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Beyond Hydrocephalus: Colpocephaly Mimicking Neonatal Ventriculomegaly

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ABSTRACT

Colpocephaly is the abnormal dilation of the posterior horns of the lateral ventricles. It is a congenital brain abnormality and is most often associated with partial or complete agenesis of the corpus callosum. A full term neonate was delivered at 39 weeks' gestation by spontaneous vaginal delivery. Antenatal ultrasound had detected right sided ventriculomegaly. The neonate was admitted to the neonatal intensive care unit. He later developed mild respiratory distress. Cranial ultrasound on admission revealed marked ventriculomegaly, suggestive of communicating hydrocephalus. Brain MRI demonstrated global microcephaly, disproportionate ventricular dilation predominantly affecting the occipital horns, radiological features of colpocephaly, and dysgenesis of the corpus callosum. Electroencephalography showed a burst suppression pattern, consistent with epileptic encephalopathy and severe cerebral dysfunction. The infant was maintained on ad lib oral feeds. Mechanical ventilation was required for two days, and intravenous antibiotics were continued for two days. The neonate stabilized clinically and was discharged with antiepileptic therapy and close multidisciplinary follow-up...

Keywords: Colpocephaly, Corpus Callosum, Lateral Ventricles, Children. .

INTRODUCTION

The aberrant dilatation of the lateral ventricles' posterior horns is known as colpocephaly. It is a congenital brain anomaly that is most frequently linked to either partial or whole corpus callosum agenesis [1]. Toxins, infections, chromosomal abnormalities, steroid use during pregnancy, and anoxic encephalopathy are only a few of the many causes. It typically manifests as neurodevelopmental problems in childhood [2]. However, it can manifest as headaches, intellectual impairments, and seizures in older age groups [3].

Since the majority of kids with colpocephaly exhibit severe delayed developmental milestones, seizures, visual abnormalities, and other neurological issues, the condition is usually detected in early childhood. It is very uncommon for neurodevelopmentally normal people to be diagnosed with colpocephaly [1]. Imaging is used to make the diagnosis, and it should primarily be distinguished from arachnoid cysts and NPH. Since there isn't a specific therapy, treatment is symptomatic. Less than 20–30 adult cases of colpocephaly have been documented in the literature as of 2025 [4].

We reviewed the literature and report a case of infant colpocephaly associated with agenesis of the corpus callosum.

CASE PRESENTATION

In a private hospital, a full-term baby weighing 2,975 grams was born via spontaneous vaginal delivery (SVD) at 39 weeks gestation. Active resuscitation was not required because the patient's Apgar scores were 8 at one minute and 9 at five. Mild metabolic acidosis was revealed by cord blood gas analysis: pH 7.22, PCO_2 59.5 mmHg, HCO_3 24 mmol/L, base excess −4.7 mmol/L, and lactate 3.9 mmol/L. The mother, a 30-year-old gravida 5, para 2, had had two previous abortions, one of which was brought on by congenital defects. There were no notable maternal comorbidities, and the pregnancy was straightforward. The mother was given intrapartum ceftriaxone antibiotic prophylaxis after an antenatal Group B Streptococcus (GBS) screening came back negative. Antibiotics were given around four hours before delivery, and the membranes spontaneously burst with clear amniotic fluid. Right-sided ventriculomegaly was discovered by antenatal ultrasonography.

Shortly after birth, the neonate developed jitteriness and transient hypoglycemia, which resolved with feeding. The neonatal intensive care unit (NICU) received the newborn. Later on, he experienced modest respiratory distress with nasal noises and chest recessions; these symptoms went away on their own without the need for more oxygen. A chest X-ray revealed no signs of pneumonia or serious lung disease, and a sepsis workup that included blood cultures and inflammatory markers was unremarkable. In the event that there was no infection, empirical intravenous antibiotics (gentamicin and ampicillin) were stopped after 48 hours. A cranial ultrasound showed significant ventriculomegaly, which may indicate communicating hydrocephalus.

For additional evaluation and care, the newborn was moved to our tertiary hospital with the diagnosis of communicating hydrocephalus for further work up after cranial ultrasonography. US finding Marked ventriculomegaly of the lateral ventricles, with mild to moderate dilatation of the third and fourth ventricles, findings suggestive of communicating hydrocephalus.

Anthropometric measures on admission to General Ahmadi hospital showed a weight at the 25th percentile and a head circumference at the third percentile. Findings were not consistent with hydrocephalus.

The neonatologist's point-of-care ultrasound (POCUS) of the brain revealed no cranial suture diastasis, a small anterior fontanelle, and a small head with a retreating forehead. The posterior horns of both lateral ventricles were severely and disproportionately enlarged, while the anterior horns were somewhat spared. There was no ventricular seeping or ballooning, and the

third and fourth ventricles looked normal. The results were consistent with colpocephaly and potential hypoplasia of the corpus callosum, and the ventricles seemed almost parallel to one another. With no indication of intraventricular hemorrhage, a formal cranial ultrasonography supported the diagnosis of colpocephaly by confirming diffuse ventricular dilatation, which was particularly noticeable in the occipital horns of the lateral ventricles (Figure 1).

After the infant experienced neurological seizures, endotracheal intubation and non-invasive intermittent positive pressure ventilation (NIPPV) were required to enable brain magnetic resonance imaging (MRI). Intravenous phenobarbital was used to manage acute seizures and levetiracetam (Keppra) was started as maintenance antiepileptic therapy. Global microcephaly, excessive ventricular dilatation mostly affecting the occipital horns, radiological signs of colpocephaly, and corpus callosum dysgenesis were all seen on brain MRI (Figure 2). A burst suppression pattern on electroencephalography (EEG) was indicative of significant brain dysfunction and epileptic encephalopathy.

Elevated aspartate aminotransferase (AST), gamma-glutamyl transferase (GGT), direct and total bilirubin, corrected calcium, platelet count, hematocrit, and anion gap were among the mild abnormalities found in the biochemical investigations. These abnormalities were all within ranges that did not indicate severe hepatic or renal dysfunction.

There was no indication of hemolysis or systemic infection, and the TORCH test came out negative. Other than that, the physical examination revealed normal facial features, ears, eyes, chest, and belly. Neurological examination was notable for microcephaly and seizure activity; primitive neonatal reflexes were present, with no obvious dysmorphic features.

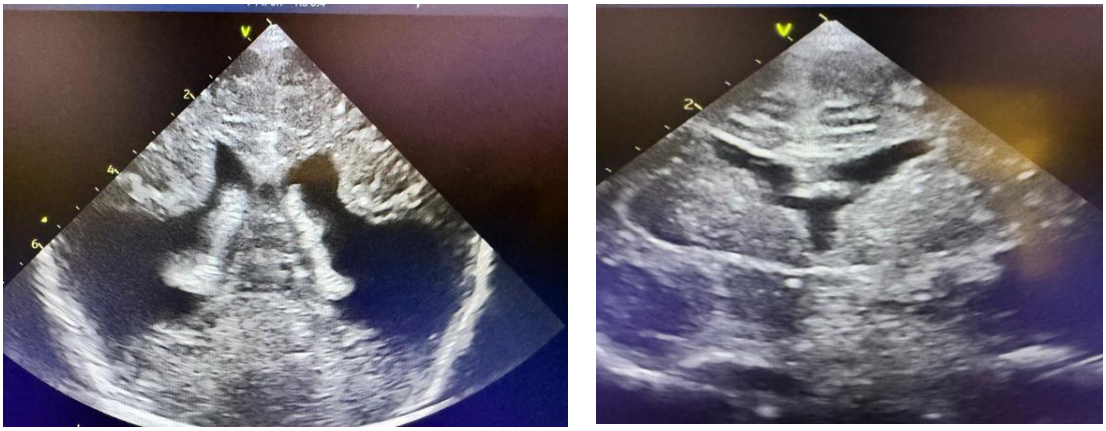

Figure 1: Diffusely dilated ventricular system; significantly more at the occipital horns of lateral ventricles (disproportionate enlargement)

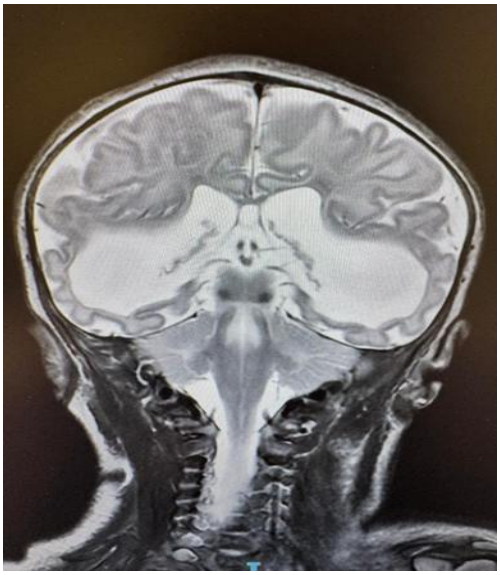

Figure 2A: Disproportionate dilatation of the ventricular system predominantly the occipital horns that are seen much dilated (Colpocephaly) . Dysgenesis of the corpus callosum

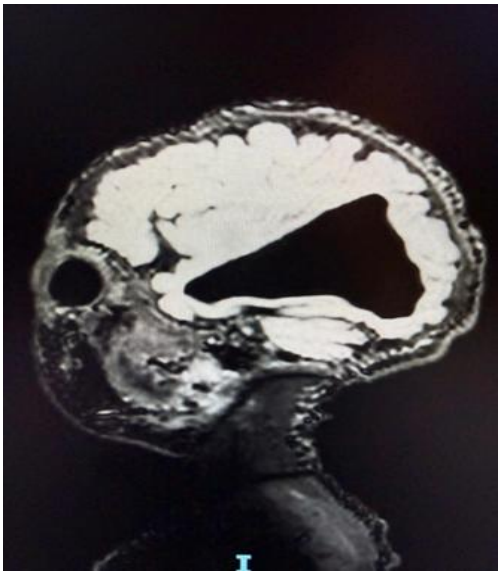

Figure 2B: Sagittal view showing dilatation of the ventricular system predominantly the occipital horns

Management and Outcome:

There were no biochemical or clinical indications of hepatic or renal impairment, and the baby was kept on ad lib oral feedings. Intravenous antibiotics were administered for two days, and mechanical ventilation was necessary for a total of two days. Pediatric neurology and ophthalmology provided consultations. Metabolic screening was finished, and routine vaccinations were given in accordance with the national schedule. In order to rule out non-ketotic hyperglycinemia, cerebrospinal fluid (CSF) amino acid and glycine levels were submitted. If the results of the initial focused testing were negative, genetic counseling was given along with DNA banking for possible whole exome sequencing.

The baby's clinical development was satisfactory, and he gained enough weight. In three weeks, a follow-up is scheduled, and the CSF amino acid values continued to be assessed. At 1-year follow-up, growth and neurodevelopment were appropriate for his age. Further genetic evaluation was planned to determine the underlying cause and guide long-term care.

Discussion

Benda [5] reported a case series in 1941. The first patient had seizures and physical and intellectual impairments, whereas the second patient had birth asphyxia. These examples are recognized as the earliest documented cases of colpocephaly, despite Benda's usage of the term "vesiculopathy." Yakovlev and Wadsworth coined the name "colpocephaly" in 1946 [6].

Perinatal ischemic encephalopathy, intrauterine infections, maternal toxin exposure, and aberrant neuronal migration or white matter formation arrest during fetal development are among the hypothesized causes [3]. It has been observed that certain patients have a history of illnesses, including meningococcal meningitis and congenital toxoplasmosis [7].

Genetic disorders, specifically trisomy 8 and 9, have also been linked to colpocephaly. Colpocephaly, or a retained fetal ventricular shape, might theoretically result from any intrauterine event that can disrupt fetal neurodevelopment [8]. The main neurodevelopmental cause of colpocephaly is thought to be the lack of white matter surrounding the occipital horns. An X-linked or autosomal dominant inheritance pattern has also been proposed based on twin and sibling colpocephaly instances [9].

The theory that colpocephaly might actually be the consequence of an anatomic abnormality brought on by white matter developmental arrest was also supported by later papers [10]. The lateral ventricles develop as big cavities of the telencephalic vesicle, according to an embryological theory [11].

Agenesis of the corpus callosum, neuronal migration disorders (lissencephaly, pachygyria), schizencephaly, microgyria, macrogyria, enlargement of the cisterna magna, cerebellar atrophy, optic nerve hypoplasia, chorioretinal coloboma, microcephaly, meningomyelocele, and hydrocephalus have all been linked to colpocephaly [12].

Insults that take place at any point between one and four months of gestation may be linked to the many brain abnormalities linked to colpocephaly. Polymicrogyria is an insult to the neurological system that occurred prior to five months of gestation.

Intrauterine hypoxia and congenital infections (such as CMV and toxoplasmosis) are potential causes of polymicrogyria [13]. Colpocephaly and polymicrogyria seem to have similar origins and onset periods. Micrognathia, hypoplastic nails, simian creases, Pierre-Robin syndrome, and neurofibromatosis were among the other related abnormalities. Periventricular leukomalacia, which results from the destruction of the white matter of the occipital lobe, is another explanation for the development of proportionally enlarged occipital horns of the lateral ventricles [14].

Furthermore, our patient's clinical and neuroimaging characteristics are similar to those of El-Hattab-Alkuraya syndrome, an autosomal recessive neurodevelopmental condition brought on by biallelic pathogenic mutations in the WDR45B gene [15]. Congenital or increasing microcephaly, global developmental delay, early-onset seizures, and spastic quadriplegia are the hallmarks of this syndrome, which is frequently linked to cerebral atrophy and ventricular dilatation on brain MRI. The severe effects of WDR45B malfunction on the development and maintenance of the central nervous system are highlighted by the fact that ventricular enlargement in reported cases is usually secondary to parenchymal loss (ex-vacuo), and affected people may also exhibit delayed myelination and brainstem atrophy [15].

The presence of severe microcephaly, epileptic encephalopathy, and marked ventricular dilatation (especially colpocephaly-like configuration) raises the possibility of a shared pathophysiological pathway and emphasizes the significance of taking El-Hattab-Alkuraya syndrome into consideration in the differential diagnosis of neonates presenting with severe neurodevelopmental impairment and complex brain malformations [16].

Colpocephaly is rarely diagnosed during pregnancy because it is frequently mistaken for hydrocephalus. Clinically, it might show up as varied degrees of microcephaly, seizures, muscle spasms, locomotor or visual abnormalities, and intellectual incapacity. It should be mentioned that neuropsychomotor abnormalities are not always present in patients with colpocephaly [17].

MRI and CT (computed tomography) scans are the gold standard for diagnosing colpocephaly. The occipital horns of the lateral ventricles inflating proportionately to the frontal horns is one of the characteristic symptoms. The maximum width of the posterior horns divided by the width of the anterior horns of the lateral ventricles is the posterior–anterior (P/A) ratio, which some researchers have proposed measuring. The diagnosis of colpocephaly is supported by a P/A ratio larger than 3, which denotes aberrant expansion of the occipital horns. In particular, it is important to differentiate colpocephaly from NPH; in this case, evaluating the P/A ratio is useful [18].

Colpocephaly is an uncommon type of ventriculomegaly, according to Assem et al. [19]. Because colpocephaly is uncommon and frequently misdiagnosed as hydrocephalus at normal pressure, its prenatal and postnatal diagnosis is challenging. As a result, appropriate postnatal imaging and early, multidisciplinary, well-codified care, including its diagnostic, therapeutic, and prognostic aspects, are required [20].

CONCLUSION

This case highlights the importance of distinguishing colpocephaly from hydrocephalus in neonates with antenatal ventriculomegaly, and underscores the role of early MRI, EEG, and multidisciplinary follow-up in defining associated brain malformations and prognosis.

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