Scholars Journal of Medical Case Reports (SJMCR) **ISSN 2347-6559 (Online)**
Abbreviated Key Title: Sch. J. Med. Case Rep. **ISSN 2347-9507 (Print)**

A United of Scholars Academic and Scientific Society, India

A Rhombencephalosynapsis Case Recognized in Adulthood without Any Neurological Findings

Prof. Dr. Kemal ARDA[1*], Dr. Sinan AKAY[2], Dr. Guray KOC[3]
[1-2]Dept. of Radiology, [3]Dept. of Neurology, Gulhane Research and Education Hospital, SB University, Bozyaka, İzmir, Turkey

***Corresponding author**
Dr. Kemal ARDA

Article History
Received: 13.04.2018
Accepted: 19.04.2018
Published:30.04.2018

DOI:
10.36347/sjmcr.2018.v06i04.017

Abstract: Rhombencephalosynapsis is reported as one of the rarest congenital anomalies of posterior fossa and characterized by absent, or hypoplastic cerebellar vermis and fusion of the cerebellar hemispheres. This entity may be seen in isolation or with other anomalies. MR is the main diagnostic tool. Typical clinical manifestation mostly includes various neurological dysfunctions, ranges from mild to severe. However, in contrary to the established information in the literature, in the light of our case, we believe that Rhombencephalosynapsis may be more often than predicted in normal appearing population. We present a Rhombencephalosynapsis case who has normal daily activities without any neurological dysfunction.
Keywords: Rhombencephalosynapsis, magnetic resonance imaging, congenital anomaly, cerebellum, neurologic dysfunction.

INTRODUCTION

Rhombencephalosynapsis (RES) is known as one of the rare congenital anomalies of the posterior fossa. It was first reported by Obersteiner in 1914, and named by Gross and Hoff in 1959 [1-4]. Although the cause of RES still remains unrevealed, it is characterized by cerebellar vermis hypogenesis or agenesis and fusion of the cerebellar hemispheres, fusion of the dentate nuclei and superior cerebellar peduncles [1-6].

RES can be seen either alone or together with complex malformations [1-3, 6]. Additionally, some other central nervous system and/or extra-nervous system anomalies, can also be associated with RES [5, 6]. Amongst these, hydrocephalus, ventriculomegaly, hypoplasia of the temporal lobes, absent/rudimentary/dysgenic corpus callosum, absence of the septum pellucidum, fused thalami, tectum and fornicles, olivary nuclei, anterior commissure and optic chiasma, holoprosencephaly, cortical dysplasia, agenesis of the posterior lobe of the pituitary, hippocampal hypoplasia, large clefts supratentorially, multiple synosthoses of cranial sutures are the most commonly seen anomalies [2, 5]. Clinical indications of RES generally include supratentorial midlline anomalies including muscular hypotonia, spasticity, truncal and/or limb ataxia, abnormal eye movements, strabismus, dysarthria, head stereotypies (head rolling), and developmental delay. Related neurological dysfunction is reported to range from mild to severe [2].

Magnetic Resonance (MR) imaging is the main diagnostic modality of RES that can readily provide the most common posterior fossa imaging demonstrating horizontally orientated foliae and fissures, which extend across the midline with absent or poorly developed vermis and the fusion of the cerebellar hemispheres. It may also show other associated intracranial abnormalities such as absence of septum pellucidum, hydrocephalus, thalamic fusion, limbic system anomalies, and cranial synostosis [4]. Fusion of the cerebellar hemispheres that forms the characteristic continuous cerebellar folial shape can be detected especially on axial and coronal images. Flat-based cerebellum without cerebellar vallecula and diamond-shaped fourth ventricle are other common indicators of RES on MR images [1]. In addition, in patients with RES and hydrocephalus, the posterior part of corpus callosum is prominantly thin and dorsally deviated [6].

In this case report, we aim to present the MR imaging findings of a RES case recognized in adulthood without any neurologic-cognitive pathology and development delay or problem.

CASE REPORT

A 47-year-old male patient was admitted to Neurology Department of our hospital with the complaint of 2-year history of discontinuous headache. It was a dull pain with the pressure around the forehead and not associated with nausea or vomiting. The frequency of headache was once or twice a month, the severity of headache was fluctuated, but not progressively worsened over time. General and

neurological examination were completely normal. His past medical history was normal. He is employed, he performs normal daily activities.

A brain MR imaging was recommended by patient's consultant. His MR imaging scan revealed fusion of cerebellar hemispheres, fused transverse cerebellar folia and partial hypogenesis of vermis. Cerebellum itself was in normal size and slightly reduced in transverse diameter, not really hypoplastic. Additionally, there was indistinct primary fissure of cerebellum. Examination also demonstrated that isthmus and splenium parts of corpus callosum were thin, the lateral ventricles were dilated and that the gyration appeared normal. The inferior olivary nuclei were present in the MR imaging of the medulla oblongata (Figure 1-3). Based on the MR imaging findings described above, the patient was recommended further diognostic tests for possible metabolic abnormalities and chromosomal rearrangements, which came out to be normal.

Here, radiological diagnosis of a suspected RES case was made. The clinical features of RES was not relevant to this diagnosis, but rather considered as a tension-type headache. A non-steroidal anti-inflammatory drug was initiated with consultation to neurosurgery due to hydrocephalus, and follow-up was planned.

FIGURE LEGENDS

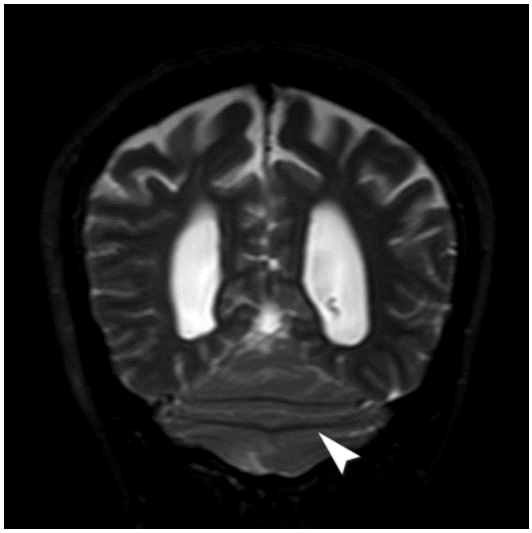

Fig-1: Coronal T2-weighted image shows fused cerebellar hemispheres and transverse cerebellar folia (arrowhead). Dilatation of bilateral lateral ventricles is also seen

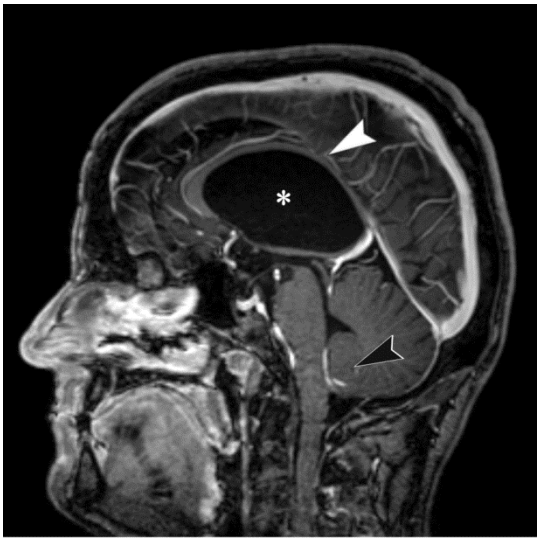

Fig-2: On sagittal postcontrast 3-dimentional T1-weighted image, thinning in isthmus and splenium of corpus callosum (white arrowhead), dilated lateral ventricle (asterisk), and tonsillar hypoplasia (black arrowhead) are readily seen

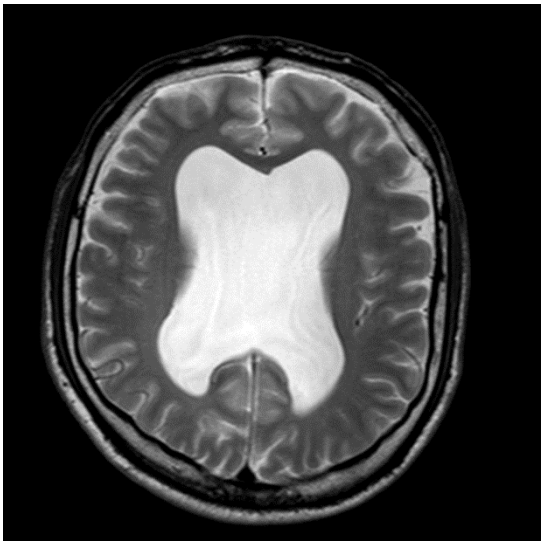

Fig-3: Axial T2-weighted image demonstrates dilated lateral ventricles and absence of septum pellicidum. Gyration is normal in brain

DISCUSSION

Although the etiology of RES is still unknown, it is believed that this congenital anomaly stems from a developmental abnormality of the cerebellar vermis around 3-6 weeks of gestation [2-5]. Literature review revealed that RES related studies are mainly on case presentations or case series involving patients with fetal or pediatric age group. Recently, Bell *et al*. [7] reported an employed male diagnosed with RES according to his MR imaging findings at the age of 55. In this case, the neuropsychological examination was in normal limits except subtle sensory-motor abnormalities, poor immediate visual memory and motor dexterity. In another representative case of a 39-year-old employed female with a history of chronic pain, Guyot *et al.* [8] reported that the patient was diagnosed with RES. The neurological examination was within normal limits except subtle ataxia of tandem gait. The patient had no remarkable medical, family, or social history. Her cognition level was average. Verri *et al*. [9] also studied an RES case of a laborer diagnosed at the age of 22. However, this patient had borderline impaired IQ and a psychiatric history consisting obsessive oral self-mutilation different from our case and the other adult RES cases described in literature. Our findings from MR imaging provided suffiecient evidence for RES diagnosis. Neurological examination was completely normal. He is employed, he performs normal daily activities. He had no remarkable medical, family, or social history.

Published reports show that the diagnosis of RES is accompanied by a neurological disorder, i.e., no case with completely normal neurological examination. This is because patients having RES can perform their daily activities with no signs of anomalies. We would like to note here that the number of incidents with RES could be much higher than what is reported in literature if patients with normal neurological examination are to be scanned for MR imaging.

CONCLUSION

Our case is a typical example of MR imaging, where RES could be diagnosed even in patients with unspecified developmental delay. Normal cognitive functions do not exclude the diagnosis either. We would like to make the clinicians aware that if the symptoms are mild, the diagnosis is usually made in adulthood. Non-syndromic RES may be even asymptomatic and that is why the diagnosis can be easily missed.

Competing interests

Authors have no competing interests.

REFERENCES

1. Bekiesińska-Figatowska M, Jurkiewicz E, Szkudlińska-Pawlak S, Malczyk K, Nowak K. Rhombencephalosynapsis-isolated anomaly or complex malformation? Pol J Radiol. 2012;77: 35-8.
2. Paprocka J, Jamroz E, Ścieszka E, Kluczewska E. Isolated rhomboencephalosynapsis–a rare cerebellar anomaly. Polish journal of radiology. 2012 Jan;77(1):47.
3. Choudhary A, Minocha P, Sitaraman S. Gomez-Lopez-Hernández syndrome: First reported case from the Indian subcontinent. Intractable Rare Dis Res. 2017;6: 58-60.
4. Guntur Ramkumar P, Kanodia AK, Ananthakrishnan G, Roberts R. Chiari II malformation mimicking partial rhombencephalosynapsis? A case report. Cerebellum.2010;9:111-4.
5. Shahrzad M, Gold M. Rhombencephalosynapsis: a rare congenital anomaly presenting with seizure and developmental delay. Acta Neurol Belg. 2015;115: 685-6.

6. Ishak GE, Dempsey JC, Shaw DW, Tully H, Adam MP, Sanchez-Lara PA, Glass I, Rue TC, Millen KJ, Dobyns WB, Doherty D. Rhombencephalosynapsis: a hindbrain malformation associated with incomplete separation of midbrain and forebrain, hydrocephalus and a broad spectrum of severity. Brain. 2012 Mar 26;135(5):1370-86.
7. Bell BD, Stanko HA, Levine RL. Normal IQ in a 55-year-old with newly diagnosed rhombencephalosynapsis. Arch Clin Neuropsychol. 2005;20: 613-21.
8. Guyot LL, Kazmierczak CD, Michael DB. Adult rhombencephalosynapsis. Case report. J Neurosurg. 2000;93: 323-5.
9. Verri A, Uggetti C, Vallero E, Ceroni M, Federico A. Oral self-mutilation in a patient with rhombencephalosynapsys. J Intellect Disabil Res. 2000;44: 86-90.