EXTREME CEREBRAL MALFORMATION – CLINICAL AND ETHICAL ISSUES BASED ON A CASE

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ABSTRACT

Hydranencephaly represents one of the most rare cerebral malformations, with poor prognosis and negative impact on both family and society. Usually the patients remain in a permanent vegetative state and require unlimited medical care. Although the majority of affected babies die in utero or in the first days-months of life there have been cases in literature with prolonged survival, for 20 (1), 22 (2) and 32 (3) years. Therapeutically, the medical care provided are purely palliative and oriented to supporting vital function.

Conclusions. Hydranencephaly is a rare brain malformation with poor prognosis, life expectancy and quality of life. Diagnosis imposes a number of ethical issues. The importance resides in the fact that prenatal detection represents an indication for therapeutic abortion. Another ethical issue raised is represented by medical care provided, which is exclusively palliative. Both surgical and novel therapeutic methods described by the literature, like endoscopic coagulation of choroid plexes, do not influence the neurological status and the neuropsychomotordevelopment (4). All this data should be clearly explained to parents and legal tutors for not raising any false hopes regarding the evolution.

Keywords: hydranencephaly, transfontanellar ultrasound, peri/intraventricular hemorrhage, prematurity

INTRODUCTION

Hydranencephaly is defined as the absence of the cerebral, hemispheres, the remaining space being filled with cerebrospinal fluid (CSF) (4). The skull and meninges are intact and most cases present with normal diencephalon and posterior fossa structures (4,5). Some times the occipital lobes can be present (4). Etiology and pathogenic mechanism are still unknown. Among the existing hypotheses, bilateral occlusion of the fetal internal carotid artery seems to be the key element (6). Other discussed causes are: intrauterine infection, toxics, iatrogenic, genetics and traumatic events produced in the first pregnancy trimester (4). Due to both increased production of CSF from the choroid plexus and decreased absorption , the intracranial pressure increases with secondary growth of the head circumference (HC). Standard treatment is represented by CSF drainage accomplished through serial ventricular/lumbar taps or by a ventriculoperitoneal or ventriculoatrial shunt. All these procedures are purely palliative and do not influence the neurodevelopment including the cognitive function. This patients maintain only the vegetative nervous system functions, being in a permanent vegetative state.

CASE PRESENTATION

A 33 days premature baby boy, BW=1,300 grams, Apgar Score 5/1 min., born by vaginal delivery at 33 weeks, from an investigated pregnancy is admitted to our hospital for rapid increasing HC. Clinical: Severe general condition, «sun setting eyes», macrocephaly (Fig.1). with HC=37 mm (P>97), moderate respiratory distress, Sp.O2=91-

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FIGURE 1. *Macrocephaly with clinical hydrocephaly features – "sunset eyes", viscerocranium extremely small comparing with neurocranium.*

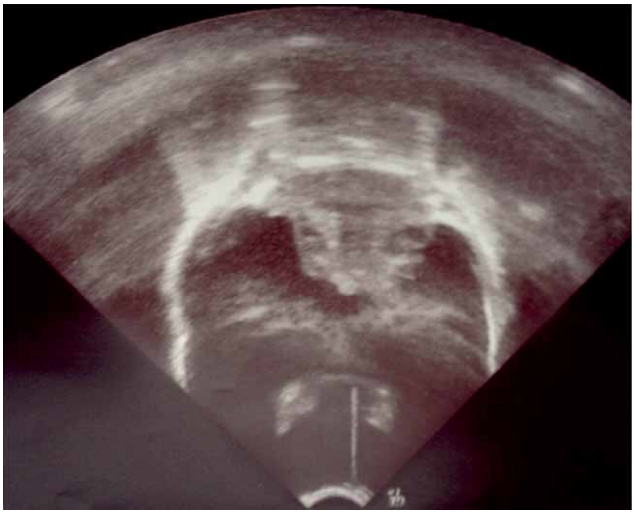

FIGURE 2. *Transfontanellar echography – coronal section. Echographic aspect of hydranaencephaly.*

92% with suplimentar oxygen administration, HR=170-180/min. Neurological: generalised hypotonia, dehiscent cranial sutures, large, bulging anterior fontanelle, 7/3 mm, weak cry, present but decreased archaic reflexes with present sucking reflex but requires assisted feeding (nasogastric tube).

Transfonatnellar ultrasound (Fig. 2) shows dilated lateral ventricles with loss of the cerebral parenchyma, visible choroid plexes, present basal nuclei and posterior fossa structures but compressed by the ventricular dilatation. Transsonic, impure intraventricular fluid sugesting ventriculitis. The IRM exam revealed the supratentorial cerebral parenchyma almost compleatly repalced by cystic cavities with mixed content-sugesting CSF and blood, along with restant bilateral temporo-basal, right fronto-basal, left medio-basal, left medio-basal and parieto-occipital parenchyma. The IRM exam confirms the head ultrasoud findings, suggestive for hydranencephaly. The EEG revealed diffuse hypovoltage, amplitude < 20 microV. Normal Karyotype analisys-46 XY.

Under medical treatment with Acetasolamide, Furosemide and Mannitol, the HC grows with a medium of 4-5 cm/week requiering serial intraventricular taps with a mean of 20 ml CSF extracted/punction.

The baby experienced several infectious events successfully treated, with no need, at that time, for assisted mechanical ventilation. At the age of 7 months a CSF drainage method is became mandatory and a ventriculoperitoneal shunt is performed. This lead to a progressive decrease of intracranial pressure with secondary decrese of HC from 57 cm before the shunt to 49 cm 7 days after surgery

DISCUSSIONS

Hydranencephaly represents a rare, encephaloclastic cerebral malformation, with extreme gravity, often fatal, characterized by absence of cerebral parenchyma that is replaced with CSF and necrotic debris (7). Usually there is no cortex identified but a portion of the occipital lobe can be preserved (8). Thalamus, brainstem, basal nuclei, choroid plexes and cerebellum are usually present. Falx cerebri might be either present or partial/total absent. There are multiple ethiopathogenic hypotheses but the commonest etiology described is bilateral oclusion of the fetal internal carotid arteries with ischemia and hemorrhage in their territory (7,9).

The antenatal diagnosis can be established by ultrasound assesment at 21-23 week of pregnancy showing the absence of cerebral parenchyma. After birth the head ultrasound identifies the presence of cystic cavities that replace the cerebral cortex and occupies the whole skull (10). In most cases death happends in-utero and the postnatal survival rate is around 2 years. The survival of this patients is related to the integrity of the brainstem, which provides vital functions such as cardiorespiratory function and thermoregulation (11,12).

The type and quality of medical care should be considered in relation to the neurological status. This case can be framed as persistent vegetative state (13).

Children with hidranencephaly raises a number of ethical, medical and legal issues.

The right diagnosis of this condition is crucial for parents for correct understanding of the situation before deciding on medical or surgical treatment that can prolong survival with no influence on the neurological prognosis.

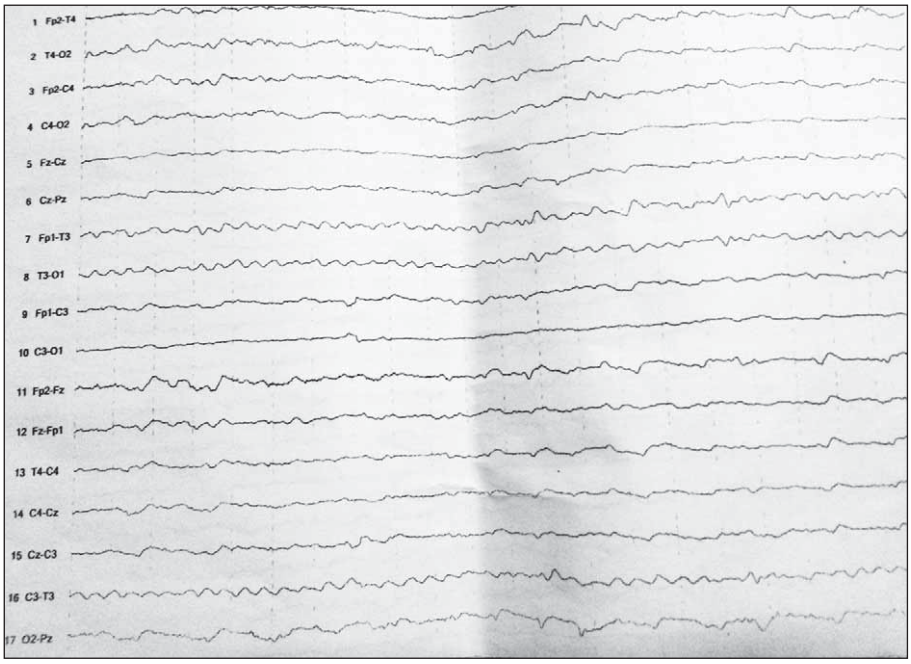

FIGURE 3. *Electroencephalography*

In terms of Romanian legislation, according to Law no 95/2006 (art. 376) and Law 46/2003 (art. 13 and art. 16), medical treatment is a legal right, as long as personal decision is not available and there are no patient's or legal carer written consent to stop medical maneuvers. The Code of Ethics for exercise of medical profession, Article 26 states: *"Incurable patient will be treated with the same care and attention as the one with chances of healing."*

CONCLUSIONS

Hydranencephaly is a rare brain malformation with poor prognosis, life expectancy and quality of life. Diagnosis imposes a number of ethical issues. The importance resides in the fact that prenatal detection represents an indication for therapeutic abortion. Another ethical issue raised is represented by medical care provided, which is exclusively palliative. Both surgical and novel therapeutic methods described by the literature, like endoscopic coagulation of choroid plexes, do not influence the neurological status and the neuropsychomotor development (4). All this data should be clearly explained to parents and legal tutors for not raising any false hopes regarding the evolution.

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