Case Report

Anencephaly: Prenatal Ultrasonographic Features and Preventive Strategies – A Case Report

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Abstract:

Anencephaly is a severe congenital anomaly characterized by the absence of a major portion of the brain, skull, and scalp. We report a case of 27-year-old woman, diagnosed at 17 weeks and 6 days gestation during her first obstetric ultrasound. The scan revealed characteristic findings of anencephaly, including absent cranial bones, brain tissue, and polyhydramnios. No family history of birth defects or miscarriages was reported. Preventive measures such as preconception folic acid supplementation are crucial, reducing NTD risk by up to 70%. The fetus was aborted, and post-abortion imaging confirmed the diagnosis.

Key words: Anencephaly, Neural Tube Defects, Ultrasound, Folic Acid, Prenatal Diagnosis, Polyhydramnios

Introduction:

Anencephaly is a lethal neural tube defect (NTD) caused by the failure of cranial neural tube closure during early development.[1] Anencephaly is part of a broader NTD spectrum, including spina bifida and encephalocele.[2] It is one of the most serious neural tube defects (NTDs), resulting from the failure of the upper part of the neural tube to close during embryonic development. This condition is incompatible with life, and early prenatal diagnosis through ultrasound is crucial for managing the pregnancy and counseling the parents. This case highlights the critical role of prenatal ultrasound in detecting NTDs and underscores the importance of folic acid in preventing these defects.[3]

Case Report:

A 27-year-old pregnant female presented to the department of radiodiagnosis of tertiary care institute for her first obstetric ultrasound at 17 weeks and 6 days of gestation. She had not undergone any previous ultrasound examinations during this pregnancy and was unsure of her last menstrual period date. She had one male child with no history of congenital defects and no miscarriages history.

Ultrasound Findings:

The ultrasound examination revealed the following features consistent with anencephaly:

- Absence of cranial bone and brain tissue: The "frog-eye" sign was evident, characterized by bulging eyes due to the lack of frontal bones. (Fig. 1)
- Absence of the calvaria: The cranial vault was not visualized. (Fig. 2)
- Polyhydramnios: Increased amniotic fluid was noted, which is often associated with impaired fetal swallowing in anencephalic fetuses.
- Gestational age estimation: Based on the fetal femur length (FL), the gestational age was estimated to be 17 weeks and 6 days.

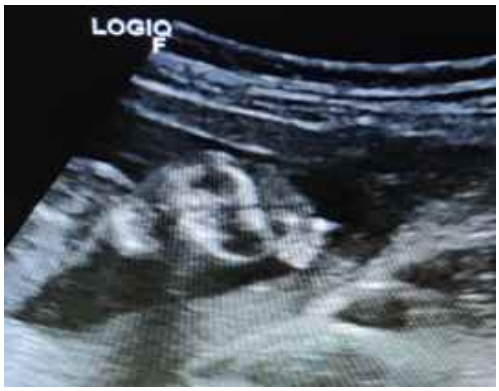

Fig. 1: Absence of cranial bone and brain tissue, giving a “frog- eye” appearance

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ISSN No. *: (p) 2348-523X, (o) 2454-1982*

DOI: 10.46858/vimshsj.110306

Date of Published : *30th September 2024*

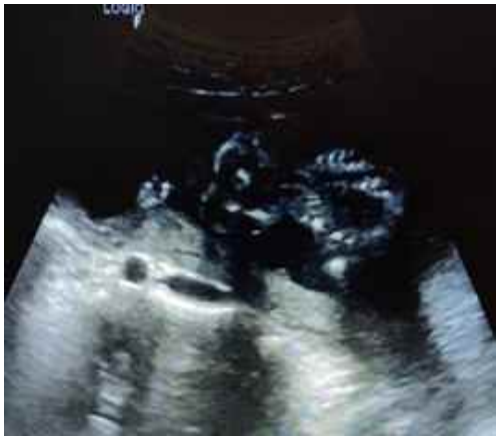

Fig. 2: Bulging eyes and absence of calvaria

Discussion:

Anencephaly can be diagnosed as early as the first trimester using ultrasound, with characteristic findings such as the "Mickey Mouse" sign due to the bi-lobed appearance of the cranial remnants. In the second trimester, the diagnosis is confirmed by the absence of the cranial vault and brain tissue, along with the frog-eye sign and polyhydramnios.[4]

Neural Tube Defects (NTDs) are a group of congenital malformations that occur due to incomplete closure of the neural tube during embryogenesis. Besides anencephaly, other common NTDs include:

- *Spina bifida:* Characterized by the incomplete closure of the spinal column, which can result in a range of disabilities from mild to severe.
- *Encephalocele:* A sac-like protrusion of the brain and meninges through a defect in the skull.
- *Iniencephaly:* A rare condition where the occipital bone of the skull and the cervical spine are defective, leading to a characteristic head-bent-back appearance.

Exencephaly and its Relation to Anencephaly: Exencephaly is a precursor to anencephaly, where the brain is present but exposed due to the lack of cranial bone coverage. Over time, the exposed brain tissue degenerates, leading to anencephaly. This progression from exencephaly to anencephaly can be observed in sequential prenatal ultrasounds.[5]

Other Common Findings in Fetuses with Anencephaly:

- Facial abnormalities: Including cleft lip and palate.
- Heart defects: Congenital heart anomalies are often present.
- Gastrointestinal malformations: Such as omphalocele.
- Renal anomalies: Including hydronephrosis or renal agenesis.

Syndromes Associated with Anencephaly: Anencephaly can occur as part of various syndromes and genetic conditions, including:

- Meckel-Gruber syndrome: Characterized by the presence of encephalocele, polycystic kidneys, and polydactyly.
- Trisomy 18 (Edwards syndrome): Associated with multiple congenital anomalies, including anencephaly.
- Roberts's syndrome: A rare genetic disorder that can include anencephaly among other skeletal abnormalities and growth deficiencies.[6]

Prevention:

The risk of NTDs can be significantly reduced by ensuring adequate maternal intake of folic acid before conception and during early pregnancy. The Centers for Disease Control and Prevention (CDC) recommends that all women of childbearing age consume 400 micrograms (mcg) of folic acid daily, as this can prevent up to 70% of NTDs.

Fig.3: Abortus showing absence of cerebral hemispheres and cranial vault.

Outcome:

In this case, the fetus was aborted in the obstetrics and gynecology (OB/GYN) department. Images of the abortus were obtained, which confirmed the diagnosis of anencephaly with classic features such as the absence of the cranial vault and cerebral hemispheres.(Fig. 3)

Conclusion:

Prenatal ultrasound evaluation plays a critical role in the early detection of anencephaly and other neural tube defects. Early diagnosis allows for appropriate medical and parental decision-making. Furthermore, preventive measures, such as folic acid supplementation before conception and during early pregnancy, are essential in reducing the incidence of NTDs. This case underscores the importance of routine prenatal care and the benefits of early nutritional intervention to prevent congenital anomalies.

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