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Anencephaly: Late Diagnosis and Management in a District Hospital in Bamako, Mali—Case Report

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Abstract

Anencephaly is a defect in the closure of the neural tube. It is a congenital anomaly characterized by the total or partial absence of the cranial vault, the brain being reduced to a small mass. Most of the cases are stillborn, but newborns have been observed to survive from a few hours to a few days. Its prevalence at birth is between one (1) case per 5000 and one (1) case per 2000 live births. Prenatal diagnosis can easily be done by using ultrasound from the first trimester of pregnancy, given the absence of visibility of the cranial vault. We report a case of anencephaly in a poorly monitored diabetic multiparous woman, discovered late after an ultrasound scan at 30 weeks' gestation. Delivery was spontaneous at 34 weeks' gestation at the same centre, resulting in a female infant with an APGAR score of 6, presenting with anencephaly and surviving for 15 minutes. The occurrence of such a pregnancy constitutes a high-risk situation that can lead to maternal and neonatal morbidity and mortality. But, early diagnosis by ultrasound and good antenatal care can improve the situation.

Keywords

Ultrasound, Antenatal Diagnosis, Anencephaly, Congenital Malformation, Mali

1. Introduction

Anencephaly is a serious malformation of the central nervous system (CNS) [1]. It is a monstrosity characterized by the absence of an encephalon [2] and results from a defect in the closure of the neural tube. The foetus is left with part of the encephalon (brain, cerebellum and brain stem) missing [3].

The prevalence is between one (1) in 1000 and one (1) in 2000, with geographical variations attributable to different genetic heritages in different populations, as well as to dietary factors which may include folic acid deficiency [4] [5]. The aetiologies are multifactorial (iatrogenic, toxic, metabolic, nutritional and rarely chromosomal). Most often, anencephaly is an isolated malformation (in 80% of cases), but in rare cases, it is associated with other malformations such as spina bifida and other NTDs. The risk of recurrence was between 1.7% and 5% [5]. Diagnosis is made by 1st trimester ultrasound between the 11th and 14th week of amenorrhoea (SA) [6].

In the absence of hydramnios, anencephaly is generally accompanied by an overdue pregnancy. However, when associated with hydramnios, it leads to premature delivery [7]. Fetal stress occurring at the end of pregnancy, is linked to the peak in the production of corticotropin releasing hormone, which is considered to be the placental clock. Spontaneously, it triggers labour by stimulating the corticotropic axis [8].

However, this mechanism is lacking in the anencephalic foetus due to the failure of secondary adrenocorticotropic hormone synthesis to hypothalamic-pituitary aplasia [9].

Anencephaly does not allow long survival [10].

In Mali, work has been carried out on NTDs in general, but few studies have focused specifically on anencephaly [11].

2. Observation

The patient was seen for an antenatal consultation in the department on July 08th, 2024. She was a 41-year-old pregnant woman. She is a grand multiparous and in her eighth gestation. Seven children live healthily, three boys and four girls. She lost her husband in February as a result of a professional accident. She is a housewife, known diabetic two years ago and she is irregularly monitored with incorrect administration of insulin. The interview did not reveal any other particular medical, surgical, obstetrical or family history. She lives in a peri-urban area of Bamako in some poor socio-economic conditions. She had two antenatal consultations on July 08th, 2024 and August 19th, 2024. No prophylaxis was taken: no iron-folic

acid supplementation or anti-tetanus vaccination, no anti-malarial or anti-helminth prophylaxis. A single ultrasound scan was performed at the District Hospital of Commune V, in the district of Bamako on July 21st, 2024. concluding in a progressive monofetal pregnancy estimated at about thirty (30) weeks with a partial absence of the cranial vault, suggesting anencephaly with moderate hydramnios (Figure 1).

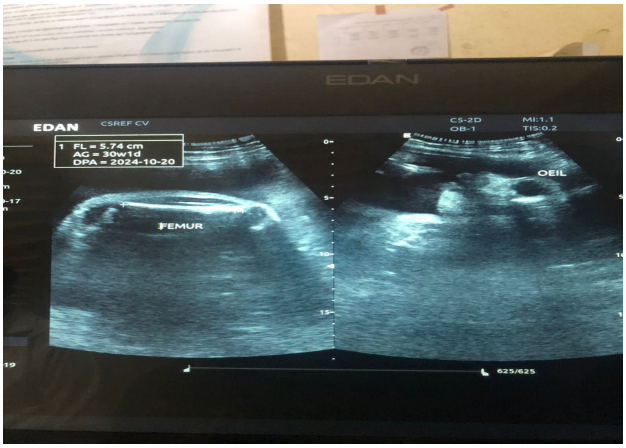

Figure 1. Double ultrasound image showing a cranial cavity and a femur.

She was tactfully informed and an appointment was given the following day with the psychologist, but the patient did not return despite repeated telephone calls.

She consulted the department, on August 19th, 2024, for painful uterine contractions. The diagnosis of active labour was done on the basis of regular contractions associated with dilatation of the cervix to 5 cm, with the sac intact. The presentation was full breech. Fetal heart sounds were regular at 136 beats per minute.

Labour was correctly monitored using the programme and resulted in the birth of a live female newborn within three hours, with a Virginia APGAR score of 8 at the first (1) minute and six (6) at the fifth minute, after a correct practice of essential newborn care (Figure 2).

Figure 2. Newborn at 5 minutes from birth.

She weighed 2450 grams with a height of 45 cm, a head circumference of 21 cm and a chest circumference of 30 cm. The cord measured 51 cm and the placenta weighed 470 grams. The amount of amniotic fluid was estimated at 2300 ml.

Clinical examination revealed a complete absence of the cranial vault, leaving a small amount of cerebral matter on the outside (absence of the brain and part of the cerebellum), making it difficult to clean the newborn. The forehead was crushed and receding backwards. The superciliary arches were prominent and separated by a very marked depression. The length of the face, the flattening of the skull and the protrusion of the eyes gave the head the general appearance of a batrachian. The eyes were globular. The eyeballs were prominent and exorbitant, like large marbles placed inside the eyelids, which were oedematous and very large. The cheeks were large. The ears were large, badly hemmed, low set and split at the top, the tongue was large and filled the mouth. The neck was very short, almost absent, with shoulders very close to the head. The diagnosis of anencephaly was accepted. There were no other visible physical malformations. She was hypotonic overall, with a temperature of 35.6 degrees. Her unstable condition meant that she could not be transferred to the neonatal unit.

She died 15 minutes after birth.

She was presented to her mother, who began to scream loudly, squaring off with the other patients on the ward. A student midwife on a placement in the maternity unit quickly left the room out of fear for the newborn. They were referred to the psychologist and subsequently improved.

3. Discussion

Anencephaly is a rare congenital malformation. Its incidence is 1/1000 births [11]. In Mali, studies by Maiga B [12] and Sidibé A [13] found respective frequencies of 12 anencephaly out of 90 neural tube defects and 5 anencephaly out of 41 neural tube defects.

Our patient was 41 years old. Studies concerning the influence of maternal age on the occurrence of NTDs including anencephaly were contradictory [14].

She was a grand multiparous with her eighth gestation and seven living children. Multiparty is also a risk factor. It seems to be associated with advanced maternal age [4].

Our patient did not take folic acid either in the preconceptional period or during pregnancy. It has been estimated that 4% - 5% of children with severe congenital anomalies are born to mothers with folic acid deficiency [5] Folic acid supplementation is no longer necessary (with the aim of preventing NTFAs) beyond the 1st trimester of pregnancy [4].

Previous studies have reported a wide range in the percentage of fetuses with associated malformations [5] [6] [11].

The malformation in our case was diagnosed at the 30-week ultrasound. Despite late discovery, the most common neural tube defects diagnosed at birth remain spina bifida and anencephaly. The latter does not allow for long survival.

However, the recommendation that all women wishing to become pregnant should take folic acid supplements is proving unsuitable to a large proportion of the population [10].

Hydramnios was associated with pregnancy in our patient.

In France and the USA, NTDs are diagnosed antenatally in 100% of cases in the first trimester, and in 60% of cases in the second trimester in Canada. Antenatal diagnosis is based on obstetrical ultrasound in the first trimester. The sonographic signs of anencephaly are the absence of frontal bones above the orbital frames and the absence of brain tissue in the anencephaly, depending on the definition used. Hydramnios is very often associated with anencephaly [11]. This may explain the spontaneous and premature onset of labour in our case.

Anencephaly represents the most severe form of neural tube closure disorder, uniformly lethal in the neonatal period, and it is one of the most lethal of congenital malformations.

Moreover, the prevalence of anencephaly is greater among spontaneously aborted fetuses than among those born at term, indicating an in-utero "selection" directed against fetuses affected by this malformation. On the other hand, full-term pregnancies are not uncommon. These are usually girls. The newborn may live for a few hours or even a few days.

The presence of hydramnios on third-trimester ultrasound is justified by the fact that, at the end of pregnancy, fetuses with anencephaly have swallowing problems. Jaquier *et al.* in a 2006 study found that 6/211 survived from 6 to 28 days at most. Minor psychological disorders were noted in our patient, who is a widow, and in a health worker: this could be explained by the monstrous morphological appearance of the anencephalic newborn, which could frighten a person unaccustomed to seeing them. This aspect has also been found in the literature, with reports of puerperal psychosis.

4. Conclusion

Anencephaly is a serious malformation. It is incompatible with life. Early folic acid supplementation and timely ultrasound can improve the situation.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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