

CASE REPORT

Anencephaly: pitfalls in pregnancy outcome and relevance of the prenatal exam

V. I. TICA^{1,2)}, M. BEGHIM²⁾, IRINA TICA^{1,3)},
M. ZAHER^{2,4)}, ESRA BEGHIM⁴⁾

¹⁾Faculty of General Medicine, "Ovidius" University, Constanta

²⁾Regional Emergency University Hospital, Constanta

³⁾Transports University Hospital, Constanta

⁴⁾Private practice

Abstract

Anencephaly is an embryological malformation of the central nervous system, invariably lethal, characterized by the absence of the brain and cranial vault and by other defects of the cranial structures. It has no cure but it can be detected during the pregnancy with ultrasonography. We present a rare case of a 15-year-old primipara, with no prenatal care, who delivered at 37 weeks an anencephalous female newborn. Ultrasonography avoided the confusion with a breech presentation, which, in conjunction with the prematurity, premature rupture of membranes and the young maternal age would have favored the cesarean section. The rarity of the case is increased by the singularity of the malformation. The importance of the prenatal care and genetic workup are stressed out in this embryologic pathology.

Keywords: anencephaly, ultrasonography, prematurity, membrane premature rupture pregnancy, prenatal.

☞ Introduction

From the embryological perspective, anencephaly results from a failure in the neural tube closure. It is a relatively rare anomaly – up to one in 20 000 infants [1].

The condition has been known since antiquity; an important role in the description and understanding of this severe congenital malformation is attributed to Etienne Geoffroy Saint-Hilaire (1772–1844) and to his son, Isidore, who is considered the founder of teratology [2].

The newborn may be blind, deaf, unconscious and usually dies during the birth or within a few hours [3].

As the primary concern of the obstetrician has always been the good evolution of the pregnancy, and the delivery in good conditions of a healthy newborn, an early diagnosis during the gestation is essential in order to advocate the termination of pregnancy, and to avoid the birth of such a child with all its implications for the parents and for the society.

The introduction of the ultrasonography in the obstetrical practice had substantially improved the survey of the pregnancy and the prenatal diagnosis of the malformations, allowing a prompt and precocious management of such cases.

We present an interesting, rarely case of anencephaly with no other associated malformations in which the obstetrical decision was almost misguided by the resemblance with another totally different situation and by the lack of any prenatal medical care and, therefore, proper diagnosis.

☞ Patient and Methods

A young, 15-year-old pregnant woman was admitted in our Department with abdominal pain and loss of amniotic liquid. From her history we found out that it was her first pregnancy, she came from a village, she was not married, she had poor education (she did not finished the school) and socio-economical condition, and she did not look for any medical consultation during her pregnancy. The clinical examination on admission favored the diagnosis of primigesta, primipara, 37 gestational weeks, singleton pregnancy with a live fetus in breech presentation, ruptured membranes, and labor. The patient had several relative indications for the cesarean section: very young nulliparous women with a fetus in breech presentation, possible prematurity (as the gestational age was not certain) and premature rupture of membranes.

For a better appreciation of the fetal weight, flexion of the cervical spine and maturity, an obstetrical ultrasonography was decided and revealed an anencephalous fetus in occipital presentation. In the view of this information, the management strategy was changed and the decision was to assist the vaginal delivery. After another 5 hours, the women gave birth to a female live newborn, with a weight of 2600 grams, Apgar score 1, with anencephaly (Figure 1). One hour after the birth, the newborn suffered an irreversible cardio-respiratory arrest. The patient had a normal puerperium. Ablactation was medically induced.

The pathological exam confirmed the clinical diagnosis by showing: anencephaly; athelectasy and lung bleeding; liver congestion; spleen dystrophy; myocardium dystrophy; IInd degree of prematurity.



Figure 1 – The anencephalous newborn with no other abnormality.

Discussion

Isolated neural-tube defect are, after cardiac defects, the most common congenital structural defects [4]. Anencephaly is the most severe defect from the most common neural-tube anomalies (spina bifida, cephalocele, anencephaly) [4]. Embryologic abnormality and the occurrence with a rate of 1:1000 to 1:20 000 infants [1], the anencephaly is an infrequent malformation of the central nervous system. It is present at the 28th day of conception and it is found at the time of birth almost every time. This is why it can be diagnosed by ultrasonography in early stages of pregnancy, even from the 12th week of amenorrhea [3]. In our case, because of the lack of any examination during the pregnancy, this opportunity was lost.

Like other forms of neural tube defects, anencephaly has a multifactorial pattern of transmission, with the interaction of environmental and genetic factors. In our case, both parents of the malformed child came from the same village and had the same low socio-economical status, with a mostly agricultural activity. There are reports in favor of the relation between the exposure of both parents to the agricultural work and an increased risk for anencephaly [5]. In addition, there are reports concerning the relation between the exposure of the mother to the passive smoking and the risk of a neural-tube defect; because of their living along with the paternal family where the smoking is usual [6].

Another particularity of the case that we report is that it does not match with any of the other known risk factors: older mother [7], maternal miscarriage history [8], twin pregnancy [9], maternal obesity, and gestational diabetes [10].

A supplementary information to be stressed out – and, thus, contributing to its rarity – is that in our case the anencephaly was a solitary birth defect without other associated malformations, like it is generally found in literature: spina bifida with/without meningocele (27%), urinary tract defects (16%), cleft lip and palate (10%), digestive tube defects (6%), heart defects (4%) and others [11]. It could also appear as part of a polymalformative syndrome, like the chromosomal defects (13th or 18th trisomia, triploidias) [11].

The anencephaly is associated with a multitude of pregnancy complications – and we have experienced

quite a few of them: premature rupture of membranes, abnormal pregnancy length (prematurity), hydramnios and fetal death. All those are described in the literature, as the sex ratio (male to female) ratio is <1 [12]. Although abnormal fetal presentation may be frequent, this was not the case. However, this matter is significant for the patient, as, because of the malformation, an initial misdiagnosis could have resulted in an unnecessary and deleterious surgical procedure. Some would again advocate in favor of the necessity of ultrasound examination on admission, especially with an unclear gestational age or presentation. An important role in the diagnosis of these and other malformations is belonging to ultrasonography. Besides the evident positive ultrasonographic diagnosis, there are studies that suggest that an echogenic amniotic fluid is a marker for first-trimester fetal acrania, condition that turn into anencephaly by the destruction of the unprotected brain [13]. For the diagnosis, it is also useful, like in all neural-tube defects, to determine the level of alpha-fetoprotein in the maternal blood that is elevated [14].

The differential diagnosis of the anencephaly (essentially by the ultrasound exam) is to be made with:

- the amniotic loop syndrome: it is usually associated with others abnormalities, like limb or fingers amputation, spinal defects and ventral abdominal wall defects. It is usually associated with oligoamnion while the anencephaly is associated with polyamnion;
- a big encephalocele: the cranial bone is very clear observed and present but is having a discontinuity (not absent like in the anencephaly);
- the microcephaly: the cranial bony outline is visible, it has no discontinuity, but it is of small dimension (the cranial ultrasound parameters can be measured and are reduced);
- the iniencephaly: the typical appearance of the cervical spine labeled retraction allows the diagnosis [11].

Anencephaly is a lethal condition, so heroic treatment maneuvers are not indicated after the birth of such an infant (although there is a recent paper that not agrees with this [15]), and so was our management. There is, neither, any cure for it; hence, the medical team efforts had been focused on providing supportive environment for the parents. The parents had been also referred to a genetic counselor.

The prevention measures are therefore very valuable, and an important role seems to belong to the diet supplementation with folic acid before pregnancy and in the first month [16, 17]. This attitude has been associated with a decrease in both the frequency and in severity of the condition [18]. Another measure to be used is the fortification of both wheat and maize flour with folic acid [19]. A secondary line of prevention is to detect the abnormality as soon as possible during the pregnancy, obtained by the implementation of the program of the prenatal diagnosis [20].

All those preventive methods are settled to avoid the phenomenon presented by us in this paper, the late presentation of gross congenital anomaly of the nervous system, a situation characteristically for the developing nations, situation associated with many complications (both physical, of the pregnancy and the labor, and psychological, for the parents). To avoid this situation health education should include issues regarding congenital malformation delivered by trained experts [21].

☐ Conclusion

The case presented in this paper is relevant by its rarity – the singularity of the anencephaly with absence of any proven risk factor – and by its obstetrical consequences. Ultrasonography clarified a difficult differential diagnosis, helped in choosing the appropriate medical strategy, and in avoiding unnecessary and deleterious decisions. Appropriate prenatal care and, in appropriate cases, genetic workup would decrease the risk of this serious embryologic pathology for the fertile population.

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Corresponding author

Vlad I. Tica, Associated Professor, MD, PhD, 1st Obstetrics–Gynecology Clinics, Emergency County Hospital, 143 Tomis Avenue, 900591 Constanța, Romania; Phone +40241–503 189, +40241–503 218, e-mail: vladtica@datanet.ro

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