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Case Report

Colpocephaly, a Very Rare Neonatal Brain Malformation: a Case Report and Literature Review

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Abstract:

Colpocephaly is a very rare brain malformation, in which the occipital horns are larger than the anterior horns of the lateral ventricles. We report the case of a newborn with colpocephaly associated with agenesis of the corpus callosum and a spina bifida.

Newborn male, from an unrelated marriage, of a 21-year-old mother, with no particular pathological history. The newborn was admitted on the first day of life of life for an early neonatal infection with pulmonary localization associated with a polymalformative syndrome made up of a facial dysmorphism, a spina bifida and a limb anomalies. The transfontanellar ultrasound showed triventricular hydrocephalus and brain CT scan showed colpocephaly associated with an agenesis of the corpus callosum. The etiological investigation did not show any drug intake or toxic substances during pregnancy. Serologies for toxoplasmosis, rubella, HIV, syphilis, hepatitis B and CMV were negative. The newborn died before a genetic study and brain MRI could'not be performed.

Antenatal and postnatal diagnosis of colpocephaly is difficult because of its rarity and its radiological appearance close to hydrocephalus, hence the need for appropriate postnatal imaging andwell-codified multidisciplinary support.

Key words: absent corpus callosum, Colpocephaly, fetal MRI, prenatal ultrasound

I. Introduction

Colpocephaly is a very rare congenital brain malformation (less than 70 cases have been published since its first description in 1940), in which the occipital horns are larger than the anterior horns of the lateral ventricles [1,2]. It is a persistence of the fetal configuration of the lateral ventricles [3]. It is assumed to be a consequence of abnormal neuronal proliferation or migration during central nervous system (CNS) embryogenesis [4]. Multiple causes are linked to this abnormal CNS development, such as intrauterine or perinatal lesions, genetic aberrations, and morphogenesis abnormalities. Colpocephaly is often associated with other neurological malformations, including partial or complete agenesis of the corpus callosum [5]. Its radiological diagnosis is usually made in the perinatal period and later suggests intellectual disability. Adult cases of newly diagnosed colpocephaly have been reported only rarely. [6], as it may be initially misdiagnosed as hydrocephalus [7,8]. We report the case of a newborn with colpocephaly associated with agenesis of the corpus callosum and spina bifida.

II. Case report :

This is a newborn male, from an unrelated marriage, of a 21-year-old mother, with no particular pathological history, the pregnancy was singleton well monitored, carried out at 36 weeks of amenorrhea plus 4 days according to the first trimester ultrasound, the delivery was vaginal and the Apgar score increased from 6/10 to 8/10 from the first to the fifth minute. The patient was hospitalized, on the first day of life, for an early neonatal infection with pulmonary localization and a weak sucking reflex associated with a polymalformative syndrome made up of facial dysmorphism, spina bifida, anal prolapse and limb anomalies. The patient weighed, at birth, 1680g (-2DS) with a height of 34 cm (-2DS) and a head circumference of 29 cm (<2DS). During his hospitalization in neonatal

intensive care, two transfontanellar ultrasounds were performed atfifth and fifteenth dayhaving objectified triventricular hydrocephalus with periventricular leukomalacia and the brain CT scan on the 28th postnatal day showed colpocephaly associated with agenesis of the corpus callosum with right parietal leukomalacia. The rest of the malformation assessment revealed aatrioventricular communicationand pulmonary stenosis requiring medical care without other associated anomalies. Concerning the etiological research we did not objectify any medication or toxic substances taken during pregnancy. Serologies for toxoplasmosis, rubella, HIV, syphilis, hepatitis B and CMV were negative. The newborn died following sepsis with pulmonary origin, before a genetic study and a brain MRI were performed.

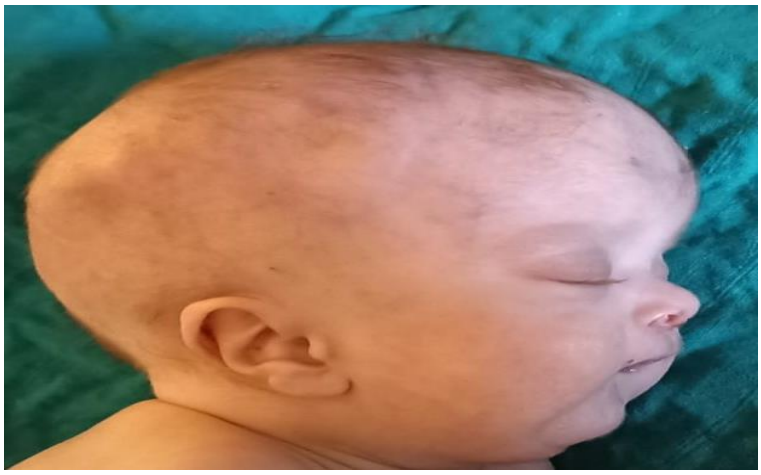

Figure 1: profile image of the face

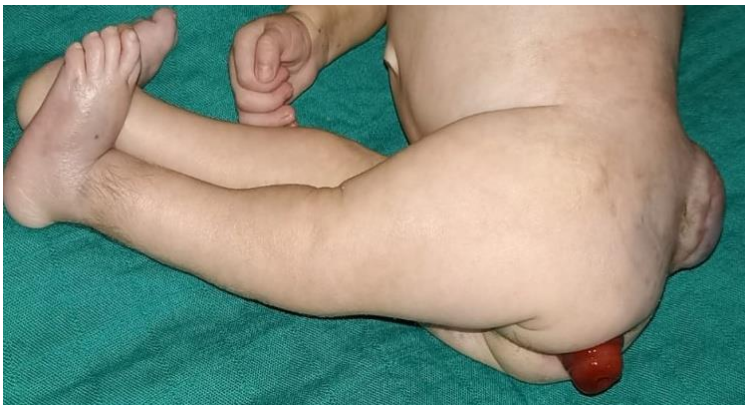

Figure 2: Abnormalities of the limbs and trunk

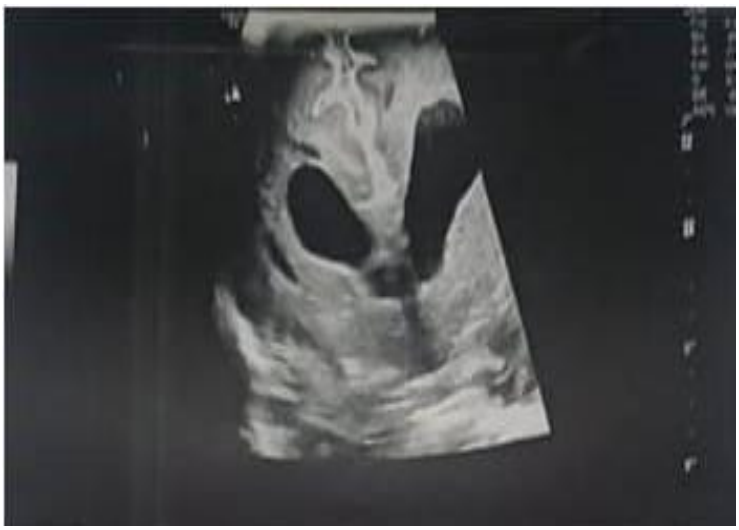

Figure 3: Trans-fontanellar ultrasound

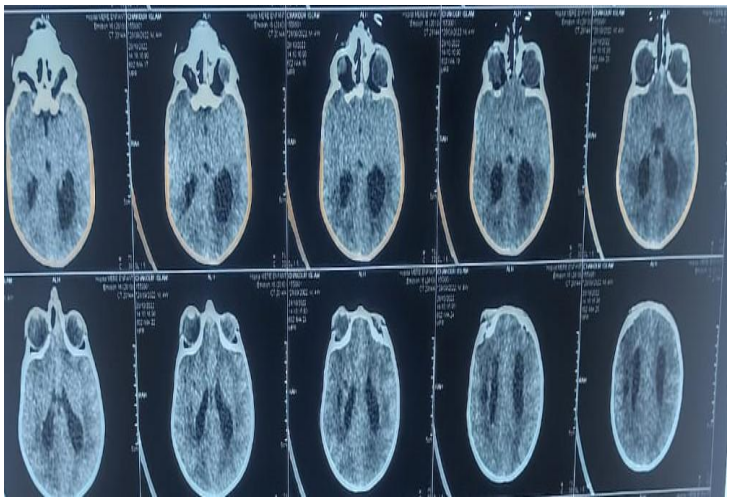

Figure 4: Brain scan

III Discussion :

Colpocephaly is an abnormal enlargement of the occipital horns of the lateral ventricles [4]. Since its first description in the scientific literature at least 70 cases have been reported worldwide [9,10]. It was initially described in 1940 by Benda, who classified this type of malformation as a developmental disorder of the cerebral vesicles [9]. In 1946, Lakovlev and Wadsworth introduced the term "colpocephaly" from the Greek term colpos meaning "hollow", to name the same brain abnormality [9]. This abnormality is secondary to a rare disorder of neuronal migration usually occurring during the first trimester of pregnancy [11]. Several studies suggested that the ventricular enlargement in colpocephaly was caused by an arrest of white matter development, occurring between the middle of the second month and the fifth month of fetal life [12]. An embryological mechanism has been proposed: the lateral ventricles appear as large cavities of the telencephalic vesicle. The normal developmental decrease in ventricular size occurs only after the formation of the foramen of Magendie, which decompresses the ventricular cavities. The occipital horns are further attenuated and shaped by the increase in size, volume, and myelination of ventricular wall fibers as well as by the association of corpus callosum and forceps fibers, the internal parieto-occipital fissure of the tapetum, and the calcarine fissure. It has been suggested that as yet unknown causes may interfere with this delicate developmental process and that the end result of such developmental arrest may be a disproportionate enlargement of the occipital horns, giving the radiological appearance of colpocephaly [13]. Several etiologies have been proposed as causes of colpocephaly, including perinatal hypoxic-ischemic encephalopathy [7], chromosomal abnormalities [3], such as mosaic trisomies [7], Familial occurrence of colpocephaly has been noted in three reports and an autosomal or X-linked recessive origin has also been suggested [3], congenital infections, such as toxoplasmosis [3] and cytomegalovirus and maternal use of drugs and toxic products during pregnancy, such as corticosteroids, salbutamol and theophylline [3]. All investigations performed in our case for etiology were unremarkable. Diagnosis of colpocephaly is uncommon during the antenatal period because it is often confused with hydrocephalus [14]. Clinically, it can manifest with varying degrees of intellectual disability, microcephaly, seizures, muscle spasms, and locomotor or visual abnormalities [15]. It should be noted that patients with colpocephaly will not always have neuropsychomotor abnormalities [3]. This pathology can be associated with serious malformations of the central nervous system, such as agenesis of the corpus callosum, lissencephaly, pachygyria, schizencephaly, macrogyria, hypertrophy of the cisterna magna, cerebellar atrophy, optic nerve hypoplasia, chorioretinal coloboma, microcephaly, myelomeningocele and hydrocephalus [15]. Among these possible concomitant congenital anomalies, agenesis of the corpus callosum remains the most frequently observed with a prevalence that can reach 40%. The estimated prevalence of agenesis of the corpus callosum in the general population is 3 to 7 per 1,000 births, compared to 2 to 3 per 100 births in children with developmental anomalies [6]. In the current literature, all adults with colpocephaly had concurrent partial or complete agenesis of the corpus callosum, except for 3 cases. Of these 3 cases, only 1 patient was diagnosed with colpocephaly with normal brain parenchyma [16]. The other 2 cases were associated with porencephaly [17] and an absent circle of Willis [18]. Magnetic resonance imaging (MRI) is the gold standard for its diagnosis [15]. Computed tomography may miss a small healthy portion of the corpus callosum [15]. Colpocephaly is usually a disease of the newborn and young infant. Review of the current literature suggests that colpocephaly is often an incidental diagnosis in adults [19]. Adult patients typically present with nonspecific symptoms such as headache, dizziness, seizures, vertigo, nausea, motor abnormalities, sensory abnormalities, cognitive abnormalities, intellectual disability, learning disabilities, loss of consciousness, and visual hallucinations [19,20]. Gungor et al. reported a case of postmortem diagnosis of colpocephaly in a patient with a lifelong history of poor academic performance [19]. Some cases had other primary diagnoses such as meningioma [19] and paraspinal abscess as a complication of spinal surgery [19]. Some patients were diagnosed with colpocephaly after being admitted for medical management of respiratory tract infection and hypertensive episode [19]. Comorbid psychiatric disorders have also been reported in some cases, such as schizophrenia [5] and obsessive-compulsive disorder [5]. These adult cases often require only symptomatic treatment [19].

IV. Conclusion :

Colpocephaly is a rare form of ventriculomegaly.Its antenatal and postnatal diagnosis of colpocephaly is difficult because of its rarity and it is often misdiagnosed as hydrocephalusat normal pressure [19],hence the need for appropriate postnatal imaging andearly, multidisciplinary and well-codified care includingits diagnostic, therapeutic and prognostic aspects. This will make appropriate professional conduct possible, thus ensuring an improvement in the quality of life of the patients concerned, both in the medium and long term [19].

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