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Hydranencephaly: A Case Report and Ethical Considerations

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Abstract
Hydranencephaly is a rare congenital structural brain abnormality with a poor prognosis, consisting of complete or almost complete absence of the cerebral hemispheres with replacement of the cerebral parenchyma by cerebrospinal fluid. The evolution of a case diagnosed in the neonatal period in which the medical decision was made to provide palliative care and not undertake aggressive interventions is described. The most significant ethical considerations in terms of treatment and care of individuals affected by hydranencephaly are highlighted.

Keywords: Hydranencephaly, Congenital Brain Abnormality, Palliative Care, Bioethics, Male, Equatorial Guinea

Introduction
Hydranencephaly is a rare but severe congenital structural brain abnormality, mostly isolated, consisting of the complete or near-complete absence of the cerebral hemispheres with the replacement of the cerebral parenchyma by cerebrospinal fluid. The cranial cavity may have remnants of glial tissue and ependyma, especially along the falx and close to the diencephalon. The cranial vault and meninges are intact. The incidence rate is around 0.01%-0.02% of pregnancies, and 1.4-2.8 per 100,000 live births. There is no difference between males and females [1, 2].

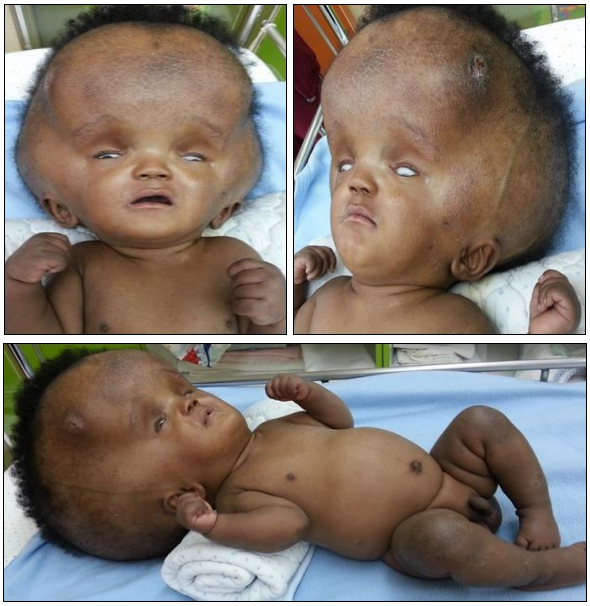

Fig 1: Phenotype of an 11-month-old male affected by hydranencephaly

Case Report
Male born in Equatorial Guinea with a history of 28-week twin gestation, perinatal asphyxia, weight 970 g., and head circumference of 27 cm. Shortly after birth, he presented signs and symptoms of hyaline membrane disease and seizures. In the neonatal period, he showed a significant increase in head circumference. Using imaging techniques, he was diagnosed with hydranencephaly. The decision was made to provide palliative care, ruling out surgical intervention due to poor prognosis. His

brother was healthy. In the following months, there was a progressive increase in head circumference, so, at 10 months of age, the mother requested an evaluation at a hospital in Spain to reconsider the therapeutic approach. Upon admission, he exhibited macrocephaly of 91 cm with occipital, parietal, and frontal ulcers, a setting-sun eye phenomenon, and disconnection from the environment. Brain ultrasound revealed at the supratentorial level the total replacement of the brain parenchyma by anechogenic fluid within which fine hyperechoic septa were identified; the posterior fossa was without apparent alterations. Head computed tomographic scan revealed the absence of supratentorial brain parenchyma that is replaced with cerebrospinal fluid; hyperdense lamellar material at the bilateral frontobasal level that may correspond to residual brain tissue and/or hypertrophic meninges; falx cerebri was present. Electroencephalogram showed globally slowed background activity with brief periods of suppression, with nonspecific irritative activity in both anterior temporal regions independently. The ophthalmoscopic examination was normal. Given the severity of the clinical-radiological findings, a multidisciplinary agreement was reached not to indicate surgical treatment for incurable underlying disease, the intervention being more life-threatening than conservative treatment. Palliative care was indicated. At 11 months of age, he was admitted to a Child Protection Center due to a social risk situation and the mother's inability to provide the palliative care he needed. He exhibited macrocephaly of 93 cm with occipital and frontal ulcers, very large fontanelle, a setting-sun eye phenomenon, and disconnection from the environment (Fig 1). During his stay, supportive care was applied (nutritional intervention, physical therapy, skincare, etc.), and no relevant pathological events occurred. At 17 months of age (head circumference 102 cm), he presented with respiratory failure that did not improve despite outpatient treatment, so he was urgently admitted to the hospital. Given the personal history and poor prognosis, in agreement with the family, it was decided to institute conservative treatment, comfort, and palliative care. After 48 hours, he presented an episode of generalized hypertonia with desaturation and bradycardia that resolved without the need for medication, and a second episode similar to the previous one with cardiorespiratory arrest and death.

Discussion

Regarding the etiology of hydranencephaly, the most commonly proposed mechanism is the ischemic stroke (infarction) involving the anterior circulation (bilateral occlusion of the internal carotid artery and, in some cases, the middle cerebral artery) usually between the 8th and 12th weeks of gestation. Damage to the brain usually is appreciated by diagnostic studies as early as the 13th to the 26th week of pregnancy (2nd trimester) when hemispheres and falx have been formed. The structures supplied by the posterior circulation (brain stem, cerebellum, thalamus, basal ganglia, and choroid plexus) are usually not involved [2].

Other different causes have been postulated: intrauterine infection (toxoplasmosis and viral infections), fetal hypoxia, genetic mutations (COL4A1 gene, LAMB1 gene, and the PI3K-Akt3-mTOR pathway), maternal exposure to toxins (cocaine, smoking, estrogens, sodium valproate, carbon monoxide, toluene), an extreme form of leukomalacia, the death of a co-twin in utero, and a rare genetic syndrome (Fowler syndrome) [2-5].

Most patients with hydranencephaly die before birth. Those who are born alive may appear completely normal at birth. Some patients present a degree of deformity in the skull and upper face if the intracranial pressure inside the skull is increased. Those with more severe disease may present at birth with evident symptoms such as seizures, myoclonus, and respiratory difficulty. Patients who appeared normal after birth, a few weeks later, start developing signs such as hyperirritability, hyper/hypotonia, seizures, increase in head circumference, and wide-open anterior fontanelles. These signs are more pronounced after several weeks. Months later, patients can present with macrocrania due to increased cerebrospinal fluid pressure or less commonly with microcephaly. Most of the children have serious cortical visual impairment but with normal eyes. Hearing is usually preserved, but occasionally sensorineural hearing impairment is noted [1, 2, 6, 7].

The majority of patients are diagnosed during pregnancy through a level 2 or higher ultrasound. If the diagnosis is not made during the pregnancy, postnatal diagnosis may be delayed from weeks to months, since patients may initially appear and function normally. Head computed tomographic scan exhibit very similar findings as described in brain MRI, although brain MRI is more accurate and precise [2].

The differential diagnosis must be done with other conditions such as severe congenital hydrocephalus, holoprosencephaly (especially the alobar type), severe open schizencephaly, and anencephaly. The differential diagnosis between hydranencephaly and severe congenital hydrocephalus is of notable importance in terms of prognosis and response to treatment. Infants with hydranencephaly diagnosed after 1 month of age, despite the placement of a ventriculoperitoneal shunt system, do not show neurological or radiological improvement. However, infants with severe congenital hydrocephalus treated with a ventriculoperitoneal shunt system usually show significant clinical improvement [1, 2, 6].

There is no cure for hydranencephaly. The prognosis is poor. Patients affected usually die in utero. Those who survive usually die within the first year of life due to complications. However, patients with survival of 20 or more years have been reported in the literature. The survival is related to the integrity of the brainstem, which regulates vital aspects, such as cardiorespiratory function, blood pressure, and temperature. For surviving patients, management of associated symptoms and/or complications and supportive care are mandatory. There is an ethical debate as to whether or not surgical treatment should be performed in cases complicated with hydrocephalus, considering the severe brain deterioration. However, it is known that the few patients described in the literature that survived past the 1st year of life did not improve after placement of the ventriculoperitoneal shunt system [1, 2, 6, 8, 9].

Hydranencephaly has significant ethical implications, both in terms of prenatal diagnosis and the treatment and care of affected individuals. Cases complicated by hydrocephalus present additional ethical challenges. Among the most important ethical considerations, the following stand out:

Prenatal diagnosis. When hydranencephaly is detected during pregnancy, parents face ethically complex decisions. They have to decide whether to continue with the pregnancy or terminate it. Here, religious beliefs, personal convictions,

and medical considerations come into play.

Informed consent. Doctors should ensure that parents are fully informed about the condition and prognosis of a baby with hydranencephaly before making treatment decisions. Informed consent is an essential part of medical ethics, allowing parents to make shared decisions about their child's care and treatment based on accurate information. It is essential to provide emotional support to parents and families during the decision-making process.

Dignity and respect. People with hydranencephaly and their families may face stigma and discrimination. Society often has deep-rooted attitudes and beliefs about disability, raising ethical questions about equality, respect, and inclusion. Medical decisions must be made with the well-being and respect of the patient in mind, even if their ability to communicate or make decisions is severely limited.

Medical decision-making. When a baby is diagnosed with hydranencephaly, parents and doctors face difficult ethical decisions about medical care and treatment. This includes the choice of providing palliative care, which focuses on alleviating the child's suffering or undertaking aggressive medical interventions that may not have a significant impact on the patient's quality of life. In some cases, medical procedures such as the placement of a ventriculoperitoneal shunt system may be considered to drain excess cerebrospinal fluid and reduce intracranial pressure. The ethics of this treatment involves weighing the possible benefits against the risks and the patient's quality of life, as well as informed parental consent.

Resources and fair distribution. In situations where medical resources are limited, such as in public health systems, the ethical question arises about how these resources should be allocated. Is it ethical to spend large sums on intensive treatments for people with hydranencephaly, especially if the results are uncertain in terms of improving quality of life?

Palliative care. In most cases of hydranencephaly, palliative care is considered the most appropriate ethical option as it focuses on alleviating suffering and improving the patient's quality of life. Palliative care can also help parents cope with the difficult situation in an ethical and compassionate way.

Quality of life. Doctors and parents must consider whether aggressive treatments will improve the child's quality of life or only prolong her suffering. This raises ethical questions about whether to prolong life with invasive measures or when it is appropriate to withdraw life support and allow a natural death.

Conclusion

Patients with hydranencephaly have a very poor life expectancy. This condition poses significant ethical challenges in the field of medicine and decision-making related to the medical care and treatment of affected patients. Medical ethics in these cases focuses on the balance between providing compassionate care that respects the patient's dignity and avoiding inappropriate medical interventions that would only prolong suffering without significant benefit.

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