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Anencephaly: A Case Report and Review of the Literature

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Abstract: *Anencephaly is a lethal malformation characterized by a lack of closure of the anterior neuropore of the neural tube with aplasia of the cortical architecture and absence of cranial box formation. Anencephaly is an isolated malformation (in 80% of cases), but in rare cases it is associated with other malformations such as spina bifida and others. Anencephaly represents 40% of neural tube defects, which is the second most common cause of nervous system anomalies after spina bifida. The prevalence rate is 1/1000. The diagnosis is made by the ultrasound of the first trimester, a prophylaxis of neural tube defects with folic acid in the periconceptional period is recommended. We are going to present a male neonate born by vaginal delivery in our department. In these antecedents, we note a low socio-economic level, a maternal age of 34 years, primigravida, with consanguinity of 1st degree and absence of prophylaxis with folic acid during the periconceptional period. The diagnosis of anencephaly was retained. The evolution was marked by the death of the newborn after two hours of life. Antenatal diagnosis of congenital malformations incompatible with extra uterine life allows parents to be informed about the risk of recurrence and the benefit of folic acid supplementation before the next pregnancy.*

Keywords: anencephaly, prenatal diagnosis, folic acid

Introduction:

Anencephaly is a congenital lethal malformation characterized by a lack of closure of the anterior neuropore of the neural tube with aplasia of the cortical architecture and absence of cranial box formation.

The prevalence is 1 case per 1000, with geographical variations attributable to different genetic heritages in different populations, as well as to dietary factors that may include folic acid deficiency [1].

First trimester obstetrical ultrasound remains the reference imaging for prenatal diagnosis.

anencephaly results from a neural tube closure defect between the third and fourth week of fetal development

The prognosis for newborns with anencephaly is extremely poor. If the baby is not stillborn, it will usually die within hours or days of birth.

Very interesting to underline the heavy psychological handicap of this pathology for the parents in case of recurrence, hence the interest of genetic counseling.

Primary prevention of neural tube defects by taking folic acid during the periconceptional period has proven to be effective not only in preventing a possible recurrence after the birth of a first affected child or fetus, but also in the general population to reduce the risk of occurrence of these malformations.

Observation

The patient was 34 years old and primigravida. The history had noted an unfavorable socioeconomic level, with consanguinity of 1st degree and absence of prophylaxis with folic acid during the periconceptional period, admitted for an abdominopelvic pain type of uterine contractions, normo carde normo tendue, an obstetrical ultrasound was realized returning in favor of a progressive mono fetal pregnancy with anencephaly (Figure 1).

The delivery was done by vaginal route and with the extraction of a malformed male newborn weighing 0250g, the newborn examined by the pediatrician, presented a temperature of 35,6°C, a bradycardia of 90 beats/minute. Neurologically, the state of alertness was altered, the archaic reflexes were absent, with axial and peripheral hypertonia. On the pulmonary level, a respiratory distress syndrome was noted with a desaturation of 75%. The morphological examination had noted a total absence of the cranial vault allowing a small part of cerebral matter to be seen (absence of the brain).

The orbits were directed upwards, with bulging and globular eyes (figure 2). The forehead was crushed, the protrusion of the eyes, the flattening of the skull gave to the head the general aspect of a batrachian. The neck was short, the ears low implanted.

In addition, no other malformation was associated. The diagnosis of anencephaly was retained.

The course of action was oxygen therapy, rewarming and venous access with the introduction of 10% glucose serum. The evolution was marked by the death of the newborn (2 hours of life).

Figure 1: aspect of the head in batrachian

Figure N° 2: anencephaly with a batrachian aspect of the head

Discussion

Anencephaly is a congenital malformation of the central nervous system that results in the partial or total absence of the encephalon, cranium and scalp. Anencephaly is an embryopathy that occurs during the 3rd and 4th week of intrauterine life and is characterized by congenital defects in the formation of the central nervous system. They represent 5% of congenital malformations. It affects 1.4 per 1000 births [2]. It is sometimes associated with spina bifida and encephalocele [3].

The origin of these malformations is multifactorial, with interaction of environmental and genetic factors (4)

The pathogenesis of neural tube defects (NTDs) remains unclear and controversial.

Various factors are considered as etiological factors, such as hyperthermia, valproic acid, fenugreek, hypervitaminosis A, folic acid and vitamin B12 deficiency during the periconceptional period and genetic factors [5-6].

The prevalence of anencephaly shows a great variation in time and space. It is 1 per 1000 in France [1], 12 per 10000 in Iran, 10.4 per 10000 in China, 0.01 to 7.42 per 10000 in Rijeka, Croatia, 1.49/1000 in Santos Dumout [7], 0.5 to 0.6 per 1000 in Singapore [8] and 0.5 to 6.5 per 1000 in India. [9-10], 7 to 8 per 1000 births in Ireland ,and it is 1 to 2/1000 in the USA and continental Europe.

In addition to geographical variations, there are ethnic variations in the frequency of anencephaly. For example, in the United States, the highest frequency is observed in Caucasians (2.8/10000) as opposed to other races (2.1/10000) [11].

Seasonal variations were observed in Great Britain between 1940 and 1958, with a peak in the frequency of children conceived in spring and a decrease in the frequency of children conceived in autumn. These seasonal variations were only found in Great Britain [12]. This may suggest the influence of exogenous dietary or climatic factors, but these are still only hypotheses.

The female sex is more affected by anencephaly with a percentage of 65.8% and a sex ratio of 0.3/1.

For most authors, the preferred age is between 25 and 30 years.

In the Panduranga et al. series [13], maternal age ranged from 19 to 28 years with a mean age of 23 years.

In the study by Shilpa et al [14], there was a variation between 20 and 40 years of age, with an average of 24.4 years.

Multiparity is also a risk factor [15]. The frequency of anencephaly is greater in twin pregnancies than in mono-fetal pregnancies. On the other hand, it is very rare that both twins are affected, whether the pregnancies are mono- or dizygotic, which implies that factors other than genetic factors are involved [16].

First trimester anencephaly diagnosis is the most successful
for fetal anomalies and is based on ultrasound.
the typical appearance of anencephaly is the "frog's eyes" sign, due to the absence of visible brain tissue above the orbits. being the absence of frontal bones above the orbital frames, according to the definition, the cranial vault is partially present, there is disorganized brain tissue, with possible meningeal envelope.
This ultrasound associated with the dosage of alphafoetoproteins in the maternal serum (between 14 and 18 SA), allows the prenatal diagnosis of anencephaly in almost 100% of cases.
Hydramnios is associated with anencephaly in 30% to 50% of cases. Factors involved include CSF secretion into the amniotic cavity, lack of normal swallowing, lack of absorption of amniotic fluid by the hypoplastic lungs, and excessive urine production due to lack of antidiuretic hormone.
Hydramnios is the most common presentation of anencephaly prior to delivery.
Anencephaly is associated with abnormalities not only of the central nervous system but also of other systems. In the newborn presented in the observation he had no associated pathology.
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The treatment of anencephaly is preventive and nowadays based on medical termination of pregnancy. It seems that this option is has become common since the 1990s in many developed countries, bringing the birth rate of anencephaly down to zero [17]. Currently in France, anencephaly results in a medical termination of pregnancy in almost all cases. The rare pregnancies that are continued correspond to the parents' choice. According to the Registre des Malformations Congénitales

de Paris, the prevalence of anencephaly among live births went from 0.6 in 1981 to 0 per 10,000 in 2000 (with 100% prenatal diagnosis, and 98.6% IMG in the period 1997-2000) [18].
Prognosis Anencephaly is a uniformly lethal anomaly.
According to the M. Jacquier et al. study [19], out of 211 cases of anencephaly, there were 15 cases of MFIU, 43 cases of intrapartum death and 153 live births. Of the 153 live birth cases, 103 (67%) died within 24 hours and 41 (28%) within 1 hour. The longest survival was 10 days (04 cases), 18 days (01 case) and 28 days (01 case). In our case the newborn survived 2 hours.
In these situations of prenatal diagnosis, the psychiatrist has a fundamental role.
If it does not seem desirable to follow a constraining and dehumanizing pre-established protocol, it is nevertheless important that the meeting with the psychiatrist be thought out and proposed according to the availability of the woman and the couple. This meeting, and particularly the first interview, is fundamental.
Primary prevention of neural tube defects by taking folic acid during the periconceptional period is necessary. Neural tube defects can occur before women know they are pregnant, so they are advised to consume folic acid-rich foods at least 3 months before planning a pregnancy, and to maintain an adequate intake if they are of childbearing age or likely to become pregnant.
It is estimated that half of all birth defects are preventable if women of childbearing age consume adequate amounts of foods rich or fortified with folic acid, or by taking vitamin supplements [20]. Predictably, rates of neural tube closure defects are higher in communities where women suffer from impoverished diets.
As a result of primary and secondary prevention studies, Canadian, British and American organizations recommend that women of childbearing age consume 0.4 mg to 0.8 mg/day of folic acid to reduce anencephaly. For women who already have an affected child, the recommended dose is between 0.8 and 4 mg.
In France, the French Pediatric Society recommends 0.2 mg/d for women without risk.

Conclusion

Prenatal diagnosis of anencephaly offers better information to parents and ensures optimal care.
A campaign to educate practitioners and patients of childbearing age about the need for folic acid supplementation before conception to prevent recurrence or occurrence of anencephaly is important.

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