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Termination of Pregnancy After Prenatal Diagnosis of Anencephaly: Case Study and Literature Review

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ABSTRACT

Anencephaly is a form of neural tube defect, which develops when the cranial neuropore is unable to close. It is one of the fatal neural tube defects. Although most causes of anencephaly are unknown, multiple risk factors are associated with this defect. The purpose of this article is to study the main risk factors and possible means of prevention of this common malformation through a clinical case and a literature review.

Keywords
Anencephaly, Neural tube defects, Ultrasound, Folic acid.

Introduction
Anencephaly is a fatal disease defined by the total or partial absence of the skull, with absence of the brain [1], infants born alive usually survive less than a day, its incidence is 1 to 5 in 1000 births. [2].

The etiology of anencephaly remains uncertain, but various environmental and genetic risk factors have been reported (diabetes, obesity, drugs, genetic polymorphisms and mutations, folate deficiency).

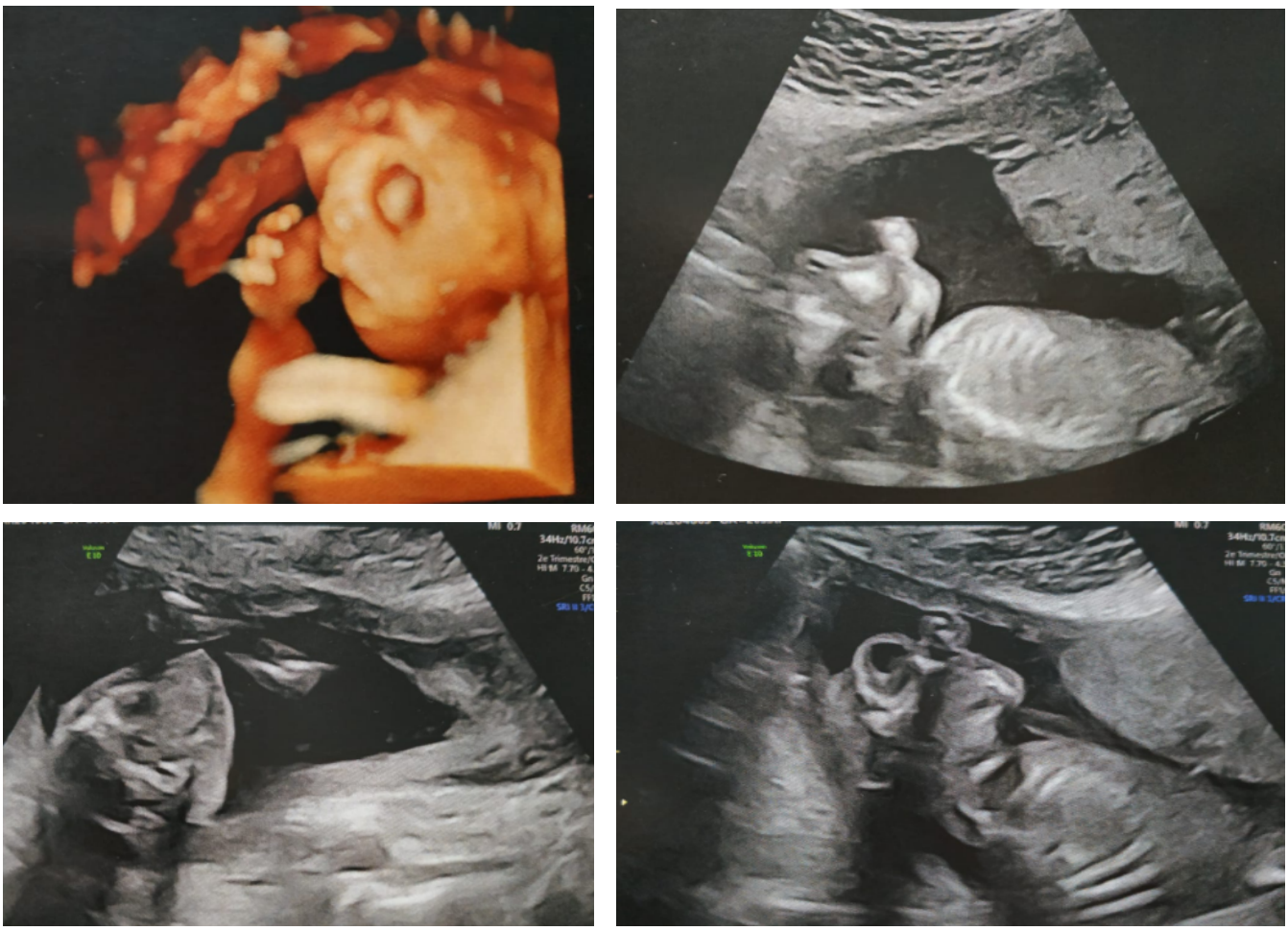
Anencephaly is accessible to screening by ultrasound in the first trimester allowing planning the best, safe and early management.

We report the case of a patient admitted to our facility for medical termination of pregnancy at 4 months for anencephaly.

Observation
Mrs SS, 40 years old, with no particular pathological history, fifth gesture, having four children by vaginal delivery, was admitted to our structure for the fortuitous discovery of an anencephaly in an unattended pregnancy presumed at 4 months. The clinical examination revealed a parturient outside of labor. The ultrasound confirmed the diagnosis by showing the absence of a cranium with the classic image of a batrachian (Figure 1- 4). Since this is a non-viable malformation, the decision was made to perform a medical termination of the pregnancy, labor induction was performed by administration of misoprostol. The vaginal delivery resulted in a female neonate with a birth weight of 500g in apparent death with an Apgar of 1/10 (Figure 5).

Discussion
Anencephaly isthe most common malformation of neural tube defects. The etymology comes from the Greek words "an", which means "without" and "enkephalos", which means "encephalon". It is defined by the total or partial absence of the calvaria with absence of the brain. The brain stem, cerebellum and diencephalon are usually present [1].

It is a relatively common type of malformation occurring in approximately 1 to 5 of every 1000 births. In the United States, approximately 1 in 4600 babies is born with anencephaly [2]. The mortality rate is 100% during intrauterine life or within hours or days of birth and the percentage of pregnancy termination is greater than 83% [3]. Other malformative anomalies are associated in 12-25% and genetic anomalies are detected in 1-5% [4].



Figures 1-4 : Ultrasound appearance of anencephaly with the classic batrachian image.

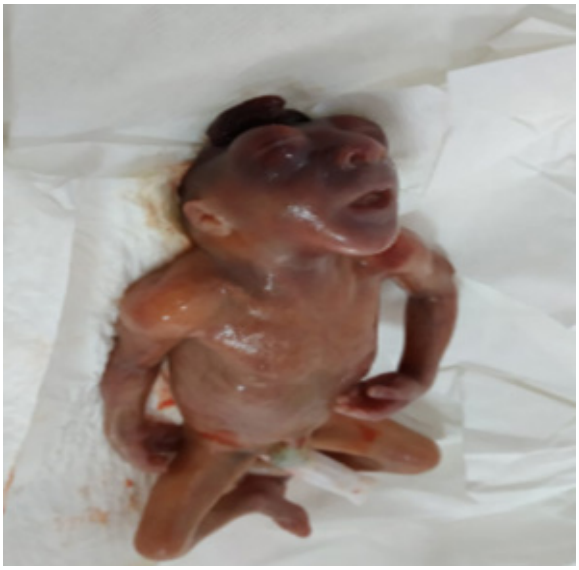


Figure 5: Birth appearance.

The exact etiology of anencephaly remains uncertain, several environmental and genetic risk factors have been incriminated, mainly folate deficiency. It could have multiple causes and systematic folic acid supplementation reduces the risk of neural tube defects [5].

Antiepileptic drugs are also incriminated in the genesis of neural tube defects; valproate is the most teratogenic and should therefore never be administered in the first instance [6]. Isotretinoin, a selective serotonin reuptake inhibitor used in dermatology, is associated with an increased risk of anencephaly [6]. Pesticide exposure is also known to be a factor exposing to neural tube closure anomalies in particular anencephaly [7].

Diabetes, hyperinsulinemia and a body mass index (BMI) of 30 kg/m 2 or more are known to be risk factors for neural tube defects [7].

It has been found that the female sex of the infants is also a factor of rsic because most of the anencephalic fetuses are female as it was in our case [8].

Anencephaly can also be part of chromosomal abnormalities, such as Edwards syndrome - trisomy 18 [9]. In addition, studies show that many anencephalic fetuses have associated anomalies, such as spina bifida, cleft palate, clubbed foot, clubbed hands and

gastroschisis, suggesting the overwhelming presence of genetics in neural tube defects [10]. These aspects highlight the importance of genetic factors in the etiopathogeny of neural tube defects [7,9,11]. Therefore, in recent years, more and more studies have evaluated the involvement of additional genetic and non-genetic risk factors in the development of anencephaly [8].

The first trimester ultrasound is the first essential examination for the detection of prenatal CNS anomalies, performed between 11-13+6 weeks of gestation, or later in the second and third trimesters, anencephaly can be detected in 100% of cases [12,13]. The current challenge in daily practice is early diagnosis when the complication rate of pregnancy termination is lower.

Once the diagnosis is established, an announcement to the parents with a proposal for termination of pregnancy is warranted [14,15]. If termination of pregnancy is not accepted, certain maternal complications should be considered such as hydramnios, scheduled cesarean section, redundant cesarean section, induction of labor, shoulder dystocia and ante/postpartum hemorrhage [16].

For women for whom termination of pregnancy is not an option, it is important that the clinician be informed of the mother's risk factors, such as obesity, diabetes, hypertension, autoimmune diseases, and provide the best method of delivery. Monitoring for the above mentioned complications is mandatory [17].

Conclusion

Anencephaly is not compatible with life. The most important aspect of managing this condition is prevention. The simplest way to reduce the incidence of anencephaly is to advise women of childbearing age to take a folic acid supplement. Any dose of 0.4 mg or more per day is effective; this is especially important for any woman taking anticonvulsants. For a young patient with epilepsy, counseling is essential regarding the risk of seizures during pregnancy for the developing fetus and the risk of teratogenicity. Valproate should be avoided. The anticonvulsant with the best teratogenicity record is lamotrigine.

Maternal and fetal ultrasound are diagnostic procedures during pregnancy for in utero diagnosis of any neural tube defect, including anencephaly. Early termination of pregnancy is proposed upon diagnosis of anencephaly.

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