



Submitted on: 05/14/2017  
Approved on: 09/05/2017

CASE REPORT

## Colpocephaly in newborn: case report and literature review

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### Keywords:

infant, newborn, neonatology, lateral ventricles, brain, agenesis of corpus callosum.

### Abstract

Colpocephaly is a rare cephalic disorder caused by an abnormal development of the central nervous system. It is characterized by a disproportionate enlargement of the occipital horns of the lateral ventricles and other potential brain malformations. Here we report the case of a preterm neonate who underwent cranial ultrasonography examinations that showed bilateral periventricular hyperechogenicity and colpocephaly. The information about the described case was obtained through medical record review, photographic documentation of the tests performed on the newborn, and a review of the current literature. This case report and the collected references seek to clarify the features of colpocephaly and provide an adequate understanding of its diagnostic, therapeutic, and prognostic aspects.

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## INTRODUCTION

Colpocephaly is a rare anatomic finding of the brain. It is characterized by a disproportionate enlargement of the occipital horns of the lateral ventricles but normal frontal horns.<sup>1,2</sup> It is assumed that this congenital malformation is a consequence of an abnormal neuronal proliferation or migration during the embryogenesis of the central nervous system (CNS).<sup>1,2</sup> Multiple causes are related to such an abnormal CNS development, such as intrauterine or perinatal injuries, genetic disorders, and errors in morphogenesis.

Other neurological malformations, particularly agenesis of the corpus callosum, may be associated with colpocephaly.<sup>3</sup> Clinically, in the early phase of life, the patient may have delayed neuropsychomotor development, muscular spasms, seizures, and visual and motor disorders.<sup>3</sup> On the other hand, there may be clinical manifestations only in the adult age, such as falls and headaches.<sup>4</sup>

The present article reports a single case of colpocephaly, which was diagnosed in a newborn who was admitted to a reference hospital in neonatology. Until March 2017, there were at least 60 reported cases, a small number to justify the findings of this report.<sup>1,3-12</sup>

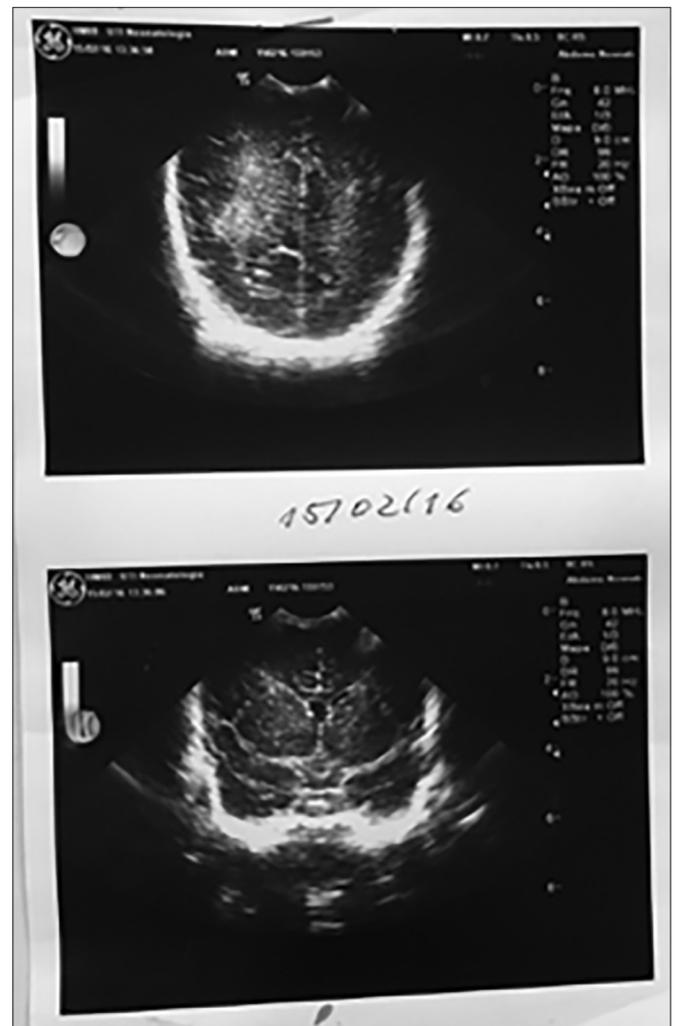
## CASE REPORT

The patient, initials G. F. N., was a male newborn, referred by a mother and child care center in the Brasília area, Brazil, with a clinical history of birth by cesarean section due to fetal distress (severe pre-eclampsia and fetal brain-sparing effect) at the gestational age of 30 weeks and 5 days. The amniotic sac was ruptured during the intervention. The patient weighed 1,125 g at birth, with a head circumference of 27 cm, a height of 38 cm, and Apgar scores of 6 and 8 at the first and fifth minutes, respectively. The mother had 7 prenatal consultations and negative serology for toxoplasmosis, HIV, syphilis, and hepatitis B in the first trimester of pregnancy.

It is also of note in the newborn's clinical history that he did not cry at the moment of birth; it was necessary to perform neonatal reanimation, with aspiration of the airways, positive-pressure ventilation, and orotracheal intubation. A surfactant was prescribed, and the patient was later transferred to the neonatal intensive care unit (NICU). Mechanical ventilation was well tolerated, with low parameters and no desaturation or complications during that period. The patient was extubated after 1 day. During his period at NICU, he had two episodes of temperature imbalance, and antibiotic therapy was started due to the impossibility of performing a lumbar puncture because of thrombocytopenia. After clinical stabilization, the newborn was transferred to the neonatal intermediate care unit of the Brasília University Hospital (HUB) to complete the treatment and for nutritional recovery.

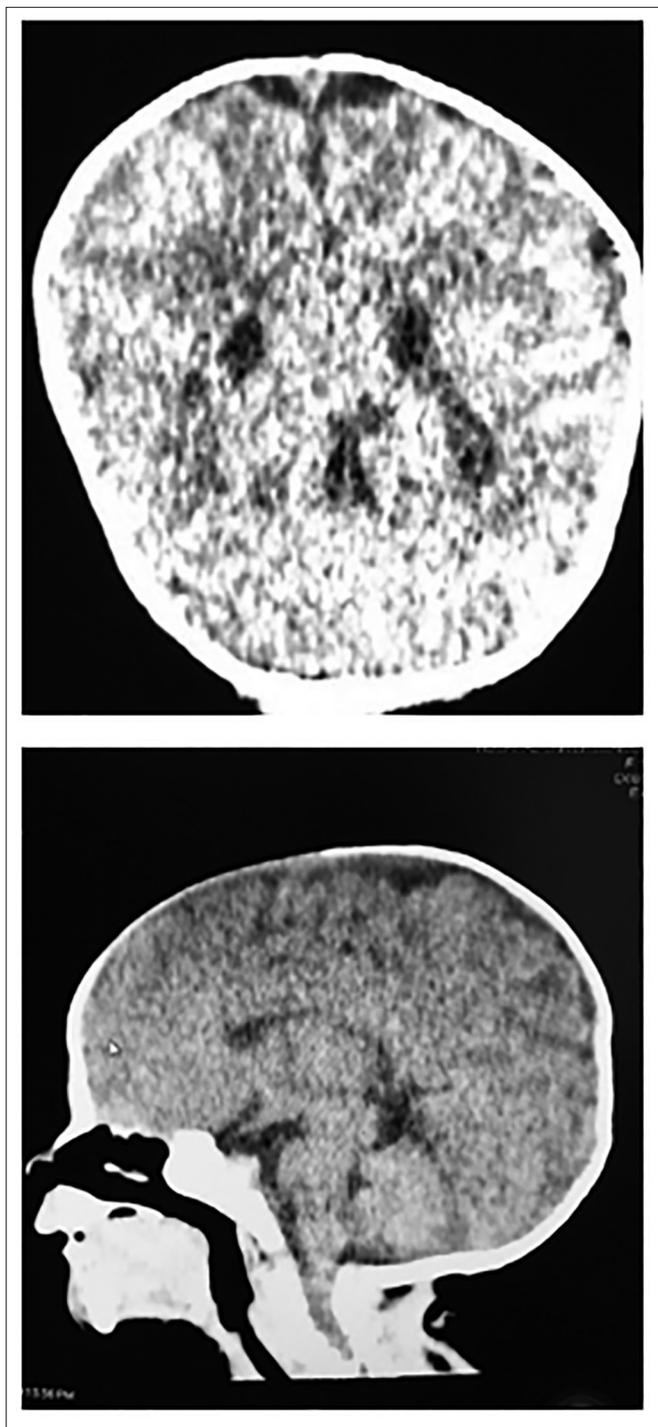
During that period, two transfontanelar ultrasound examinations were performed. The first one, on the eighth postnatal day, showed left periventricular hyperechogenicity (Figure 1), and the second one, on the 17th postnatal day,

showed bilateral periventricular hyperechogenicity and left-side colpocephaly. Later, on the 43rd postnatal day (or the 6th week after birth), a computed tomography scan of the head was performed, and it showed an asymmetry of the occipital horns, larger on the left side, with no other anatomical changes (Figure 2). A magnetic resonance scan would have better shown the existing anatomical changes and any other simultaneous malformations but could not be performed because the equipment was down for maintenance at the time the newborn was hospitalized.



**Figure 1.** Transfontanelar cranial ultrasound showing left periventricular hyperechogenicity.

The newborn had a satisfactory clinical evolution, including an adequate weight gain. Thus, he was discharged from the hospital with recommendation for follow-up at HUB's high-risk pediatrics outpatient clinic. An outpatient evaluation at 4 months and 26 days of chronological age (2 months and 20 days of corrected age) showed that the patient had a social smile and proper head postural control. It should be noted that his growth and development were adequate for his age during the first year of life.



**Figure 2.** Computed tomography of the head, showing an asymmetry of the occipital horns, larger on the left side.

## DISCUSSION

Colpocephaly is an abnormal enlargement of the occipital horns of the lateral ventricles. It was initially described in 1940 by Benda, who classified this type of a malformation as a developmental disorder of the brain vesicles.<sup>13</sup> In 1946, Yakovlev

and Wadsworth introduced the term “colpocephaly” from the Greek term *kolpos*, meaning “hollow,” to name the same brain anomaly.<sup>14</sup> It is a rare disorder of neuronal migration, which usually occurs during the first trimester of pregnancy.<sup>5,15</sup> It should be emphasized that since colpocephaly was first described in the scientific literature, 67 years before the present report, at least 60 cases have been reported worldwide.<sup>1,3-12</sup>

Several clinical entities have been proposed as probable causes of colpocephaly, including (1) perinatal hypoxic–ischemic encephalopathy; (2) chromosome anomalies, such as mosaic trisomies 8 and 9; (3) congenital infections, such as toxoplasmosis and cytomegalovirus; and (4) use of medications by the mother during pregnancy, such as corticosteroids, salbutamol, and theophylline. Genetic factors are also observed in colpocephaly formation, which could stem from either autosomal or X-linked recessive inheritance.<sup>3,5,15</sup>

Unfortunately, the diagnosis of colpocephaly is infrequent during the prenatal period, and it is often mistaken for hydrocephalus.<sup>7</sup> Clinically, colpocephaly can be evidenced by varying degrees of intellectual disability, microcephaly, seizures, muscular spasms, and locomotor or visual abnormalities.<sup>1,3,5,15</sup> It should be noted that patients with colpocephaly will not necessarily have neuronal proliferation disorders or delayed neuropsychomotor development.<sup>3</sup>

Colpocephaly may be associated with severe CNS malformations, such as agenesis of the corpus callosum, lissencephaly, pachygyria, schizencephaly, macrogyria, enlarged cisterna magna, cerebellar atrophy, optic nerve hypoplasia, chorioretinal coloboma, microcephaly, myelomeningocele, and hydrocephalus.<sup>2, 3,16</sup>

Of these possible concurrent congenital anomalies, agenesis of the corpus callosum is most frequently observed, being associated with 40% of colpocephaly cases; magnetic resonance (MR) is the gold standard for its diagnosis.<sup>3,8</sup> Computed tomography may not show a small healthy part of the corpus callosum.<sup>3</sup> Therefore, after a prenatal diagnosis of colpocephaly, neuroimaging examinations should be ordered for a detailed investigation of concurrent neurological malformations.<sup>16</sup>

## CONCLUSION

This reported case together with the current literature elucidates the characteristics of colpocephaly, a rare disorder that should be studied for a complete understanding of its diagnostic, therapeutic, and prognostic aspects. This will make appropriate professional conducts possible, thus ensuring an improvement in the quality of life of the affected patients, both in the mid and long term.

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