Case Report

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Radiology Section

MRI Findings in a Rare Isolated Cerebellar Malformation: Rhombencephalosynapsis

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ABSTRACT

Rhombencephalosynapsis is a rare posterior fossa malformation presenting with absent vermis and fused cerebellar hemispheres. Diagnosis on MRI is straight forward with no differential diagnosis. Herein, we report the pathoanatomic and MR imaging features in a case of symptomatic isolated rhombencephalosynapsis and correlate with the functional abnormality seen clinically.

Keywords: Absent vermis, Ataxic movements, Fused cerebellum, Nystagmus

CASE REPORT

A 14-month-old male infant was referred to our department for a MRI brain. The parents informed that he had abnormal eye movements, involuntary head movements and aggressive behavior in the form of banging head and hyperactivity with lack of normal sleep pattern. Clinical examination revealed motor and mental retardation, nystagmus, and ataxic movements. He was the first child of a second-degree consanguineous marriage born at full term by spontaneous vaginal delivery. There were no history of apnoea or seizures. There were no other dysmorphic features or associated extracranial abnormalities. Personal or familial history was unremarkable.

After obtaining the parent's consent, Multiplanar MRI was performed on 0.35T machine (Magnetom C Siemens, Germany) using MPRAGE, T2W AND FLAIR SEQUENCES in axial, coronal and sagittal planes after sedation. A midline sagittal T2-WI [Table/Fig-1] demonstrated the absence of normal cerebellar vermis. Cerebellar tissue was present in the midline, but it lacked the normal vermian lobular configuration. Coronal MPRAGE [Table/Fig-2] showed midline fused cerebellar lobes.

Cerebellar folia crossed the midline, with no intervening vermis. An axial MPRAGE at the level of the medulla [Table/Fig-3] confirmed fusion of the cerebellar hemispheres across the midline and lack of formation of the vermis as opposed to the normal cerebellar appearance (inset image).

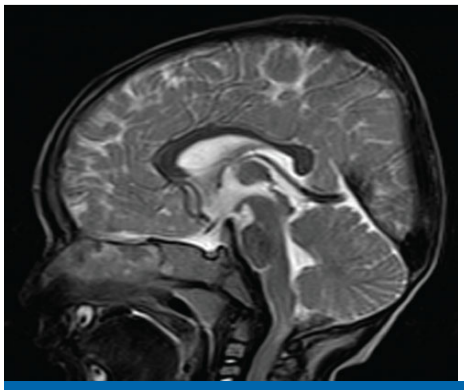
An axial FLAIR -WI [Table/Fig-4] at the level of the superior cerebellar peduncles showed fusion of the cerebellar hemispheres with no definable midline cerebellar vermis. The fourth ventricle was oval pointing posteriorly. Size of the posterior fossa was normal with cerebrospinal fluid filling the subarachnoid space. Corpus callosum and septum pellucidum were preserved, the lateral ventricles were not dilated. Myelination and the gyration appeared normal.

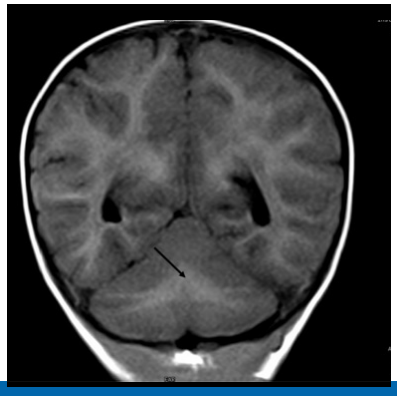
Based on these pathognomic MRI findings, a diagnosis of isolated rhombencepalosynapsis was made without any differential diagnosis. Further workup for metabolic abnormalities and chromosomal aberrations were excluded. No definite treatment was offered except counselling for this disorder.

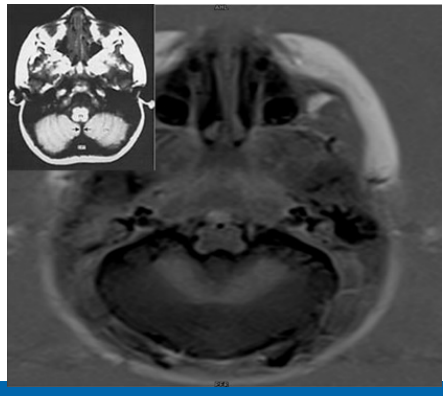
The parents were counselled regarding the varying degrees of cerebellar dysfunction and neurodevelopmental delay. At 4 years follow-up, the child shows features of aggressive behavior with delayed milestones.

DISCUSSION

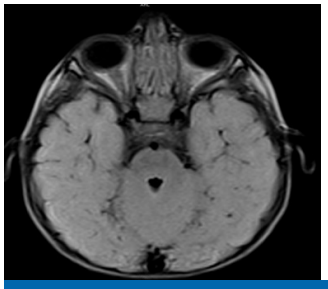
Rhombencepalosynapsis is an unusual disorder of cerebellar development with around 50 cases reported worldwide till now, majority of them recognized on MRI [1,2]. Interestingly, it was, first described by Obersteiner, in 1914 as a postmortem description of a 28-year-old man. It has striking pathoanatomic findings seen as seamless continuation of the cerebellar hemispheres across the midline which presents with cerebellar fusion and absence of cerebellar vermis on imaging studies [1-3]. This is in contrast to other

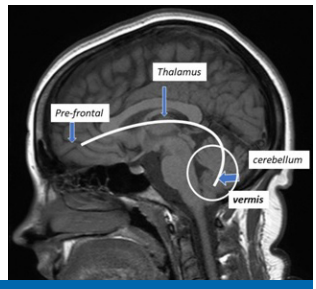






[Table/Fig-1]: A midline sagittal T2-WI demonstrates absence of the normal cerebellar vermis. Cerebellar tissue is present in the midline, but note that it does not have the normal vermian lobular configuration. **[Table/Fig-2]:** A coronal MPRAGE image shows midline fused cerebellar lobes. Cerebellar folia cross the midline, with no intervening vermis (arrow).
[Table/Fig-3]: An axial MPRAGE image at the level of the medulla confirms fusion of the cerebellar hemispheres across the midline and lack of formation of the vermis (inset image showing normal morphology).





[Table/Fig-4]: An axial FLAIR at the level of the superior cerebellar peduncles shows fusion of the cerebellar hemispheres with no definable midline cerebellar vermis. The fourth ventricle is "heart" shape pointing posteriorly instead of normal crescent shape. **[Table/Fig-5]:** Thalamo prefrontal cerebellar circuit.

vermian maldevelopment syndromes (such as Dandy-Walker complex, Joubert syndrome or tectocerebellar dysraphia), where striking structural defects on imaging studies are clefts between the cerebellar hemispheres associated with vermian agenesis and dysgenesis of the isthmus and brain stem creating constellation of imaging findings [1,2].

Disturbed cerebellar development between 28 and 41 days of gestation results in RES. True causative factors regarding genesis of RES are controversial with multiple theories and hypothesis proposed [1-5]. The most plausible hypothesis to explain the fused cerebellum is a primary failure of vermian differentiation possibly due to defective gene expression in early patterning centers of the brain rather than a primary maldevelopment of the vermis. Thus, there would be undivided hemispheres instead of fused ones [1,2].

The most distinctive feature on gross pathology is agenesis or poor differentiation of the vermis, cerebellar hemispheres appear midline fused, with no intervening cyst, folia and fissures are transversely oriented (single-lobed cerebellum).

The striking structural defects of rhombencephalosynapsis are appreciated on imaging studies across all ages including prenatal imaging [1,3,4,6]. The distinctive MRI findings are-

1. A small, narrow fourth ventricle with a "keyhole" or "diamond" shape, instead of the normal crescent shape, is seen on axial sections due to the vermian agenesis and fusion of the dentate nuclei and middle cerebellar peduncles behind the pointed fourth ventricle.

2. Flat and uninterrupted continuity of the white matter of the folia across the midline in the base of the cerebellar hemispheres, transversely oriented folia in the inferior cerebellum represents single-lobed cerebellum [4]. Till date, no case was diagnosed based upon CT-findings solely.

Associated anomalies can be supratentorial as deficiency or absence of the septum pellucidum, dysgenesis of the corpus callosum and the anterior commissure, fused fornices, and fused thalami [1,2,4]. Extracranial anomalies can involve respiratory, genitourinary, musculoskeletal abnormalities [1,3,4].

The severity of neurological dysfunction ranges from mild to severe depending on the associated supratentorial anomalies. There is paucity of literature regarding clinical manifestations and organic correlation in isolated RES [1,4]. Cerebellar vermis is an unpaired midline structure connecting the two hemispheres. It is composed of multiple lobules with characteristic foliar pattern seen on midline sagittal images. Vermis acts as a relay center with sensory inputs from cerebrum, cerebellum, and spinal cord. Functionally, vermis co-ordinates speech, movement of body, eyes in addition to maintaining equilibrium and controlling emotional processes. Anatomical absence/distortions/destructions of vermis gives rise to inappropriate emotional displays in addition to the movement and equilibrium disorders.The cerebellar-thalamo prefrontal circuit [Table/Fig-5] controls inhibition, motor, and executive function and may be disturbed in RES patients due

to an absent vermis.

The child described in this report demonstrated attention deficit disorder, abnormal eye movements, relatively mild motor abnormalities and cognitive impairment. Probably, an impaired cerebellar-thalamo-prefrontal circuit is responsible for the aggressive behaviour. Other studies report similar data [1-4]. Based on the literature data, cognitive functions in RES are mostly impaired [1-5]. In contrast, there are RES patients with normal intellectual development, cognitive and language functions [1,5].

Treatment is none, intellectual impairment and life expectancy are variable, although many affected individuals die in infancy and childhood. There is no differential diagnosis [2-5].

CONCLUSION

RES is a unique entity with pathognomic MRI appearance and no differential diagnosis. Wide spectrum of outcomes emphasizes the importance of precise and complete identification of RES which is essential to prognosticate and counsel the parents.

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