Rhombencephalosynapsis Unmasked: A Rare Congenital Anomaly Masquerading as Progressive Cognitive Decline

Jeongwon Lee, DO; Kyle Hamann, DO; Arjun Bagai, DO; Dhruv Patel, DO; Bruce Dodt, MD
Riverside Medical Center, Kankakee, Illinois

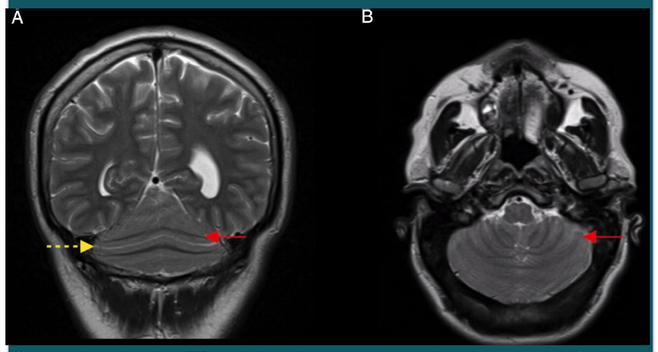
Background

Rhombencephalosynapsis (RES) is a rare congenital cerebellar malformation characterized by partial or complete agenesis of the vermis with midline fusion of the cerebellar hemispheres, dentate nuclei, and superior cerebellar peduncles. RES in an adult is an exceedingly rare phenomenon, with some sources stating an incidence of approximately 1 per 1,000,000. We present a case of RES in a 42-year-old woman first diagnosed during outpatient evaluation for progressive cognitive decline, illustrating variable phenotypic expression and diagnostic challenges.

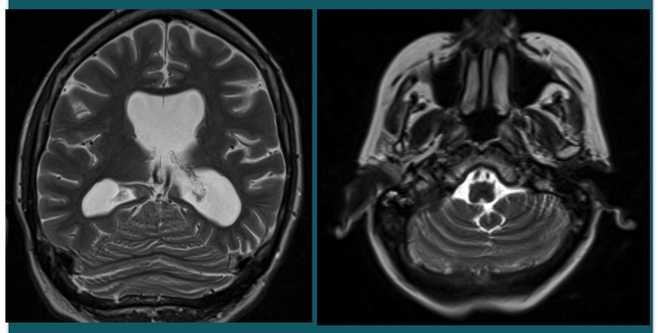
Rhombencephalosynapsis (RES) is a rare congenital cerebellar malformation characterized by partial or complete agenesis of the vermis with midline fusion of the cerebellar hemispheres, dentate nuclei, and superior cerebellar peduncles. RES in an adult is an exceedingly rare phenomenon, with some sources stating an incidence of approximately 1 per 1,000,000. We present a case of RES in a 42-year-old woman first diagnosed during outpatient evaluation for progressive cognitive decline, illustrating variable phenotypic expression and diagnostic challenges.

Case Presentation

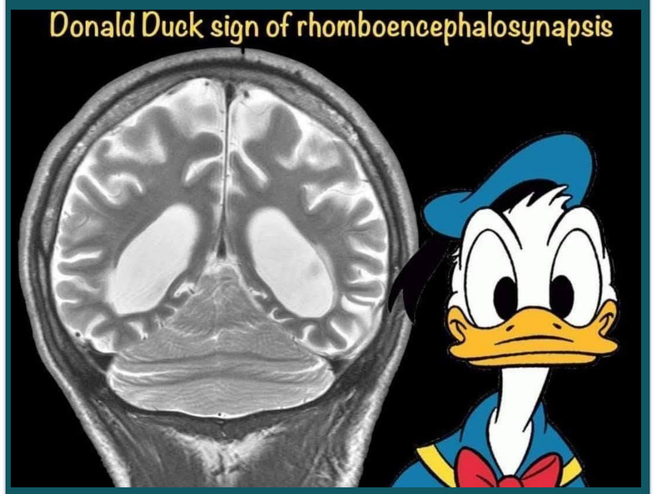
The patient is a 42-year-old female with a history of traumatic brain injury (TBI) complicated by post-traumatic seizure disorder managed with lacosamide, right eye blindness from prior trauma, and alcohol use disorder who presented with worsening memory impairment and balance difficulties over 3-4 months, resulting in multiple falls. Prior history was notable for delayed bipedal ambulation at 17 months, childhood strabismus diagnosed at age 4, frequent falls with multiple head injuries, and difficulty learning motor tasks. Despite this, she excelled academically and completed a university level education. Neurological examination revealed significant memory deficits and decreased attention span. Gait and balance were notably impaired. Prior CT imaging demonstrated stable ventriculomegaly when compared to prior brain imaging. As repeated CT scans of the brain failed to show acute pathological changes coupled with progressive clinical decline, an MRI brain was obtained revealing continuity of the right and left cerebellar hemispheres consistent with RES, mild lateral ventriculomegaly, symmetric hippocampi, and age-appropriate cerebral volume loss.



Coronal and axial T2-weighted sequences, showing partial fusion of the cerebellar hemispheres and continuity of the folia (solid red arrow) and fissures (dashed yellow arrow) across the midline



MRI brain without contrast 9/6/2025
Continuity of the right and left cerebellar hemispheres consistent with rhomboencephalosynapsis.
There are age appropriate chronic white matter microvascular changes.



Conclusions

RES results from defective dorsoventral patterning early on in embryogenesis. Clinical presentation can vary widely from mild truncal ataxia to severe developmental delay and early mortality. Classic features include developmental motor delay, ataxia, abnormal eye movements, and characteristic head stereotypies. In older populations, neuropsychiatric manifestations are much more common including emotional dysregulation, increased impulsivity, and impairment in executive function. Confirmatory MRI findings aid diagnosis, including absent or hypoplastic cerebellar vermis, midline fusion of cerebellar hemispheres with transversely oriented folia, fused dentate nuclei, and a pathognomonic keyhole or diamond-shaped fourth ventricle. Evidence of RES on imaging should prompt further investigation for other malformations particularly vertebral, rib, renal, limb, and cardiac abnormalities as patients with VACTERL features and/or severe RES are at greater risk for severely abnormal clinical outcome. No curative treatment is available, though surgical intervention may be indicated for associated hydrocephalus via ventriculoperitoneal shunt or endoscopic third ventriculostomy. This case demonstrates the need to consider RES as part of a broader differential when evaluating certain constellations of neurological symptoms. The patient's retrospective medical history with delayed developmental milestones, childhood strabismus, and motor difficulties characterize classic presentation of RES that went unrecognized until advanced neuroimaging. Increased awareness of RES and appropriate imaging findings may prompt thorough evaluation of lifelong motor/developmental histories and guide multidisciplinary management including seizure management, neuropsychiatric evaluation, nutrition optimization and neurorehabilitation.

Disclosure Information

Nothing to disclose.