



ANENCEPHALY: A CASE REPORT AND REVIEW OF THE LITERATURE

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Article Received on 09/04/2022

Article Revised on 30/04/2022

Article Accepted on 20/05/2022

ABSTRACT

Anencephaly is a congenital malformation of the nervous system of the fetus, which occurs due to a defect in the closure of the neural tube. The fetus is left with a part of the brain (brain, cerebellum and brain stem) missing. Anencephali can be diagnosed by ultrasound in the first trimester. Therapeutic termination of pregnancy has been recommended in developed countries since the 1990s and the birth rate of anencephaly is almost zero in these countries. This makes it possible to avoid giving birth to a newborn with a severe handicap, a difficult choice for the parents, but it is important to take into account an important fact: anencephalic newborns survive only a few hours after birth and are often blind, deaf, cannot feel pain and remain unconscious. This observation was made at the Ibn Rochd Hospital in Casablanca, Morocco, at the Lalla Meriem Maternity Hospital in a case of anencephaly whose parents refused to terminate the pregnancy. The interest of this work was to underline the heavy psychological handicap that this pathology has on the parents, hence the interest of genetic counselling and folic acid prophylaxis in women at risk.

KEYWORDS: Anencephaly; congenital malformation; fetus.

INTRODUCTION

Anencephaly is a congenital malformation of the nervous system of the fetus, which occurs as a result of a defect in the closure of the neural tube. The fetus is left with a part of the brain (brain, cerebellum and brain stem) missing. It is a rare malformation with a prevalence of 1 in 1000, with geographical variations as well as dietary factors that may include folic acid deficiency.^[1] This disease generally affects the fetus from the first month of pregnancy and results from disruption of neural tube closure.^[2] Anencephaly can be diagnosed by ultrasound in the first trimester.^[3] Therapeutic termination of pregnancy has been recommended in developed countries since the 1990s and the birth rate of anencephaly is almost zero in these countries.^[4] This makes it possible to avoid giving birth to a newborn with a severe handicap, a difficult choice for the parents, but it is important to take into account that anencephalic newborns survive only a few hours after birth and are often blind, deaf, cannot feel pain and remain unconscious.^[2,3] The interest of this work was to underline the heavy psychological handicap that this pathology has on the parents, hence the interest of genetic counselling and folic acid prophylaxis in women at risk.

CASE REPORT

This is Mrs E.F aged 28 years, second gesture second pares: a child living by vaginal delivery with no notion of consanguinity or particular pathological history; during the first prenatal consultation the diagnosis of anencephaly was made on the first trimester ultrasound: Figure 1. The family refused therapeutic termination of the pregnancy. The delivery took place by the basic route giving birth to a live polymalformed newborn of sex F weighing 2650g; motivating its transfer to the paediatric neonatology department; On admission the newborn presented hypothermia at 35.1°C, a bradycardia at 69 beats/minute. Neurologically, the state of wakefulness was altered, the archaic reflexes were absent, with axial and peripheral hypertonia. Pulmonary distress was noted with 45% desaturation. Morphological examination: Figure 2: noted a total absence of the cranial vault, leaving a small amount of cerebral matter (absence of the brain and part of the cerebellum). The orbits were directed upwards, with prominent superciliary arches and bulging, globular eyes (figure 1). The forehead was crushed, receding backwards. The length of the face, the facial angle, the protrusion of the eyes, the flattening of the skull gave the head the general appearance of a batrachian. The neck was short, the ears low set and badly hemmed, the tongue protruding. No other malformation was associated. The diagnosis of anencephaly was retained.



Figure 1: Anencephaly in the first trimester.



Figure 2: Morphological Appearance.

DISCUSSION

Anencephaly is the most severe form of neural tube closure disorder, uniformly lethal in the neonatal period, and is one of the most lethal of all congenital malformations.^[5] The term "neural tube defects" (NTDs) refers to a group of congenital malformations resulting from a neural tube closure defect of varying extent in the fourth week of embryonic development.

Epidemiological studies of NTDs have led to the suspicion of certain risk factors such as: Factors linked to seasonal variations were observed in Great Britain between 1940 and 1958, with a peak in the frequency of children conceived in spring and a drop in the frequency of children conceived in autumn. These seasonal variations have only been found in Great Britain^[6];

geographical origin; folic acid deficiency; maternal age and multiparity are also risk factors^[7]; the frequency of anencephaly is greater in the case of twin pregnancies than in the case of a mono-fetal pregnancy. However, it is very rare for both twins to be affected, whether the pregnancies are mono- or dizygotic, which implies that factors other than genetic factors are involved.^[8]

Antenatal diagnosis is based primarily on first trimester obstetric ultrasound. The ultrasound signs of acrania/anencephaly are the absence of frontal bones above the orbital frames and the absence of brain tissue; in anencephaly, according to the definition, the cranial vault is partially present, there is disorganised brain tissue, with a possible meningeal envelope. This ultrasound, combined with the determination of alpha-

fetoprotein in the maternal serum (between 14 and 18 weeks of age), allows prenatal diagnosis of anencephaly in almost 100% of cases.^[3]

The treatment of anencephaly is preventive and nowadays based on medical termination of pregnancy. It seems that this option has become common since the 1990s in many developed countries, bringing the birth rate of anencephaly down to zero.^[4] Currently in France, acrania results in medical termination of pregnancy in all cases. The rare pregnancies that are continued correspond to the parents' choice. According to the Paris Congenital Malformations Registry, the prevalence of anencephaly among live births rose from 0.6 in 1981 to 0 per 10,000 in 2000 (with 100% antenatal diagnosis and 98.6% IMG in the period 1997-2000).^[9]

In terms of the prognosis of anencephaly, survival at birth is only a few minutes to a few days (24 hours to 6 days). Jaquier et al in a 2006 study found that 6/211 survived between 6 and 28 days at most.^[10] In our case the newborn survived 4 hours. Primary prevention of neural tube defects (anencephaly being one of the most severe forms) by taking folic acid during the periconceptional period has proven to be effective.^[4]

CONCLUSION

Prenatal diagnosis of anencephaly provides better information to parents and allows consideration of medical termination of pregnancy. A campaign to raise awareness among practitioners and patients of childbearing age of the need for folic acid supplementation from the beginning of pregnancy to prevent recurrence or the occurrence of acrania/anencephaly is important. It is also important to legislate on the medical termination of pregnancy in order to avoid psychological trauma to our parturients who are likely to give birth to polymalformed newborns.

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