholesterol levels as well as for pregnant and lactating women [12-13].

Also, in the Maghreb region, it is used in the treatment of wounds, diarrhea, acne, dehydration, anemia, bronchitis, rheumatism, stomach aches, constipation and arterial hypertension, either in the form of decoctions or of seeds reduced to flour and mixed with the honey [14]. Several prospective studies have shown that fenugreek have teratogenic effects in humans and animals. Likewise, cases of noticeable birth defects such as hydrocephalus, anencephaly, cleft palate and spina bifida have been found in women who consumed fenugreek seeds during gestation.

Some studies have also shown that fenugreek is neuroprotective [15,16] while, the developing nervous system appears to be particularly sensitive to the toxicity of fenugreek, as earlier reported in Moroccan studies showing that children are more likely to develop encephalopathy such as hydrocephalus, anencephaly and spina bifida.

In another study done by Seneffs et al[17] documented that several pathways by which glyphosate is unfavorable and lead to anencephaly. It is an amino acid analogue of glycine and it is alleged to interrupt glycine dependent processes mostly in the forebrain but not in the hindbrain. It is a key component of herbicides and infants born in agricultural population where glyphosate-based herbicides are profoundly used have a higher rate of anencephaly when either of the parents were unprotected. A contrary report was documented by Okunola et al[18], wherein they reported similar prevalence of anencephaly with non-agarian community.

Several studies have shown that more females fetuses have anencephaly [8, 8-19], however, in this case report it's a male fetus, this corroborates with the finding in El-Moussaoui et al[20].

Fetal abnormalities detection during the first trimester yield high results. Ultrasonography during the second trimester,

gives away the characteristic appearance of anencephaly "frog eyes sign", due to the absence of visible brain tissue above the eye sockets(Figure 4). Polyhydramnios is referred as a pathological increase in the volume of amniotic fluid during gestation, the diagnosis is made by ultrasound by measuring the amniotic fluid index/ using a single largest pocket (Figure 2a and 2b). It is connected with anencephaly in 30% to 50% of cases. This corroborates with the finding in this case. Some of the causative factors comprise secretion of cerebrospinal fluid into the amniotic cavity, lack of absorption of amniotic fluid by the hypoplastic lungs, excessive urine production due to lack of antidiuretic hormone and lack of normal swallowing. Polyhydramnios is the commonest demonstration of anencephaly before child birth. Anencephaly is a consistently fatal anomaly. It seems to be of multifactorial origin, subsequently consumption of foods rich in folic acid at least 3 months prior to planning their pregnancy, and to maintain proper intake if they are of child bearing age or patients desirous of birth is advised. Based on studies carried out on primary and secondary prevention, Canadian, British and American organizations recommend that women of childbearing age should consume 0.4 mg to 0.8 mg/day of folic acid to diminish cases of anencephaly. Note that women with past history of anencephaly should consume a dose ranging from 0.8mg and 4.0mmg on a daily basis.

CONCLUSION

This case report has shown that radiological investigation is fundamental in every successful journey of pregnancy. Therefore, regular interval obstetric scan should be scheduled for all expecting mothers and our African expecting mothers should enroll early for ante-natal care. Prenatal diagnosis still has its own challenges since there is no clear law regulating medical abortion and being a religious sets of people abortion is criminalized in our setting. Multidisciplinary approach should be engaged.

CONSENT

Verbal/written consent was obtained.

AUTHORS CONTRIBUTION: VNA- Manuscript conceptualization, reviewed and edited the manuscript, performed and interpreted the radiological studies, CW- reviewed and edited the manuscript, also assisted with the interpretation of the radiological studies, CLE-E-clerked, scanned and interpreted images, reviewed and edited the manuscript.

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