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Case Report

Anencephaly in A 32 Weeks old Intrauterine Fetus: The Ultrasonographic Findings and a Case Report

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Abstract

Anencephaly is a serious neural tube defect in which parts of the brain and skull are not developed, this may be associated with other congenital malformations.

Anencephaly is caused by failure of closure in the cranial neuropore between the third and fourth week of gestation (23rd and 26th embryonic day) resulting in the absence of a major portion of the brain, skull and scalp.

This is a case of a 32 weeks female fetus carried by a 40-year-old $G_7 P^{6+0}$ house wife who was referred for an obstetric ultrasonography scan on account of exaggerated symphysio-fundal height and routine pregnancy check-up. The findings of the scan were polyhydramnios, a single tone live fetus with absent skull vault and protruding eyes bilaterally likened to the frog eye appearance of anencephaly. We present this case because of its peculiarity and rarity in the literature.

Keywords: anencephaly; intrauterine; ultrasonography; fetus

Introduction

Anencephaly is a serious neural tube defect in which parts of the brain and skull are not developed, this may be associated with other congenital malformations [1].

Anencephaly is caused by failure of closure in the cranial neuropore between the third and fourth week of gestation (23rd and 26th embryonic day) resulting in the absence of a major portion of the brain, skull and scalp [2].

Anencephaly is also characterized with abnormal vascularization of the embryonic exencephalic brain which leads to degeneration of the nervous tissues making the brain to remain like a spongy vascular mass following failure of closure of the cranial neuropore during the fourth week of intrauterine development [1,4,5]

In anencephaly the brain lacks part or the entire cerebrum with the remaining brain tissue often exposed to injury from amniotic fluid [2,3].

The cause of anencephaly is still debatable but the defect is primarily that of failure of closure of the rostral neuropore and can affect many systems individually [1,6]. In the etiology of anencephaly which is unknown, some associated factors play some role, these factors are either socioeconomic status, environmental conditions, and genetics of both population and familial ancestry [2,7,8].

Anencephaly is associated with anomalies of not only the central nervous system but other systems respectively. Some of the associated anomalies are spina bifida, cleft palate, cleft lip, hypospadias, gastroschisis, clubbed feet and hands, omphalocele and many more [1].

Anencephaly is preponderant in the female gender, this has been reported by many researchers [1,2,9,10].

Anencephaly can be diagnosed by ultrasound examination and by elevated maternal alpha feto protein level (AFP). In most cases pathological examination of the abortus is needed due to common association with systemic anomalies [11,12]. Anencephaly was the first fetal malformation diagnosed prenatally by Campbell and colleagues using trans-abdominal ultrasonography [11,13].

Case Report

This is a case of a 32 weeks female fetus carried by a 40-year-old $G_7 P^{6+0}$ house wife who was referred for an obstetric ultrasonography on account of routine pregnancy check-up, lower abdominal pain and discomfort with intermittent abdominal cramps.

The mother had a poor antenatal history; this was late and had no history of routine folate intake in the index pregnancy. No history of congenital anomaly in the family. The previous pregnancies were uneventful.

The findings of the scan was polyhydramnious (free single pocket of amniotic fluid devoid of fetal part of about 14cm perpendicular dimension), a single tone live female fetus with absent skull vault and protruding eyes bilaterally likened to the "frog eye" appearance of anencephaly. Figure 1.

No associated facial anomalies, spinal anomaly, limb anomalies, gastric and renal anomalies were demonstrated following the ultrasonographic examination.

The pregnancy was terminated with expulsion of the fetus, the mother was reassured, and advised on appropriate antenatal care and benefits of early commencement of antenatal care with intake of folate especially in the early trimester of pregnancy

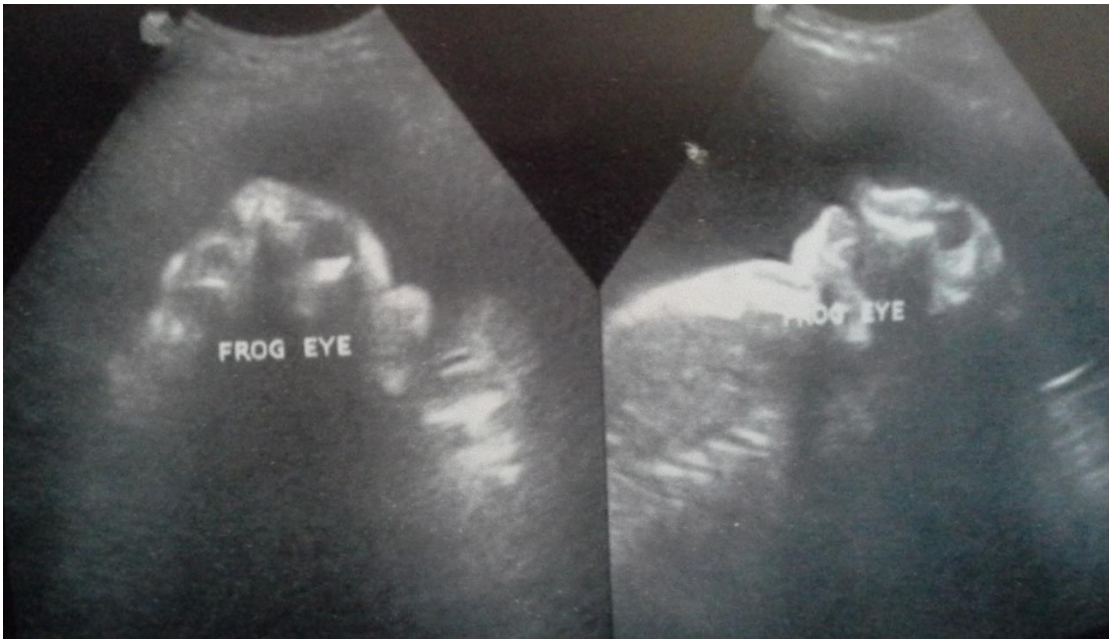

Figure 1: *These are obstetric ultrasonograms demonstrating the so called "frog eye" appearance of anencephaly and absent skull vault in both images. Associated marked amount of amniotic fluid volume (polyhydramnious) is also demonstrated.*

Discussion

Anencephaly is a neural tube defect (NTD) and mainly diagnosed prenatally by trans-abdominal ultrasonography; this is reported by numerous researchers and similarly the index case was also diagnose prenatally following trans-abdominal ultrasonography.

Anencephaly is preponderant in the female population; this was observed and documented in some literatures, the index case was also a female fetus.

Anencephaly been an NTD is more likely to occur with advancing maternal age and parity; this is a documented fact in the literature. This index case was from a female aged 40 years old and happens to be the seventh pregnancy of the mother.

Most NTD's were commonly associated with poor obstetric management likely failure or inadequate folate/folic acid intake especially in the first trimester; this has been observed by some researchers and we also reported failure of folate intake by the mother in the first trimester and subsequently in this index case/pregnancy.

The cause of anencephaly is unknown in most instances, we could not demonstrate the possible cause in this index case ultrasonographically but we presumed that it might be associated with failure of folate intake in the first trimester of this pregnancy.

Polyhydramnious which is an excess of amniotic fluid is a common finding associated with most NTD like anencephaly; this had associated excess amniotic fluid volume of a free pocket more than 14cm.

The classical finding of absent skull vault, exposed fetal brain and protruding eye balls which were synonymous with anencephaly ultrasonographically and reported by most literatures were also demonstrated ultrasonographically in this index case.

Some of the associated anomalies like spinal masses, clubbed hands and foot, obvious renal and gastric with abdominal wall defects that were documented in the literatures were not seen in this index case.

The commonest gestational age of delivery was 32 intrauterine weeks; the index case was also diagnosed at this age most likely from the history of lower abdominal discomfort and abdominal cramps reported by the mother. We assumed that these symptoms may be that of spontaneously preterm induced labor associated with the condition.

Termination of pregnancy with expulsion of the fetus and other products of conception happens to be the mainstay of treatment, the index case also had similar treatment option, conforming to that documented in the literature

Conclusion

Anencephaly been part of the NTD is predominantly associated with failure of adequate folate intake especially in the first trimester pregnancy. Trans-abdominal ultrasonography remains the main and first important diagnostic tool in the diagnosis of intrauterine anencephaly.

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