CASE REPORT

RHOMBENCEPHALOSYNAPSIS: A CASE SERIES

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PJR July - September 2023; 33(3): 159-162

ABSTRACT

Rhombencephalosynapsis(RES) is a very rare congenital cerebellar anomaly with limited number of cases reported worldwide. It is characterised by partial or complete agenesis of cerebellar vermis with variable cerebellar hemisphere fusion. Resultant clinical presentation is highly variable ranging from delayed developmental milestones to cognitive and intellectual impairment. This article reports a case series of two child patients presenting with intellectual disability and delayed milestones.

Keywords: Rhombencephalosynapsis, fused cerebellum, intellectual disability.

Introduction

Congenital malformations of posterior cranial fossa are divided into two subtypes on MRI brain, some of them are those with large posterior fossa and others with small or normal sized posterior fossa. Rhombencephalosynapsis corresponds to the later subtype in which there is partial or total agenesis of cerebellar vermis with midline fusion of cerebellar hemispheres.[1] It has pathognomonic appearance on MRI with no other differentials.[2] This anomaly is usually considered as sporadic disease, however, there is some presumption about genetic predisposition.[3] In published data most of the cases were diagnosed as isolated and some of them were associated with other congenital malformations, e.g. VACTERL association or G mez-L pez-HernÆndez syndrome.[4]

Case No. 1

A 9 years old male child with history of developmental delay, delayed speech, delayed walk after 2 year of age and intellectual disability was referred to at Islamabad diagnostic center for MRI brain, which was performed in GE Signa Explorer 1.5 T MRI scanner. MRI documented small sized posterior fossa with fused appearance of cerebellar peduncles, dentate nuclei and vermis in midline with slightly low lying cerebellar tonsils reaching the level of C1 vertebra. Absent rostrum of corpus callosum and anterior commissure along with mild hydrocephalus was also noted. (Fig.1,2 & 3)

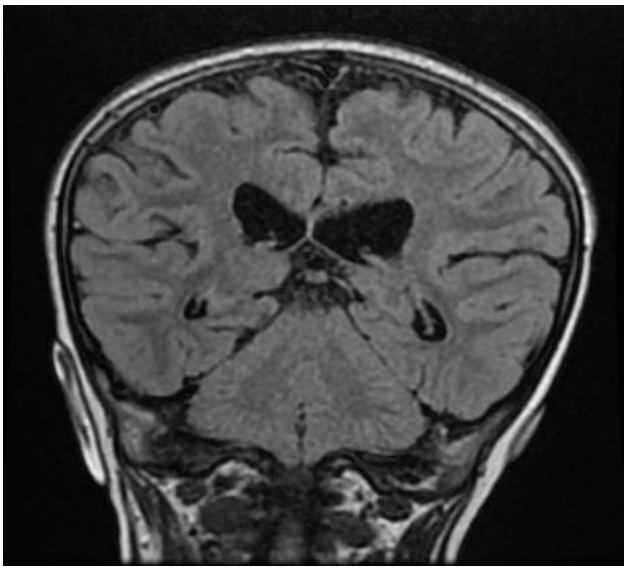

Figure 1: Coronal FLAIR sequence shows midline fused cerebellar hemispheres, peduncles and vermis.

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Submitted 23 August 2023, Accepted 20 September 2023

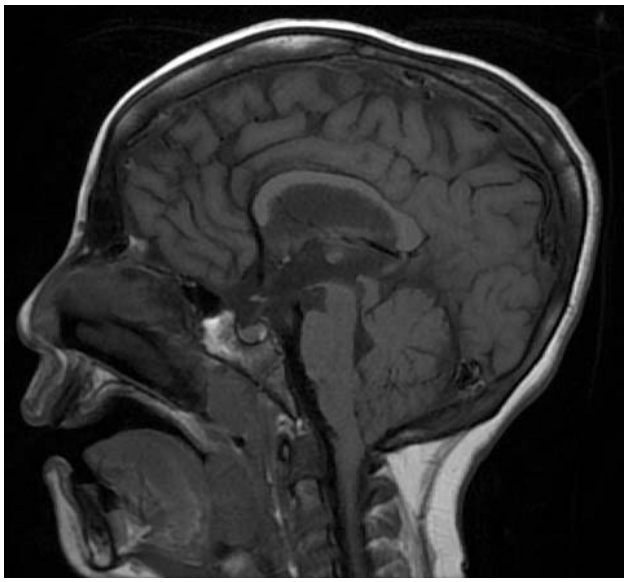

Figure 2: Sagittal T1 sequence shows slightly low lying cerebellar tonsils and absent rostrum of corpus callosum.

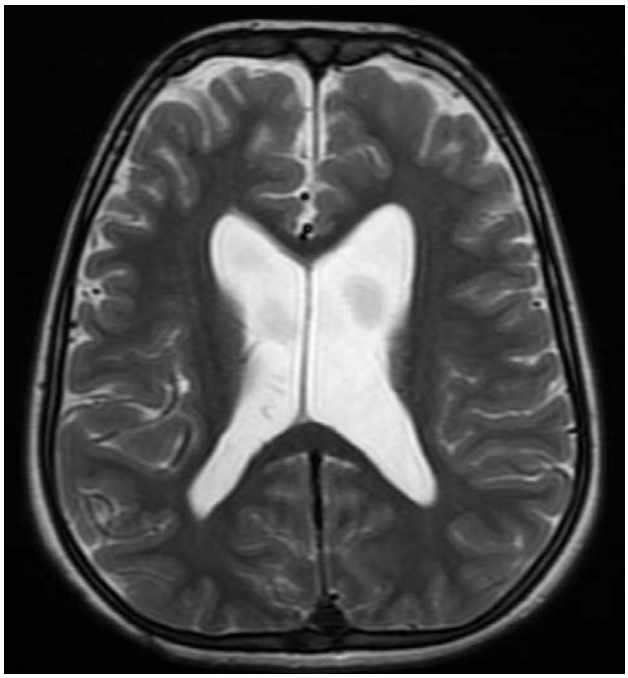

Figure 3: Axial T2 sequence shows mild hydrocephalus

Case No. 2

A 10 year old male child with history of delayed milestones and intellectual disability, abnormal head shape and frequent falls since the age of 3 years was referred to Islamabad diagnostic center for MRI brain, which was performed on GE Signa Explorer 1.5 T MRI scanner. MRI documented abnormal skull shape with flattened appearance of occiput. Posterior fossa structures were abnormal with fused bilateral cerebellar hemispheres with dentate nuclei and cerebellar peduncles in midline. Cerebellar tonsils were also irregular and dysmorphic in shape. The posterior part of falx cerebri appears deficient with interdigitation of cerebral sulci and gyri in midline in the region of bilateral occipital lobes. Tentorium was not clearly visualized.(Fig.4 & 5).

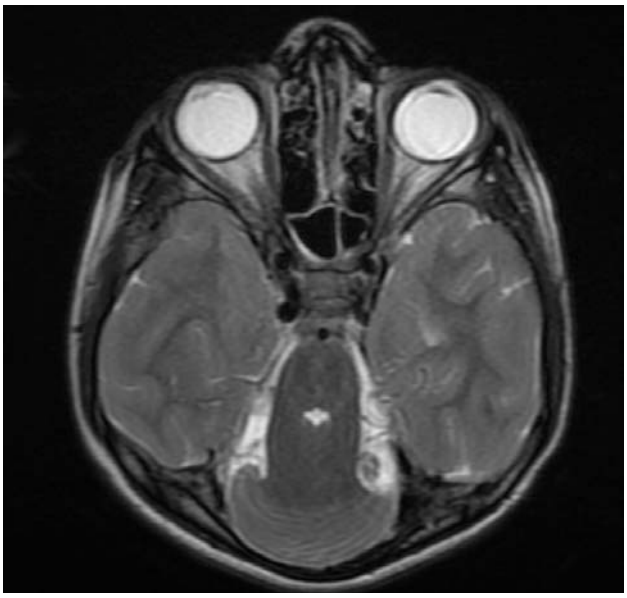

Figure 4: Axial T2 sequence shows flat and uninterrupted continuity of the white matter of cerebellar folia across midline at base of cerebellar hemispheres visible as single lobed cerebellum with transversely oriented folia.

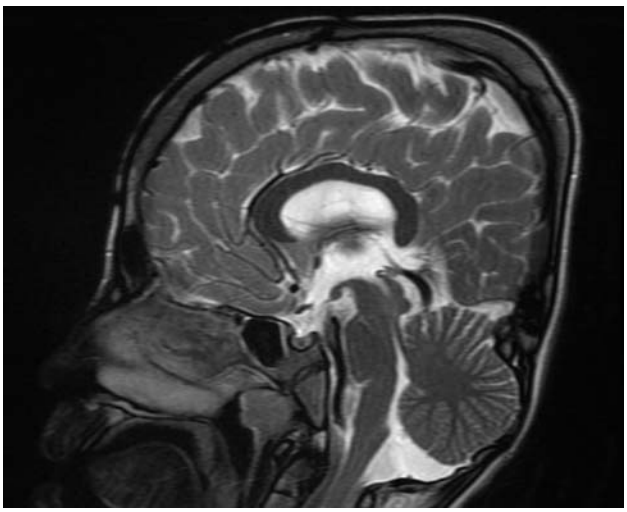

Figure 5: Saggital T2 sequence shows irregular and dysmorphic cerebellar tonsils. Tentorium cerebelliis not clearly visible.

Discussion

Very few case reports of Rhombencephalosynapsis(RES) are available indicating that it is a rare disease. Its aetiology is not exactly understood, however, it is considered a sporadic disorder.[5] Despite of many efforts on stereotyping, no genetic association has been diagnosed.

RES can be diagnosed in antenatal life by ultrasound performed by an expert and is usually detected by presence of ventriculomegaly.[6] In the traditional concept, rhombencephalosynapsis was considered to be result of abnormal development of the vermis with subsequent fusion of the hemispheres.[7] However, some authors suggested that this anomaly is more likely due to a failure of vermian differentiation with undivided development of hemispheres.

There are still many doubts about the exact disorder of development.[8] Other associated congenital anomalies can vary from patient to patient. Most commonly associated intracranial anomalies include hydrocephalus or ventriculomegaly, corpus callosum dysgenesis, and the absence of septum pellucidum. Relatively less frequent and occasionally associated anomalies include agenesis of the posterior lobe of pituitary, thalamus, tectum and fornices fusion, hypoplasia of the temporal lobes, olivary nuclei, anterior commissure and optic chiasm. RES in rare cases has co-existing extracranial anomalies as well.[9]

In our study we reported case series of rhombencephalosynapsis in two patients of nearly similar age group (9-10 years) with clinical symptoms of intellectual disabilities and delayed milestones. In our patients no associated other extra cranial anomalies were detected.

Conclusion

Rhomboencephalosynapsis is a rare brain disorder with unknown aetiology. It can be diagnosed with MRI brain and should be considered as a potential diagnosis when children present with clinical symptoms of intellectual disabilities and delayed milestones. More research work is required to know associated developmental defects and syndromes for better understanding of disease.

Conflict of Interest: None

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