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Case Report

Hydranencephaly first case in our hospital

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Abstract

Several types of encephalo-dysplasias have been described associated with cystic formations. Thurnbull in 1904 described a case under the term of cystic aplasia where both parietal lobes were involved. In 1950 Hamby, Krauss and Beswick reported 7 children with dysplasia using the term hydranencephaly as it is now known, in such patients the cerebral hemispheres were completely or almost completely absent and their place was occupied by cerebrospinal fluid. Male term newborn patient obtained by cesarean section with Apgar, Silverman and normal somatometry of a 23-year-old HIV-bearing mother without prenatal control who was identified at birth as obstetric ultrasound as hydranencephaly vs severe obstructive ventriculomegaly. Ultrasound and tomography check the diagnosis. The patient dies 7 days after his birth.

Keywords: Hydranencephaly; Carotid arteries, Prenatal diagnosis; Postnatal diagnosis

Introduction

Several types of encephalo-dysplasias have been described associated with cystic formations. Thurnbull in 1904 described a case under the term of cystic aplasia where both parietal lobes were involved. In 1950 Hamby, Krauss and Beswick reported 7 children with dysplasia using the term hydranencephaly as it is now known, in such patients the cerebral hemispheres were completely or almost completely absent and their place was occupied by cerebrospinal fluid.

This condition was also described in 1940 by Bettinger and for the first time by Cruveilhier in 1835. The oldest case corresponds to Ambroise Paré.[1,2,3]

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Case Report

Male term newborn patient obtained by cesarean section with Apgar, Silverman and normal somatometry of a 23-year-old HIV-infected mother without prenatal control who was identified by obstetric ultrasound with hydranencephaly vs severe obstructive ventriculomegaly. New ultrasound reported: supratentorial region with cyst occupying the upper space. No middle line or brain mass is identified, only incomplete internal septum and choroid plexuses, neither thalamus nor basal ganglia are visible. Structures of the posterior base are observed and there is no flow in color Doppler or in dependent Doppler (Figures 1 and 2). He was asked for a tomography that reports skull bones and open fontanelles. A cystic cavity that occupies the entire brain space, a pontine fossa with a structure similar to the left cerebellar reminiscence and brainstem (Figures 3, 4). The patient dies 7 days after birth in the neonatal intensive care unit.

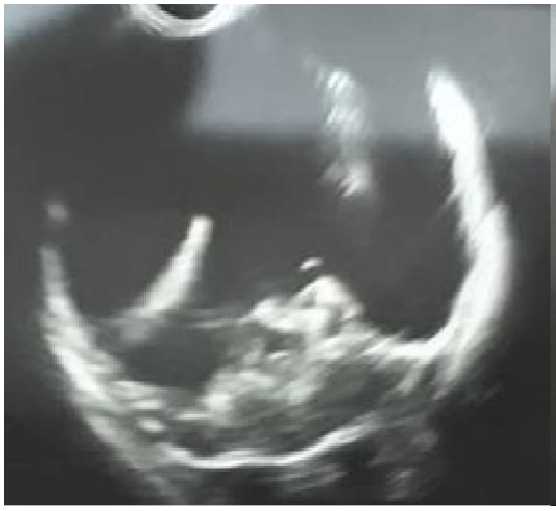

Figure 1.

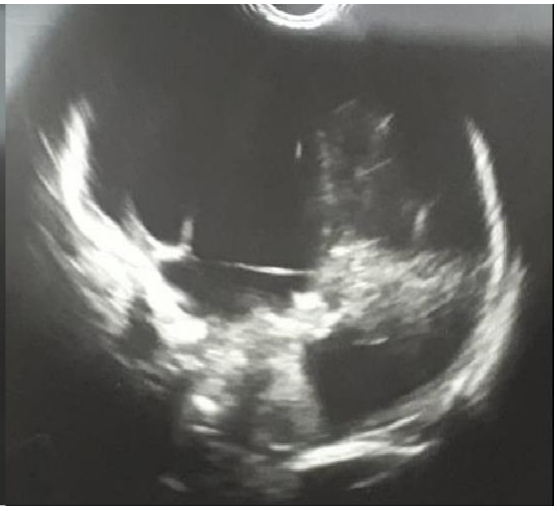

Figure 2.

Transfontanelar ultrasound images showing a cystic image that covers almost the entire supratentorial cerebral parenchyma.

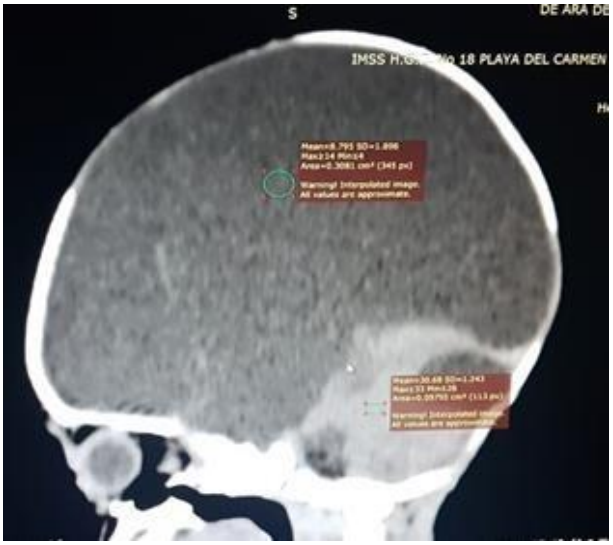

Figure 3.

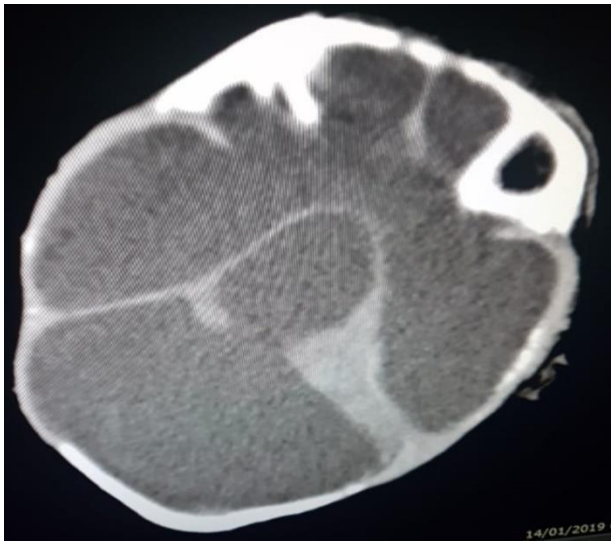

Figure 4.

Sagittal (F.3) and coronal (F.4) tomography showing scarce cerebral parenchyma with predominance in the posterior region and cystic image occupying the supratentorial region that displaces the superior structures.

Discussion

The etiology of hydranencephaly is controversial but since the meninges and the cranium are intact it is presumed that from the neopallium rudiments the defect is already present during the period of embryological formation of these structures.[4]

On the other hand, hydranencephaly is a rare malformation, with an approximate incidence of 1 in 10,000 live births; it is characterized by the absence of cerebral hemispheres, which are replaced by a membranous sac containing cephalospinal fluid. The origin of this pathology is unknown. The most probable explanation for the disappearance of the fetal cerebral hemispheres is the occlusion of the supraclinoid portion of both carotid arteries (hypoplastic arteries or bilateral aplasia have been found in postnatal studies with the consequent hemorrhagic destruction of the cerebral parenchyma.[5]

The variable detection of brain remains seems to reflect the development path of the cerebral arteries. In addition, data from fetal and postnatal neuroimaging and histopathological findings point to an early occlusion of the internal carotid arteries, which occurs mainly between the eighth and twelfth weeks of gestation, as the main pathogenic mechanism.[6] In such a way that it is a challenge for the imageologists to detect this anomaly in the early stages of fetal development.

On the other hand, its etiology has been attributed to multiple causes that lead to ischemic injury and viral infections are the main ones involved, such as herpes virus, parvovirus, cytomegalovirus, among others; another possible etiology is toxic (alcohol) or genetic processes such as Fowler's syndrome.[7,8] Hydranencephaly does not occur after a hydrocephalus, while it can start as a multicystic encephalopathy, and progress to hydranencephaly, since there is also a severe vascular base lesion.[10]

Cases of hydranencephaly associated with congenital toxoplasmosis have also been reported as another etiological possibility of this condition, although in these cases it is most likely hydrocephalus10 as well as associated with systemic lupus erythematosus.[11]

Genetics has also focused on unraveling this mystery in such a way that Abdel-Hamid MS, et al.,[12] and led them to consider neurodevelopment protein 1 gene (NDE1) and identify a new variant. Non-sensitive homozygous (c.54G> A, p. W18) as a possible cause of the disease. The variability of the degree of brain malformations and the apparent fusion of the thalami were elusive and delayed the recognition of the genetic etiology. Their results provide the first prenatal description of this rare syndrome.

Regarding diagnostic imaging, Chih-Ping Chen, et al. report a case with type II thanatophoric dysplasia detected by prenatal ultrasound observing nuchal translucency at 14 weeks of gestation and a new study at 25 weeks revealed hydranencephaly associated with other defects.[13] Kline-Fath BM, et al. reports that fetal ventriculomegaly is a common reference for prenatal MRI with possible etiologies such as hydrocephalus and hydranencephaly.[14]

Gardea-Loera G, et al., discuss the clinical aspects of neuroimaging and electrophysiological behavior of hydranencephaly where there is great similarity between hydranencephaly and severe congenital hydrocephalus, both clinically and in imaging studies.[15] In hydranencephaly, the electroencephalogram (EEG) usually shows no electrical activity and auditory brainstem evoked potentials are normal. In our hospital it is the first case of this rare anomaly.

Author contributions

RBC, GPA took care of the patient and made the literature research. RBC, GPA drafted the manuscript and supervised the manuscript. The final version has been read and approved by all authors.

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Conflict of interest

All authors declare that they have no conflict of interest.

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