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Rhombencephalosynapsis with Aqueductal Stenosis: A Rare Cause of Adolescent Hydrocephalus

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ABSTRACT

Rhombencephalosynapsis (RES) is a rare congenital malformation of the hindbrain characterized by fusion of the cerebellar hemispheres due to partial or complete agenesis of the cerebellar vermis. It presents with a spectrum of neurological manifestations, including motor dysfunction, cognitive delay, and hydrocephalus secondary to aqueductal stenosis.

We report the case of a 15-year-old female who presented with a six-month history of progressive headache and blurred vision, along with developmental delay. MRI revealed classic features of RES, including complete agenesis of the cerebellar vermis and fusion of the bilateral cerebellar hemispheres. Additional findings included fusion of the inferior colliculi and absence of the septum pellucidum. The supratentorial ventricular system was moderately dilated, indicating hydrocephalus secondary to stenosis at the level of the aqueduct of Sylvius. The corpus callosum appeared thinned and showed upward bowing, suggestive of chronically elevated intracranial pressure. A thinned, upward-bowed corpus callosum was noted, suggestive of chronic raised intracranial pressure.

This case underscores the importance of early neuroimaging in children with neurological symptoms and developmental delay. Recognizing RES and its associated anomalies is essential for appropriate management, including CSF diversion procedures and neurodevelopmental support.

Keywords: Rhombencephalosynapsis, Aqueductal stenosis, Hydrocephalus, Cerebellar malformation, MRI, Adolescent neurology

1. INTRODUCTION

Rhombencephalosynapsis (RES) is a rare congenital brain malformation characterized by midline fusion of the cerebellar hemispheres due to partial or complete loss of the intervening vermis. [1–3] The exact etiology remains unclear, but one hypothesis suggests that it results from dorsal–ventral patterning defects during brain development, leading to midline loss and fusion of lateral structures. [2] Unlike other vermian malformations such as Dandy-Walker complex, Joubert syndrome, or tectocerebellar dysraphia, RES does not involve cerebellar hemisphere separation but rather a failure of differentiation, resulting in undivided hemispheres. [4]

Since the cerebellum develops from both the inferior mesencephalon and metencephalon, mesencephalic anomalies are often associated with RES. The quadrigeminal plate may be dysmorphic, leading to deformation of the cerebral aqueduct and noncommunicating hydrocephalus of varying severity. Many patients also exhibit partial or complete absence of the septum pellucidum, which may be congenital or secondary to chronic hydrocephalus. Aqueductal stenosis, although inconsistently present, is frequently observed in RES, likely due to genetic defects at the mesencephalic-metencephalic junction. [2]

2. CLINICAL PRESENTATION AND IMAGING FINDINGS

Patient History

A 15-year-old female presented with a six-month history of progressively worsening headache and blurred vision. Her medical history was notable for global developmental delay, primarily affecting motor milestones and cognitive function. There was no history of seizures, trauma, or prior neurological illness. Family history was unremarkable for congenital or hereditary neurological disorders. Ophthalmologic evaluation revealed bilateral papilledema, suggestive of increased intracranial pressure. The remainder of the systemic examination was unremarkable.

MRI Findings

Magnetic resonance imaging (MRI) of the brain demonstrated characteristic features of RES. The cerebellar vermis was completely absent, with fusion of the bilateral cerebellar hemispheres, resulting in a continuous folial pattern across the midline. The inferior colliculi were fused, and the septum pellucidum was absent. A single midline dentate nucleus was observed instead of the usual paired structures. The middle cerebellar peduncles were either absent or severely hypoplastic, while the superior cerebellar peduncles were also underdeveloped. There was stenosis at the level of the aqueduct of Sylvius, leading to obstructive hydrocephalus with moderate dilation of the lateral and third ventricles. A thinned, upwardly bowed corpus callosum suggested chronic intracranial pressure changes.

3. MANAGEMENT PLAN

Given the presence of hydrocephalus, the patient was referred for neurosurgical evaluation. The options for treatment included cerebrospinal fluid (CSF) diversion procedures such as ventriculoperitoneal (VP) shunting.

This case highlights the importance of early neuroimaging in children presenting with neurological symptoms and developmental delays. Recognizing RES and its associated anomalies is crucial for guiding appropriate management, including neurosurgical intervention for hydrocephalus and supportive therapy for neurodevelopmental impairments.

Clinical Features and Associated Anomalies

Clinically, RES presents with a broad spectrum of neurological symptoms, largely influenced by associated supratentorial anomalies. Common features include truncal and limb ataxia, hypotonia, spasticity, abnormal eye movements, strabismus, dysarthria, and developmental delays. The severity of impairment varies, though most cases do not significantly restrict daily activities. Cognitive deficits are frequent, with attention disorders and dysfunction in the cerebellar-thalamo-prefrontal circuit being common. Some cases also report self-mutilation and obsessive-compulsive behaviors, though normal cognitive function has been observed in certain individuals. [3]

Hydrocephalus, often associated with aqueductal stenosis, is the most frequently observed supratentorial anomaly in RES. Other supratentorial abnormalities include fused thalami, fornices, and cerebral peduncles, absence of the septum pellucidum, limbic system dysgenesis, cortical malformations, and multiple suture synostoses. Patients with additional supratentorial anomalies and rudimentary cerebellar development tend to exhibit more severe symptoms, including hypotonia, motor dysfunction, cerebellar ataxia, strabismus, and developmental delays. [5] Given the clinical suspicion of raised intracranial pressure, neuroimaging was performed.

4. RADIOLOGIC DIAGNOSIS

MRI Characteristics

Axial MRI best demonstrates fused hemispheres, while coronal images highlight the horizontal folial orientation. Fusion of the superior and middle cerebellar peduncles, dentate nuclei, and inferior colliculi is also common, leading to a characteristic diamond-shaped, narrowed, and posteriorly oriented fourth ventricle. However, this feature is not consistently present. High-resolution volumetric sequences may be necessary to accurately assess cerebellar peduncle fusion. Additionally, olivary nuclei may be hypoplastic or absent. [6]

MRI plays a crucial role in diagnosing RES, particularly in patients with unexplained developmental delay. [3] Since cognitive function may be preserved, the diagnosis is often delayed until adulthood.

CT vs. MRI

While CT imaging has been used in some cases to reveal the characteristic keyhole-shaped fourth ventricle and associated supratentorial ventriculomegaly, its utility is limited by artefacts that obscure detailed evaluation of the posterior fossa. MRI, on the other hand, provides superior imaging capabilities, offering high-resolution, multiplanar views that allow for better differentiation between white and gray matter. This makes MRI the most effective imaging modality for assessing the posterior fossa, particularly for visualizing small but crucial structures such as the dentate nuclei, cerebellar peduncles, and colliculi. [7]

5. CONCLUSION

MRI played a pivotal role in diagnosing RES in this adolescent patient. The absence of the cerebellar vermis and fusion of the cerebellar hemispheres (with a continuous midline folial pattern and single dentate nucleus) are pathognomonic MRI findings of RES. These, combined with fused midbrain colliculi and resultant obstructive hydrocephalus, reflect a developmental insult at the midbrain-hindbrain junction. Recognizing RES and its associated features is important for guiding management: unlike other vermian malformations (e.g., Dandy-Walker or Joubert syndromes), RES frequently necessitates treatment of non-communicating hydrocephalus. Early neurosurgical intervention for CSF diversion, along with supportive therapies, can greatly improve outcomes. This case illustrates how understanding the embryologic basis of the malformation explains its coexistent anomalies and helps guide comprehensive patient care.

REFERENCES

[1] Ishak GE, Dempsey JC, Shaw DWW, Tully H, Adam MP, Sanchez-Lara PA, et al. Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain. 2012 May;135(5):1370–86.

[2] Whitehead MT, Choudhri AF, Grimm J, Nelson MD. Rhombencephalosynapsis as a cause of aqueductal stenosis: an under-recognized association in hydrocephalic children. Pediatr Radiol. 2014 Jul;44(7):849–56.

[3] Paprocka J, Jamroz E, Ścieszka E, Kluczewska E. Isolated rhomboencephalosynapsis – a rare cerebellar anomaly. Pol J Radiol. 2012;77(1):47–9.

[4] Guyot LL, Kazmierczak CD, Michael DB. Adult rhombencephalosynapsis: Case report. J Neurosurg. 2000 Aug;93(2):323–5.

[5] Truwit CL, Barkovich AJ, Shanahan R, Maroldo TV. MR imaging of rhombencephalosynapsis: report of three cases and review of the literature. American journal of neuroradiology. 1991 Sep 1;12(5):957–65.

[6] Mendonça JLF, Natal MRC, Viana SL, Coimbra PPA, Viana M a. CB, Matsumine M. Rhombencephalosynapsis: CT and MRI findings. Neurol India. 2004 Mar;52(1):118.

[7] Danon O, Elmaleh M, Boukobza B, Fohlen M, Hadjnacer K, Hassan M. Rhombencephalosynapsis diagnosed in childhood: Clinical and MRI findings.

FIGURE LEGENDS:

Figure 1:

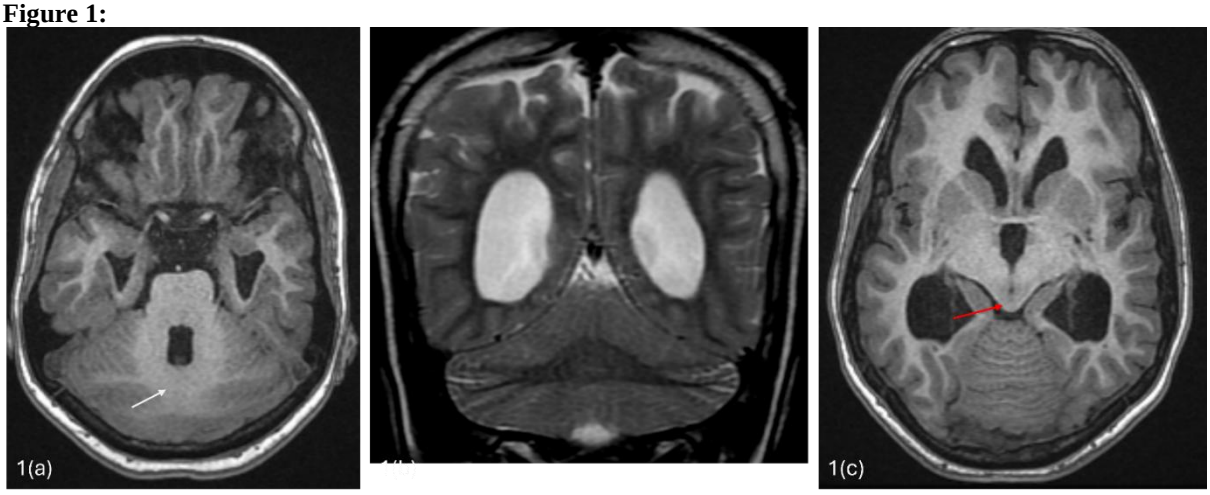

Figure 1A: Axial T1-weighted image demonstrates the absence of the cerebellar vermis (white arrow) with fusion of the bilateral cerebellar hemispheres, consistent with rhombencephalosynapsis.
Figure 1B: Coronal T2-weighted image confirms these findings, showing uninterrupted continuity of the white matter across the bilateral cerebellar hemispheres.
Figure 1C: Axial T1-weighted image highlights the fusion of the inferior colliculi (red arrow), further supporting the diagnosis.

Figure 2:

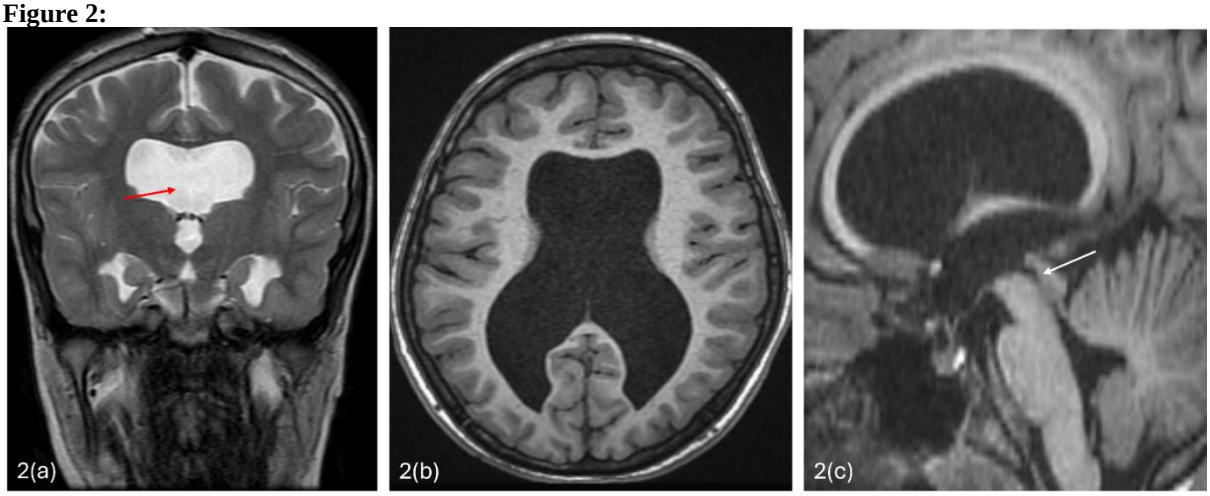

Figure 2A: Coronal T2-weighted image demonstrates the absence of the septum pellucidum (red arrow), indicative of a midline developmental anomaly.
Figure 2B: Axial T1-weighted image further confirms the non-visualization of the septum pellucidum, accompanied by moderate dilation of the supratentorial ventricular system, suggestive of associated hydrocephalus.
Figure 2C: Sagittal T1-weighted image reveals stenosis at the level of the aqueduct of Sylvius (white arrow), contributing to the ventricular dilatation. Additionally, mild thinning and upward bowing of the corpus callosum are noted, suggestive of chronic intracranial pressure changes.