Radiology Case Reports 19 (2024) 2048–2051

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journal homepage: www.elsevier.com/locate/radcr

Case Report

Colpocephaly in an adult: A rare case report ☆

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ARTICLE INFO

Article history:
Received 28 January 2024
Revised 6 February 2024
Accepted 12 February 2024

Keywords:
Colpocephaly
Corpus callosum agenesis
Corpus callosum dysgenesis
Seizure
Headache
Lateral ventricles

ABSTRACT

Colpocephaly is the disproportionate enlargement of the occipital horns of the lateral ventricles. It is usually diagnosed in the neonatal period or early childhood due to symptom severity. Adult cases of colpocephaly are rarely reported and often incidentally diagnosed. We report a case of colpocephaly with partial agenesis of the corpus callosum in a 30-year-old female with no past medical history. The patient presented after a syncopal episode with associated complaints of dizziness, vomiting, and chronic intermittent headaches. This case highlights the clinical and radiological features of colpocephaly in adults.

Introduction

Colpocephaly is the disproportionate enlargement of the occipital horns of the lateral ventricles [1–4]. It is a congenital brain abnormality and is most often associated with partial or complete agenesis of the corpus callosum [1,5]. It is typically diagnosed in early childhood as most patients present with severely delayed developmental milestones, seizures, visual abnormalities, and other neurological disorders [4–6]. Diagnosis of colpocephaly in neurodevelopmentally normal adults is extremely rare [3,4,6].

To the best of our knowledge, there are less than 20 publications describing colpocephaly in adult patients in current literature. With this case, we aim to highlight the radiological findings of colpocephaly in an asymptomatic adult.

Case report

A 30-year-old female with no past medical history presented to the medicine outpatient department after a syncopal episode. The episode was preceded by dizziness, headache, and vomiting. According to her family members, it lasted for 2-3 minutes. Fecal incontinence, stiffening of the body, and throat sounds were also reported by the family. The patient denied any confusion before or after the episode. Apart from a similar short episode 2 months ago and frequent intermittent headaches, she reported being healthy throughout her life and had no complaints.

A neurology consult was obtained. With seizure disorder as the preliminary diagnosis, electroencephalogram (EEG), brain imaging, and routine laboratory investigations were ordered.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2024.02.035

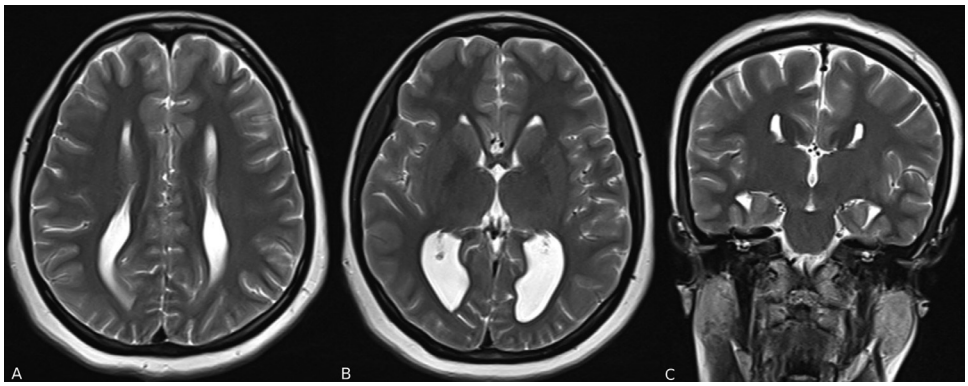

Fig. 1 – (A) Axial T2 MRI shows parallel orientation and separation of frontal horns with separation and enlargement of occipital horns of the lateral ventricles. (B) Axial T2 MRI shows enlargement of occipital horns of lateral ventricles (colpocephaly), also known as the "racing car sign" seen due to partial agenesis of the corpus callosum. (C) Coronal T2 MRI shows near-parallel orientation of frontal horns and eversion of cingulate gyri into the frontal horns bilaterally, also known as "moose head appearance" or "Viking helmet sign" seen due to partial agenesis of the corpus callosum.

Magnetic resonance imaging (MRI) obtained on a 3 T magnet demonstrated enlarged occipital lobes of the lateral ventricles with the "racing car" sign and "moose head appearance" or "Viking helmet sign" (Fig. 1) and partial agenesis of the corpus callosum (Fig. 2). The patient had no known previous imaging for comparison. Complete blood count showed an elevated white blood cell count (13,100/mm^3). Differential leukocyte count showed neutrophilia (91.8%) and lymphocytosis (6.20%). EEG done with an awake patient demonstrated mild intermittent diffuse slowing. The patient was prescribed levetiracetam and sodium valproate and regular follow-ups were scheduled.

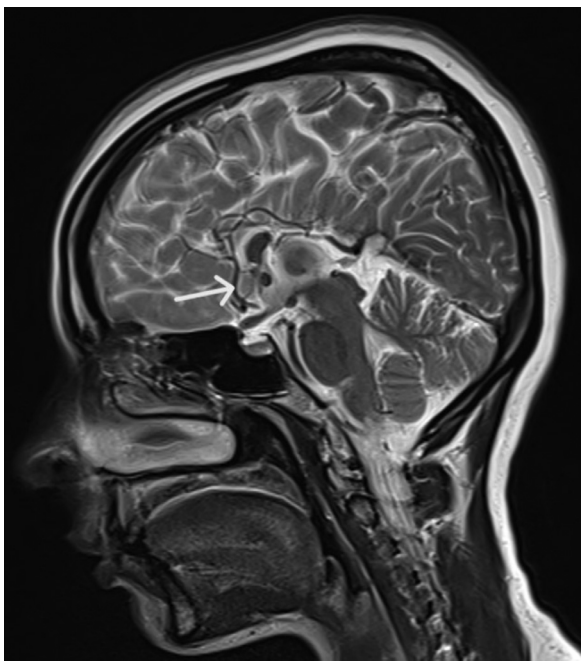

Fig. 2 – Sagittal T2 MRI shows the presence of a small distorted genu (arrow) with a prominent anterior commissure. Body, rostrum, and splenium of the corpus callosum are not visualized (partial agenesis of the corpus callosum).

Discussion

In 1941, Benda published a case series, with the first patient suffering from seizures, and intellectual, and physical disabilities and the second patient suffering from birth asphyxia. Although Benda used the term "vesiculopathy", these cases are widely known as the first-ever recorded cases of colpocephaly [7]. The term "colpocephaly" was first used by Yakovlev and Wadsworth in 1946 [8].

Postulated etiologies include abnormal neuronal migration or white matter formation arrest during fetal development [1,3], intrauterine infections, maternal toxin exposure, and perinatal ischemic encephalopathy [3]. Some patients have been noted to have a past medical history of infections such as congenital toxoplasmosis [9] and meningococcal meningitis [1]. Colpocephaly has also been associated with genetic abnormalities, namely trisomy 8 and 9 [10]. Any intrauterine process that can interfere with fetal neurodevelopment can theoretically lead to a retained fetal ventricular shape, i.e. colpocephaly [9,11]. Neurodevelopmentally, the absence of white matter around the occipital horns is considered the primary cause of colpocephaly [12]. Based on cases of colpocephaly involving siblings and twins, an X-linked or autosomal dominant inheritance pattern has also been suggested [10,11].

Apart from abnormalities of the corpus callosum, other associated malformations include microcephaly, myelocele, macrogyria, microgyria, schizencephaly, lissencephaly, cerebellar atrophy, optic nerve atrophy, Chiari malformation, periventricular leukomalacia, and enlargement of the cisterna magna [1,3,9,10,13]. In 1941, Benda reported the young patient's autopsy findings, namely agenesis of the corpus callosum, dilated lateral ventricles, thin and undifferentiated occipital lobes, and microgyria [7]. In recent times, cases with

concurrent agenesis of the corpus callosum have been diagnosed prenatally using antenatal MRI [12]. The estimated prevalence of corpus callosum agenesis in the general population is 3-7 per 1000 births, as compared to 2-3 per 100 births in children with developmental abnormalities [1]. In current literature, all adults with colpocephaly were noted to have concurrent partial or complete agenesis of the corpus callosum, except for 3 cases. Out of these 3 cases, only 1 patient was diagnosed with colpocephaly with normal brain parenchyma [14]. The other 2 cases were associated with porencephaly [15] and an absent circle of Willis [10].

Review of current literature suggests colpocephaly is often an incidental diagnosis in adults. Adult patients usually present with nonspecific symptoms such as headache [1,2,3,10], dizziness [2,6], seizures [6,11], vertigo [1], nausea [6], motor abnormalities [16], sensory abnormalities [12], cognitive abnormalities [15,17], intellectual disability [18], learning disability [17], gait instability and falls [9], loss of consciousness [13] and visual hallucinations [19]. Gungor et al reported a case with postmortem diagnosis of colpocephaly in a patient with a lifelong history of poor academic performance [20]. Some cases had other primary diagnoses such as meningioma [2] and paraspinal abscess as a complication of spine surgery [4]. Some patients were diagnosed with colpocephaly after being admitted for medical management of respiratory tract infection [13] and hypertensive episode [14]. Concurrent psychiatric disorders were also reported in some cases, such as schizophrenia [21] and obsessive-compulsive disorder (OCD) [22]. Our patient presented with syncope, vomiting, frequent long-standing headaches, and dizziness, and colpocephaly was an incidental diagnosis. Apart from a short-lived similar episode of syncope a few months ago, she had no past medical history and no known perinatal complications. Our patient denied any history of neurobehavioral symptoms.

As most patients present with nonspecific symptoms, computed tomography (CT) is typically the initial investigation done at most institutions, with MRI being the imaging of choice [1]. This is further supported by a case of colpocephaly associated with partial agenesis of the corpus callosum in which CT did not show a small fragment of the corpus callosum [1]. Radiologically, colpocephaly is diagnosed by a disproportionate enlargement of the occipital horns with the presence of normal anterior horns of the lateral ventricle [9]. When associated with dysgenesis or agenesis of the corpus callosum, findings usually include disproportionate occipital horn dilation with an abnormal separation of frontal horns. The frontal horns are typically parallel in orientation [1], and the same was noted in our patient.

Noorani et al. described the posterior-to-anterior ratio (P/A ratio), which refers to the ratio of the maximal width of the occipital horn and the maximal width of the anterior horn of the lateral ventricle [5]. A P/A ratio greater than or equal to 3 represents disproportionate dilation of the occipital horns, making colpocephaly more likely [5,9,13]. Despite its high specificity, the P/A ratio has low sensitivity, and the radiological diagnosis of colpocephaly is mainly based on the visualization of typical features on imaging [3]. Our case supports the low sensitivity of the P/A ratio, as it was only 1.8 for our patient. Dysgenesis or agenesis of the corpus callosum concurrent with colpocephaly may give rise to the "racing car" sign, representing the frontal horns, body, and occipital horns of the lateral ventricles bearing resemblance to a racing car on axial imaging [23]. Another sign associated with agenesis of the corpus callosum is "moose head appearance" or the "Viking helmet" sign, seen on coronal imaging at the level of the frontal horns [23]. Our patient's imaging demonstrated both the abovementioned signs. In a study of 90 patients, Durmaz et al. reported that if the occipital horn of the lateral ventricle was greater than 20 mm and the other horns were smaller than 7 mm, colpocephaly could be predicted with a sensitivity and specificity of 94% and 95% respectively [24]. Nevertheless, the sensitivity and specificity of these radiological findings need to be studied further.

The most important differential diagnosis of colpocephaly is normal pressure hydrocephalus (NPH) [3,4,9,13,14]. Clinically, colpocephaly is associated with nonspecific symptoms whereas NPH usually presents with the triad of dementia, gait abnormalities, and urinary incontinence. The term "probable NPH" is used when a patient presents with at least 2 of the 3 triad features, normal opening pressure, and radiological evidence of ventricular system dilation that is not proportionate to the level of brain atrophy [9]. Evans index (ratio of maximum width of the frontal horns of the lateral ventricles and the maximum internal diameter of the skull at the same level; a ratio >0.30 indicates ventriculomegaly) and callosal angle (angle between medial superior borders of the left and right ventricle through the posterior commissure, perpendicular to the anterior-posterior commissure on coronal images; an acute angle can indicate idiopathic NPH) are 2 imaging markers that are often used in the diagnosis of NPH [12]. Esenwa et al noted high specificity of the P/A ratio for colpocephaly in cases where the differential diagnosis was NPH [9].

There is no specific preferred management for colpocephaly [1]. Asymptomatic adults in whom colpocephaly is diagnosed incidentally usually require no treatment [3,11]. For symptomatic adult patients, management is symptomatic: the most common example being antiepileptics for patients presenting with seizures [11,12,19]. Our patient was prescribed antiepileptics due to the symptoms reported by her family as well as abnormal EEG findings. For patients presenting with cognitive or neurological symptoms, a multidisciplinary approach involving neurology, occupational therapy, speech therapy, and physiotherapy can be beneficial [1,19].

Conclusion

Colpocephaly is a rare congenital brain malformation that is typically known as a pediatric diagnosis. Adults being diagnosed with colpocephaly is exceptionally rare, with most patients presenting with nonspecific symptoms. Partial or complete agenesis of the corpus callosum is the most commonly associated malformation. The most important differential diagnosis in adults is NPH. Since it is not well studied in adults, this case aims to contribute to and summarize the existing adult cases of colpocephaly. Knowing the clinical and radiological features and associations is important to avoid unnecessary diagnostic and therapeutic interventions.

Patient consent

Written informed consent for the publication of this case report was obtained from the patient.

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