CASE REPORT AND REVIEW OF LITERATURE

A Rare Cerebellar Malformation: The Donald Duck Sign

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Abstract

In the developing world, congenital developmental disorders are very common to get identified quickly and early by advanced medical technologies. A rare congenital central nervous system developmental condition is called rhombencephalosynapsis (RES). The origin and pathogenesis of RES are still unknown, even though it has been identified for many years. Its symptoms overlap because it occurs in conjunction with other syndromes. The severity of the cerebellar vermismal development or malformation determines how this condition manifests clinically. Here, we describe a 52-year-old male who was found to have a cerebellar malformation known as RES. Clinical approaches, treatment strategies, and MRI features of RES have been explored.

Keywords: Case report, Central nervous system, Cerebellar vermis, Congenital maldevelopment, Rhombencephalosynapsis.

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Background

Cerebellar maldevelopment leading to the fusion of cerebellar hemispheres and hypogenesis or agenesis of the cerebellar vermis is the hallmark of rhombencephalosynapsis (RES), which is a congenital condition.[1] It is also linked to aqueductal stenosis, which causes hydrocephalus, and other CNS problems. Rhombencephalosynapsis rarely manifests as a standalone case but frequently occurs in conjunction with other disorders. Ataxia, body incoordination, dysarthria, hypotonia, seizures, head shaking, and in severe cases, mortality in early adulthood, are some of the symptoms of this condition, which has an unclear etiology and pathogenesis. Recent developments have made it possible to diagnose it prenatally using fetal MRI scans. Rhombencephalosynapsis should be suspected when ventriculomegaly is seen on fetal sonography.[2] Brain MRIs are performed on symptomatic patients if a diagnosis is not made during pregnancy. Supportive care and surgical management for related congenital abnormalities, such as hydrocephalus, are the only available treatments for RES.[3]

Case Presentation

A 52-year-old male was brought to the clinic with a complaint of repetitive and irrelevant speech for 1 month, 10–12 days of headache, balance problems and giddiness, and 2 days of vomiting. He was getting admitted to the ward. He has been on treatment for diabetes and hypertension for 3 years. Under admission, he was presented with the same complaints like vomiting, abnormal speech, and balance/gait problems. There was no other history related to these symptoms or any other disease.

After initial symptomatic treatment, the patient appeared to be in good general health upon inspection. He was aware and had a good sense of place, time, and people. A normal Glass Glow Coma Scale score was obtained after a nervous system assessment. His pupils responded to light and were equal in size and diameter on both sides. Extraocular movements were within normal limits. All four limbs had normal muscle tone and power. All of the limbs with +5/+5 had deep tendon reflexes. The patient showed no symptoms of biological pathology, including stiff neck and facial weakness. Lab examinations like blood examinations, urine examinations, etc. were in the normal range.

We proceeded with an MRI brain detail study and found dilated left lateral and third ventricles, indicative of RES; absence of cerebellar vermis; a fusion of both cerebellar hemispheres and dentate nuclei. The Donald Duck Sign was clearly noted and marked incidentally (Fig. 1). Chronic ischemic changes are also present with age-related cerebral and cerebellar cortical atrophy.

Treatment

Symptomatic treatment for hypertension and diabetes started and other medications for vertigo, and vomiting were also added. After the patient's symptoms subsided and his condition stabilized, he was released from the hospital. For the next 1-year case follow-up is done every month regularly patient has not experienced any worsening or recurrence. Rhombencephalosynapsis was the diagnosis.

Review of Literature and Discussion

In 1914, one autopsy report of a young male was reported as the first case in the presently available literature of RES.[4] A rare congenital

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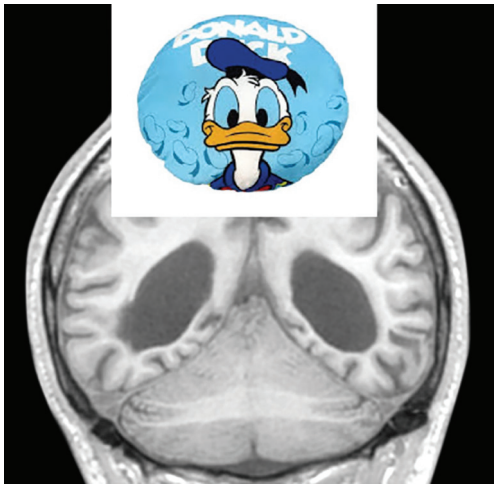

Fig. 1: The absence of the cerebellar vermis, fusion of the dentate nuclei with both cerebellar hemispheres and inflated lateral and third ventricles (The Donald Duck Sign)

deformity of the posterior fossa, RES is typified by a dorsal fusion of the cerebellar hemispheres, a fusion of dentate nuclei and superior cerebellar peduncles, and hypogenesis or agenesis of the vermis. The degree of cerebellar vermis maldevelopment determines the severity of the condition. The condition has no known specific environmental or genetic origins. Although the exact cause of the disease is unknown, several studies have demonstrated that environmental and genetic variables are significant contributors to its etiopathogenesis.

Cerebellar maldevelopment may have resulted from several genetic abnormalities, including tetrasomy 9p and interstitial deletion of the long arm of chromosome 2q.[5]

Based on the available literature, RES can be classified in various ways:

On the Basis of Severity

The degree of hemisphere fusion and the Vermian agenesis pattern are used to categorize RES. The vermis is absent in full RES. The missing portion of the vermis determines the classification of partial RES.

Based on Correlation with Other Conditions

Rhombencephalosynapsis may be solitary or linked to other conditions including atypical holoprosencephaly, VACTERL, and GLHS.[6]

On the Basis of Associated Findings

Based on related results, RES is frequently linked to additional findings such as hydrocephalus, mesencephalon synapsis, holoprosencephaly, pontocerebellar hypoplasia, dysgenesis of the corpus callosum, and absence of the septum pellucidum.[6]

The disease's clinical spectrum ranges from loss of cognition to asymptomatic. The patient often exhibits no symptoms when rhomboencephalosynapsis manifests as an isolated defect. Although the illness typically first appears in early childhood, it can also appear in early infancy if it is severe. Ataxia, hypotonia, epilepsy, spasticity, irregular eye movements, decreased or normal cognitive function depending on severity, dysarthria, the distinctive figure of eight, and side-to-side head shaking are among the most prevalent clinical signs.[7] Atypical craniofacial characteristics such as hypertelorism, a flat midface, a large forehead, and low set posteriorly situated ears are also typically present in the patient. From normal IQ to intellectual impairments, neurocognitive development varies.

Symptomatic patients have a lower life expectancy and die during childhood, while asymptomatic patients lead normal lives with minor balance and coordination problems. Imaging or neuropathological evidence of a hypoplastic single-lobed cerebellum is used to diagnose RES.[8] If fetal ultrasonography shows cerebellar hypoplasia and ventriculomegaly, it can be identified prenatally using fetal MRI. The prognosis is typically worse if the diagnosis is made during pregnancy. The preferred diagnosis method is brain magnetic resonance imaging. Known as the Donald Duck sign, it typically exhibits an aberrant posterior fossa and fourth ventricle that create a diamond or keyhole form.[9] Brain MRI can effectively distinguish RES from the most prevalent differentials for this disorder, which include Joubert syndrome and the Dandy-Walker spectrum.[10] If an MRI is unavailable or not recommended, the diagnosis of rhomboencephalosynapsis can then also be made with a brain CT scan.

Rhombencephalosynapsis does not have a proven cure. In exceptional occurrences, RES does not require medical care. In addition to ventriculostomy or ventricular shunting for obstructive hydrocephalus, the affected infant receives supportive care.[7,8] Patients who receive a diagnosis after birth need physical, social, and psychological care in addition to symptomatic treatment. With physical therapy and rehabilitation, their symptoms progressively become better. Associated abnormalities are managed surgically.

Rhombencephalosynapsis is an uncommon condition that frequently goes undiagnosed, leading to the birth of the affected neonate. To avoid future difficulties for the kid and parents, prenatal counseling for this disease should be promoted.[9,11]

Conclusion

There are numerous theories on the etiopathogenesis of RES, a congenital illness. Due to its comorbidity with other syndromes, the diagnosis is sometimes overlooked. The primary modality for diagnosing RES is still brain MRI. Medication won't do anything because it's a cerebellar maldevelopment disorder. Other related anomalies are managed surgically. Even though new research has revealed its imaging results and clinical characteristics, further gene studies are required to get over the shortcomings of the methods used in genetic studies today. To further understand the etiopathogenesis of the condition, comprehensive prenatal history data is required.

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