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Hydranencephaly: Anatomoclinical Descriptions of a Rare Congenital Anomaly – a Case Report

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ABSTRACT

Introduction: hydranencephaly is a rare congenital anomaly in which the cerebral hemispheres are completely or partially absent, with the basal ganglia, brainstem, and meninges preserved.

Objective: to report a case of hydranencephaly in a newborn, describing the anatomoclinical findings and the importance of using computed tomography as a complementary examination for correct diagnosis.

Case report: a 33-year-old female patient was admitted to the hospital and referred for an emergency cesarean section secondary to infectious chorioamnionitis due to premature rupture of the amniotic membranes. Obstetric ultrasound revealed findings suggestive of malformation of the upper regions of the central nervous system, such as a single lateral ventricle in its anterior horn without visualization of the anterior portion of the midbrain line, suggesting holoprosencephaly. After birth, macrocrania, enlargement of the nuchal region, open and tension-free fontanelles, among other findings, were noted. The baby was referred for a cranial computed tomography scan for differential diagnosis, where diffuse morphological alteration of the brain was evidenced, without characterization of the parenchyma and cerebral cortex, ventricles or corpus callosum. The cerebellum, brainstem and posterior part of the thalami were present. The appearance was highly suggestive of hydranencephaly.

Conclusion: this paper reports a case of a rare congenital anomaly known as hydranencephaly, highlighting its anatomical and clinical aspects and showing the importance of using postnatal computed tomography as an important exam for differential diagnosis.

Keywords: Hydranencephaly; Congenital malformation; Diagnosis, Anatomy.

Introduction

Hydroencephaly is a term that etymologically originates from the Greek, where "hydro" means "water" and "encephaly" means "brain", where the first description whose existence was recorded as "anencéphalie hydrocéphalique" occurred between the years 1830-56 by Cruveilhier[1,2,3]. The term "hydranencephaly" was probably introduced by Spielmeyer in 1905, being improved in 1958 by Crome and Sylvester[4,5,6,7]. Hydranencephaly is a rare congenital disorder, with a prevalence rate of 0.2 to 0.4 cases per 10,000 live births[8] and which manifests itself in the period of embryogenesis after the process of neurulation of the neural tube, occurring around the second trimester of pregnancy[8-10].

In this sense, it is a neurological condition characterized by the total or partial absence of the cerebral cortex and which represents an extreme in the variability of congenital brain malformations[11]. It has been described as a multifactorial result involving the interruption of brain development during the first trimester of pregnancy and which has been associated with iatrogenic, infectious, toxic, genetic or hypoxic causes[8,12]. However, it is essential to emphasize that the pathophysiology associated with hydranencephaly has not yet been clearly defined[13]. Regardless of the genesis, the outcome is the formation of a space filled with cerebrospinal fluid, residual neurons, glial cells[14], and remaining blood vessels in the place where the cortex anatomically belongs, linked to severe complications given the functional importance of the telencephalon[8,15-17].

From this perspective, most cases of hydranencephaly can be diagnosed in the second half of pregnancy[9] but are limited in terms of correct diagnosis[1,8]. The inability to identify the condition prevents the efficient delimitation of the extent of associated damage, which in most cases presents as low survival rates and a potentially poor prognosis[8,18,19].

This study aimed to provide an anatomoclinical description of a case report of prenatal diagnosis of a newborn with findings suggestive of brain malformations on obstetric ultrasound examination followed by postnatal diagnostic confirmation

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through computed tomography confirming absence of parenchyma cerebral cortex, lateral ventricles and corpus callosum.

This study has been approved by the local Institutional Ethics Committee (CAAE: 79994724.8.0000.5060 / Decision number: 6.885.114)

Case Report

A 33-year-old female patient, with 32 weeks and 2 days of gestation, was admitted to the hospital and referred for an emergency cesarean section, secondary to infectious chorioamnionitis due to premature rupture of ovular membranes. Obstetric history: G3/P2/A0 (cesarean sections); Incomplete prenatal card; Denies alcoholism and smoking; Vaccination card: DTaP, with no reports of other vaccines; Positive GBS test; Negative serologies, serological panel of immunity for toxoplasmosis. Obstetric ultrasound revealed findings suggestive of malformation of the central part of the nervous system, such as a single lateral ventricle in its anterior portion without visualization of the anterior portion of the midbrain line, with no other associated morphological abnormalities. Such findings suggested the diagnosis of holoprosencephaly.

After birth and during hospitalization in the Neonatal Intensive Care Unit, serial physical examinations revealed macrocranial enlargement, widening of the nuchal region, open and tension-free fontanelles, diastasis of all cranial sutures, trivascular umbilical stump, global hypertonia with predominance in the upper limbs, and a tendency toward opisthotonus. In the immediate postnatal period, on the first day of birth, the baby underwent transfontanellar ultrasound, confirming doubts about the initial diagnosis. The definitive imaging diagnosis was only made on the second day of birth, by computed tomography of the skull. The examination demonstrated diffuse morphological alteration of the brain, without characterization of the parenchyma and cerebral cortex, ventricles, or corpus callosum. The cerebellum, brainstem, and posterior part of the thalami were present. This aspect was highly suggestive of hydranencephaly.

The baby was discharged from hospital at 1 month and 10 days of age, with a calculated gestational age of 39.1 weeks, weight of 3,220 grams, head circumference of 40 cm, and body length of 48.5 cm. Multidisciplinary monitoring was planned with a pediatrician, neurosurgeon and team from the institution "Associação de Pais e Amigos dos Excepcionais" – APAE (Association of Parents and Friends of the Special Needs Children).

Discussion

Fetal anomalies of the central nervous system are quite common, with an incidence of 0.1% to 0.2% of live births and an occurrence of 3% to 6% in stillbirths, presenting itself as the second leading cause of fetal death[20]. Hydranencephaly is a malformation that corresponds to this classification and is characterized by the absence of the telencephalic hemispheres. Given the above, it has a prevalence of 5% among ultrasound diagnoses, where, according to findings by Barros *et al.*, the highest prevalence of neurological disorders is in cases of hydrocephalus.

Hydranencephaly is a significant clinical challenge given the complexity of establishing correct diagnoses and delimiting causes analogous to the condition[12]. Diagnoses associated with hydranencephaly are based on radiological techniques such as transillumination, computed tomography, brain magnetic resonance imaging and cerebral angiography, in addition to neurological clinical signs that present low cognitive development or motor impairment[11]. Early diagnosis is performed through ultrasound, such as Doppler and volumetric ultrasound, which screen for fetal malformations and assist in treatment planning and delimiting the patient's prognosis[22]. The clinical similarity of this condition to other neurological malformations, such as holoprosencephaly, porencephaly and anencephaly, directly affects the prognosis regarding the chances of survival and associated impairments[23]. This initial confusion between diagnoses occurred in our case, where, in a first examination, still in the obstetric ultrasound, the findings suggested it was a case of holoprosencephaly (figure 1). However, on the first day after birth, a transfontanellar ultrasound was performed, generating suspicion about the possibility of it being hydranencephaly (figure 2). On the second day of life, a computed tomography exam was performed, which demonstrated that it was indeed hydranecephaly (figure 3). We also emphasize that the limited availability of specialized imaging exams, together with the lack of knowledge about the condition, contribute to inconsistent diagnoses.

Therefore, the malformation in question presents a morphology linked to the absence or atrophy of the cerebral cortex, which is replaced by a sac-shaped membrane filled with cerebrospinal fluid[23,24]. In imaging exams, the condition results in an empty or virtually absent appearance of the cerebral hemispheres. Within this membrane formed by the pia mater and arachnoid mater leptomeninges, there will be cerebrospinal fluid, necrotic dendrites and residual neurons, with or without partial preservation of the occipital lobe[8,9,14,15]. Structures such as the brainstem, diencephalon (thalamus), basal ganglia, choroid plexus and cerebellum are generally preserved[9,25]. Hydranencephaly can present with a wide range of aspects of destruction, corresponding to the bilateral absence of the telencephalic hemispheres or absence of one of the hemispheres, the left or the right, and finally, the cortical absence and structures of the diencephalon such as the thalami and basal ganglia[9,23-27].

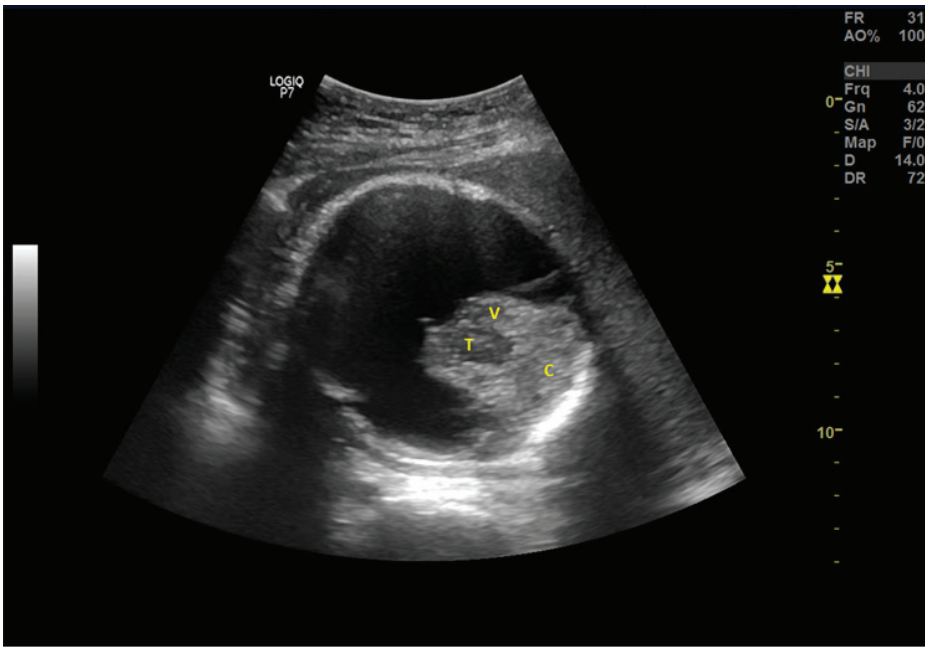

Figure 1. Obstetric ultrasound at the gestational age of 32 weeks and 2 days.
Figure caption: note a single cerebral ventricle (V), communicating anteriorly, with the anterior portion of the cerebral midline not being visible, suggesting holoprosencephaly. Thalamus (T), Cerebellum (C).

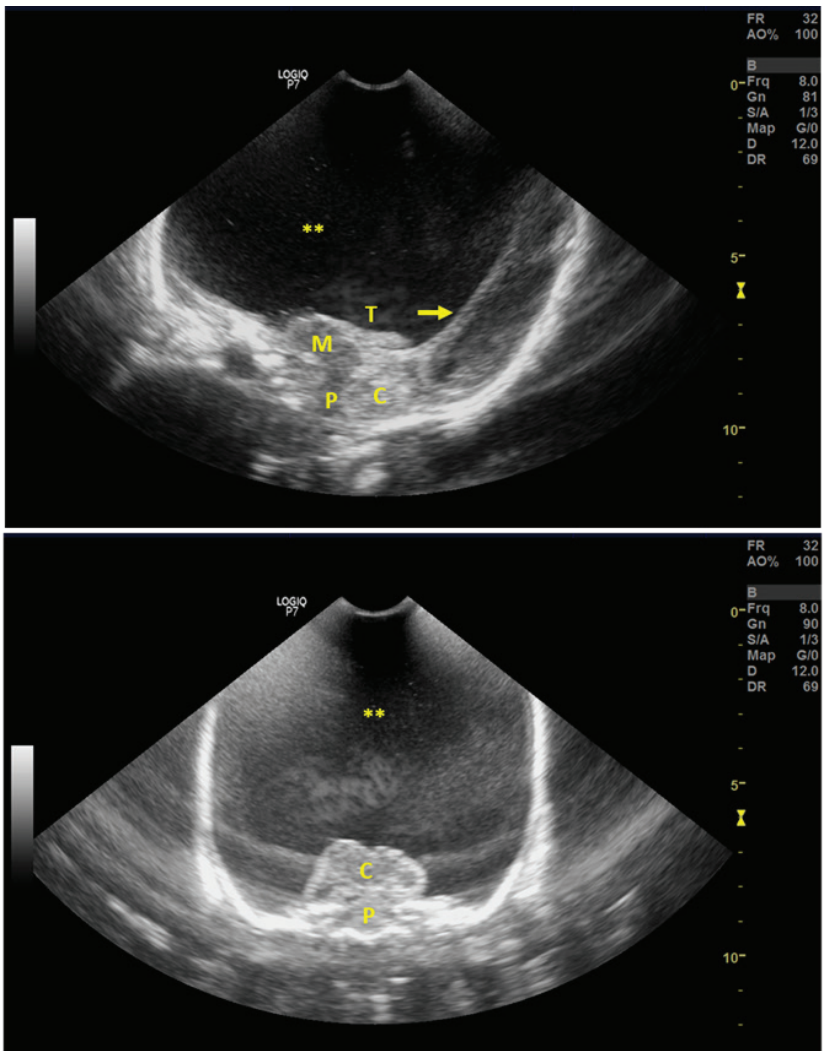

Figure 2. Transfontanellar ultrasound performed on the day of birth.
Figure caption: note the cranial cavity with no brain tissue and filled with cerebrospinal fluid (**), cerebellar tentorium (arrow), thalamus (T), cerebellum (C), midbrain (M), and the pons (P).

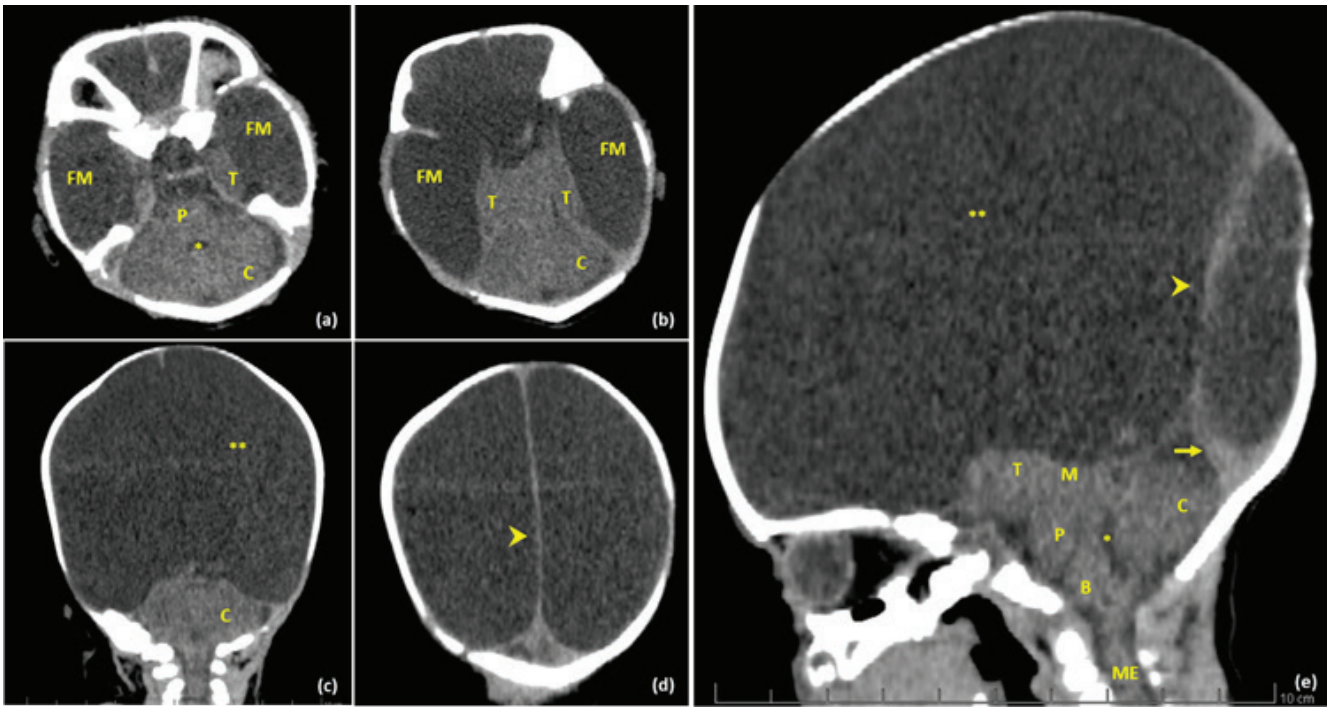

Figure 3. Computed tomography performed on the second day after birth.
Figure caption: serial computed tomography scans in transverse/axial view (a,b,d), frontal/coronal view (c) and lateral/sagittal view (e). The images show the middle cranial fossa with the absence of nervous tissue (FM), the thalamus (T), the midbrain (M), the pons (P), the medulla (B), the cerebellum (C), the spinal cord (ME), remnants of the brain falx (arrowhead), the cerebellar tentorium (arrow), the fourth ventricle (*) and the cranial cavity with bilateral absence of the telencephalon filled with cerebrospinal fluid (**).

Thus, delimiting morphological aspects is essential for correct diagnoses and projections of neurological damage from the condition of hydranencephaly.

In the case report in question, the absence of structures was diagnosed by computed tomography of the skull, confirming the absence of brain parenchyma in the cerebral hemispheres, absence of lateral ventricles and corpus callosum with increased head circumference. Among the structures that were preserved, the cerebellum, brainstem and posterior part of the thalami are observed, as demonstrated in figure 3.

When destruction associated with the cortex and brain tissue is noted due to failure in their development, intracranial pressure is altered, leading to abnormal development of the skull and associated structures[28]. Although important structures of the telencephalon do not coexist in these cases with modification of the skull perimeter and morphological rearrangement, there is compatibility with life given the preservation of fundamental centers present in the respiratory and vasomotor centers respectively, located in the reticular formation of the bulb in the brainstem. However, changes will be observed concerning motor and sensory impairments that affect functional capacities in projection and association, and in the long term, they corroborate by drastically reducing the life expectancy of patients[11].

Hydranencephaly is a serious condition that can significantly reduce the life expectancy of patients, in addition to leading to numerous complications. It is a malformation for which there is no cure and its treatment involves reducing symptoms and providing support to the patient[8,23]. The quality of life of surviving patients is seriously compromised due to the presence of numerous neurological impairments and clinical signs that correspond to the compromised structures, which may include developmental delays, seizures (including those that are resistant to medication), spastic diplegia, cerebral palsy, respiratory distress, facial paralysis, hemiparesis, generalized hypertonia, strabismus, blindness, abducens nerve palsy, nystagmus, among others[24,29-31].

Most patients die in their first year of life[8] but there are reports of patients with longer survival, such as that of Dias *et al.*[32] who survived until 21 years, associated with less structural impairment due to the condition of hemihydranencephaly. This data is also supported by what was reported by McAbee *et al.*[23], who demonstrated a case of a patient who survived until 19 years without improvement in neurological outcomes and the findings of Ulmer *et al.*[33], who described the survival of a 36-year-old patient with left hemihydranencephaly. According to a review by McAbee *et al.*[23], the survival factor of patients would be directly associated with the preservation of subcortical regions such as the brainstem, cerebellum and diencephalon. Some studies, such as that of Hassenein *et al.*[24], report the possibility of existing cortical areas taking over and executing the non-existent functions of the destroyed hemisphere, which would explain greater chances of survival in some patients, but when

there is a total absence, such as the case portrayed in this article in question, this possibility would not be a reality.

Conclusion

This study demonstrates a rare case of a congenital anomaly known as bilateral hydranencephaly, addressing the diagnostic trajectory through imaging exams, correlating the clinical and anatomical aspects of the disease. We emphasize the importance of computed tomography exams of the skull for the definitive examination associated with the history of previous imaging exams and physical examination of the patient. Knowledge of the anatomical and clinical aspects of the disease and understanding the challenges to make the appropriate diagnosis accurately and more quickly is of great importance for the health professionals involved to be attentive and able to provide patient care with greater efficiency and accuracy.

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Mini Curriculum and Author's Contribution

1. Fernanda Dazilio dos Reis: M.D. student. Contribution: case study, manuscript preparation and data collection.

2. Caio Pedrosa Beber: M.D. Contribution: case study, manuscript preparation and data collection.

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