

Asymptomatic Dandy-Walker Variant in adulthood: A Case Report and Review of the Literature

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Abstract

Background: Brain and body magnetic resonance imaging (MRI) is increasingly utilized for both clinical and commercial screenings, making it inevitable to detect incidental findings that are oftentimes unrelated to the purpose of the imaging. These cases pose a clinical challenge due to a lack of established protocols for their follow-up and management.

Case description: A 43-year-old asymptomatic male patient presented to our neurology clinic with an incidental brain MRI lesion concordant with an imagenological diagnosis of asymptomatic Dandy Walker Malformation variant (DWMv). The patient had undergone a voluntary full-body MRI after suffering from a sport's related ankle injury.

Conclusion: This case report represents one of the few reported in medical literature. Herein, we aim to elucidate the need for a consensus on established guidelines and clinical recommendations for the appropriate management of incidental findings that can be associated with life-threatening clinical scenarios.

Keywords: Dandy-walker syndrome; Dandy-walker variant; adult; nervous system malformations; hydrocephalus; incidental finding

Introduction

Dandy-Walker syndrome (DWS) is a rare posterior fossa abnormality disorder first described in 1987 by Sutton and finally declared as a Dandy Walker syndrome by Benda in 1954. It is mainly characterized by hypoplasia of the vermis, cysts that communicates with the fourth ventricle and it is usually diagnosed intrauterine [1, 2]. Although the signs and symptoms in DWS are nonspecific, patients can present with developmental delay, hypotonia or spasticity, ataxia, or augmented head circumference. According to the National Organization for Rare Disorders in the US it affects about 1 per 30,000 live births and according to the gender is more frequent in females than males [3]. In the United States racial, gender, and age analyses indicate that an increased age and elective admission decreases mortality and the incidence of adverse discharge deposition (ADD) in association with DWS [4]. The spectrum of each presentation includes multiple factors including intracra-

nial hypertension and hydrocephalus [5]. The majority of the diagnoses are made within the first year of life, with macrocephaly being the most common clinical manifestation [4, 5]. Furthermore, the new onset diagnosis in the adult population can include rare events such as: brainstem infarction, psychosis, neuromuscular disease, ischemic stroke, and hemorrhagic stroke of the basal ganglia [6]. Given the aforementioned, herein we present an asymptomatic case of DWS in adulthood as one of the few cases described in the medical literature and its importance to further our understanding of management protocols for asymptomatic DWS and its variants.

Case Presentation

A 43-year-old asymptomatic male patient was referred to our neurology clinic due to incidental abnormal findings on brain magnetic resonance imaging (MRI) examination. **(Figure 1)** The patient refers that the MRI was routinely conducted

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following a fall injury while playing tennis, denying any direct head injuries or loss of consciousness (LOC) resulting from the fall itself. Patient refers to solely suffering a minor left ankle injury and voluntarily decided to undergo a whole-body MRI.

Initial MRI examination revealed a large extra-axial cerebrospinal fluid (CSF) collection in the posterior aspect of the posterior cranial fossa measuring up to 7 cm wide x 4.2 cm antero-posterior (AP) x 6.2 cm superior to inferior, consistent with a diagnosis of mega cisterna magna versus an arachnoid cyst. There were no acute abnormalities in the brain or intracranial hematomas reported. Thus, the radiological diagnosis of asymptomatic Dandy Walker malformation variant (DWMv) was established. Patient's perinatal history includes being born full term through assisted vaginal breech delivery. Patient received full immunization throughout childhood and adulthood. Furthermore, there were no identifiable medical history of neurocognitive impairments or learning deficits. Patient denied medication use, medical, surgical or hospitalization history.

A complete neurological evaluation was conducted and was unremarkable other than hyporeflexia on lower extremities, bilaterally. The Rinne test was unremarkable for whereas the Weber test demonstrated left sided lateralization. Cognitive evaluation was conducted using MoCA and SLUMS questionnaires in which scores were unremarkable for cognitive impairment. Due to the unremarkable clinical findings, a 6 month follow-up was recommended.

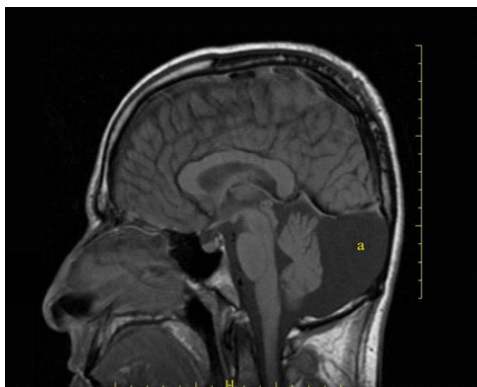


Figure 1: T1-weighter midsagittal MR imaging plane of the brain without contrast. (a) Large extra-axial CSF collection in the posterior aspect of the posterior cranial fossa consistent with Dandy Walker malformation variant. MR: magnetic resonance, CSF: cerebrospinal fluid

Discussion

Most cases of DWS are described as sporadic, although there are several literature references of chromosomal abnormalities that can be associated with teratogens, chromosomopathies, fetal alcohol exposure and congenital rubella [7]. Deletion of 3p24.3 with ZIC1 and ZIC 4 genes are amongst the first described DWM genes. Other chromosomal abnormalities include deletion or duplication of 6p25.3, tetrasomy 9p, deletion of 13q32.2 or q33.2, deletion of 2q36.1 (the location of the gene PAX3), deletion and duplication of 7p21.3 (NDUFA4 and PDF 14 gene location) [4, 8]. Additionally, isolated DWM has also been reported amongst siblings, suggesting an autosomal recessive or X-linked inheritance pattern [9]. Although

no genetic testing was carried out for this particular case, given the lack of positive family history we determined that this case was associated with a sporadic form of DWS.

Imaging findings in DWM

According to Gumz Correa, G. et al., the diagnosis can be done intra-uterus as early as 14 weeks of gestation to differentiate from other cystic posterior fossa lesions, with the most commonly used imaging modality being ultrasonography (US) [8]. The use of MRI has also been discussed in the prenatal diagnosis, which may aid in the visualization of the position of the torcular, an important finding in the DWM syndrome [10]. In adult patients, diagnosis through MRI over CT scan has an advantage with added multiplanar reconstruction to further analyze characteristic lesions on the posterior fossa [8, 11]. Given that this incidental finding was made through an unrelated and voluntary full-body MRI scan, no further imaging was required.

Differential Diagnosis

Secondary Arachnoid Cyst

The first description of an arachnoid cyst was made by Bright in 1831 [12]. Arachnoid cysts represent approximately 1 % of all atraumatic expansive lesions [13]. As part of the differential diagnosis, one of the most common diagnosis include the retro-cerebellar arachnoid cyst [6]. MRI findings are consistent with cystic intrasellar lesion with suprasellar extension that are balloon-shaped, regular, molding with no invasion of the medial wall of cavernous sinus [13]. Furthermore, they can be large enough to cause compression of the cerebellar hemisphere and the fourth ventricle which can also be associated to DWS as already described above [8].

Hydrocephalus

Hydrocephalus is a common finding on patients with DWS but can also be a differential diagnosis due to the similarity of the posterior fossa abnormalities that can coexist in both DWS and hydrocephalus [8]. More commonly, hydrocephalus is described as a clinical entity which is characterized by three factors: increased cerebrospinal fluid volume, increased intracranial pressure and dilatation of the cerebrospinal fluids spaces [10].

Megacisterna magna

Megacisterna Magna was first described by Richard Gonsette in 1962, this description was made in patients who presented with cerebellar atrophy [4]. Mega cisterna magna is an enlarged cisterna magna that can measure up to 10 mm or more from the posterior aspect of the vermis up to the internal aspect of the skull's vault. There are no identifiable structures abnormalities either infra or supratentorially [14]. Mega Cisterna magna is the most common differential diagnosis, however, in this pathology the fluid collection is found or located posteroinferior to a normally and well developed cerebellum [15].

Conclusion

Incidental brain MRI findings of DWMv in asymptomatic adult patients are rare. The consequences and implications of these findings are yet to be described in the literature, making it hard to appropriate treatment and management guidelines. Furthermore, given the rarity of asymptomatic DWMv in adulthood, there are no established protocols that optimize the clarity of the final diagnosis or the potential complications, for which the individualized management heavily relies on the clinician's expert opinion.

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References

1. Trevino Alanis MG, Gonzalez Cantu N, Montes Cruz JV, Garcia Flores JB, Martinez Menchaca HR, Rivera Silva G. [Dandy Walker malformation]. *Arch Argent Pediatr*. 2014; 112(1):103-104.
2. Cardoso J, Lange MC, Lorenzoni PJ, Scola RH, Werneck LC. Dandy-Walker syndrome in adult mimicking myasthenia gravis. *Arq Neuropsiquiatr*. 2007; 65(1):173-175.
3. Kalidasan V, Carroll T, Allcutt D, Fitzgerald RJ. The Dandy-Walker syndrome--a 10-year experience of its management and outcome. *Eur J Pediatr Surg*. 1995; 5(1):16-18.
4. McClelland S, 3rd, Ukwuoma OI, Lunos S, Okuyemi KS. Mortality of Dandy-Walker syndrome in the United States: Analysis by race, gender, and insurance status. *J Neurosci Rural Pract*. 2015; 6(2):182-185.
5. U.S. Department of Health and Human Services Dandy-Walker Syndrome Information Page. <https://www.ninds.nih.gov/Disorders/All-Disorders/Dandy-Walker-Syndrome-Information-Page>. Accessed Feb 26, 2022.
6. Abdul H, Burns J, Estevez A, Nasr El-Nimer C, Ekinde B, Lacaille S. Hemorrhagic Stroke in a Young Adult with Undiagnosed Asymptomatic Dandy-Walker Malformation. *Case Rep Neurol Med*. 2019; 2019:1450703.
7. Robinson AJ. Inferior vermian hypoplasia--preconception, misconception. *Ultrasound Obstet Gynecol*. 2014; 43(2):123-136.
8. Correa GG, Amaral LF, Vedolin LM. Neuroimaging of Dandy-Walker malformation: new concepts. *Top Magn Reson Imaging*. 2011; 22(6):303-312.
9. Bragg TW, St George EJ, Wynne-Jones GA, Hockley A, Morton JE. Familial Dandy-Walker syndrome: a case report supporting an autosomal inheritance. *Childs Nerv Syst*. 2006; 22(5):539-541.
10. Bernardo S, Vinci V, Saldari M, et al. Dandy-Walker Malformation: is the 'tail sign' the key sign? *Prenat Diagn*. 2015; 35(13):1358-1364.
11. Gibson LM, Paul L, Chappell FM, et al. Potentially serious incidental findings on brain and body magnetic resonance imaging of apparently asymptomatic adults: systematic review and meta-analysis. *BMJ*. 2018; 363:k4577.
12. Jafrani R, Raskin JS, Kaufman A, Lam S. Intracranial arachnoid cysts: Pediatric neurosurgery update. *Surg Neurol Int*. 2019; 10:15.
13. Holanda Ferreira TS, Malveira LRC, Gomes Neto A. Spontaneous subdural and intracavitary hemorrhage of temporal arachnoid cyst in an adult patient. *Interdisciplinary Neurosurgery*. 2021; 23.
14. Milani HJF, Barreto EQS, Ximenes R, Baldo CAR, Araujo Junior E, Moron AF. Fetal posterior fossa malformations: review of the current knowledge. *Radiol Bras*. 2019; 52(6):380-386.
15. Barkovich AJ, Kjos BO, Norman D, Edwards MS. Revised classification of posterior fossa cysts and cystlike malformations based on the results of multiplanar MR imaging. *AJR Am J Roentgenol*. 1989; 153(6):1289-1300.