A Case Report of Mega Cisterna Magna with Extra-cranial Features of Dandy-Walker Malformation in an Adult: Implications for the Dandy-Walker Complex Continuum

Eugene John F. Balmores Jr., MD[1,2]

[1]*College of Medicine, Isabela State University, Echague Campus, Echague, Isabela, Philippines*
[2]*Department of Family and Community Medicine, Cagayan Valley Medical Center, Cagayan, Philippines*

ABSTRACT

Posterior fossa malformations are rare cyst-like pathologies of the central nervous system. Outcomes of patients with these conditions are largely documented in fetal or early childhood studies as most cases are non-compatible with life. Also, different schemes to categorize these occurrences have been proposed. One classification puts forth that the entities are a continuum of pathologies, called the Dandy Walker Complex, wherein Mega Cisterna Magna is the most benign to Dandy Walker Malformation as the most severe form. We report the first case of a patient with a mixed presentation of Mega Cisterna Magna having extracranial manifestations of Dandy Walker Malformation reaching her adult years. The patient is a 26-year-old female who was apparently well until she presented with recurrent headache and seizure episodes of 2-year duration. She had an unremarkable birth and childhood history, apart from learning difficulties in school. In her adult years, she gave birth to an infant with multiple physical anomalies. She has a maternal uncle with abnormal facie and intellectual disability. Physical examination of the patient exhibited a bulging occiput, hypertelorism, down-slanting palpebral fissures, large globular nose, large ear lobules, high arched palate, and clinodactyly. Neurologic examination was unremarkable. Magnetic resonance imaging confirmed Mega Cisterna Magna and was deemed non-surgical. She remained seizure-free during admission as well as on follow-up, maintained on Phenytoin. The existence of a Dandy-Walker continuum is still debated, as the link among the disease entities are yet to be established using developmental or genetic studies. This case, however, supports the Dandy-Walker Complex classification by demonstrating a rare combination of Mega Cisterna Magna with features of Dandy-Walker Malformation in an adult. This can contribute to disease definition and eventually to the discovery of the pathobiological mechanisms of posterior fossa cysts, and to appropriate diagnosis and management.

Keywords: Mega cisterna magna, Dandy-Walker Malformation, Dandy Walker Complex, rare disease, case report

INTRODUCTION

Posterior fossa malformations are rare cyst-like intracranial pathologies which are believed to arise from the maldevelopment of the hindbrain.[1] The variety includes Dandy-Walker Malformation (DWM), Dandy-Walker Variant (DWV), and Mega Cisterna Magna (MCM); the respective prevalence of each is 6.70 per 100,000, 2.08 per 100,000, 54% of all cystic posterior fossa malformations, and with overall prevalence of 1 per 35,000.[2-5] These conditions presenting in the adult years are extremely rare.[6]

DWM is a complex malformation that is usually diagnosed on second-trimester ultrasound examination and is defined by (1) partial or complete agenesis of cerebellar vermis, (2) enlargement of the posterior fossa with the upward displacement of the tentorium, transverse sinus and torcular, and (3) cystic dilation of the fourth ventricle.[7] MCM,

eISSN 2094-9278 (Online)
Published: December 18, 2025
https://doi.org/10.47895/amp.vi0.12682

Corresponding author: Eugene John F. Balmores Jr., MD
College of Medicine
Isabela State University - Echague Campus
San Fabian, Echague, Isabela, Philippines
Email: ejbalmoresmd@gmail.com
ORCiD: https://orcid.org/0000-0002-4053-9110

meanwhile, has been considered a normal variant and is defined as an enlargement of the cisterna magna greater than 10 mm on imaging without vermian hypoplasia and fourth ventricle anomaly. Whereas DWM presents with extra-cranial syndromic features of the face, extremities, heart, and urinary system, most children with isolated MCM develop normally.[8-10] Few studies, however, have documented non-isolated MCM with associated extra-cranial manifestations (i.e., talipes, renal agenesis, pectus excavatum, heart defect, etc.). Follow-up of these patients revealed poor prognosis, with outcomes including fetal demise and termination of the pregnancy.[11-13] Moreover, developmental studies on patients with MCM share a consensus of developmental delay occurring in non-isolated MCM cases.[14,15]

Over the years, a number of perspectives on the differentiation of posterior fossa anomalies have emerged. Barkovich et al. described a classification based on multi-planar magnetic resonance imaging and suggested that the abnormalities represent a continuum called the Dandy-Walker Complex (DWC).[16] However, differences in the cerebellar vermis and fourth ventricle anatomy of the entities led some to believe that they cannot be grouped together under a continuum. A link among the members has not been demonstrated.

In this case report, we demonstrate a rare occurrence of a patient with non-isolated MCM having multiple extra-cranial features of DWM reaching her adult years. The importance of studying the genetics of syndromic cases and benign cases for diseases in a continuum is likewise presented.

CASE PRESENTATION

Patient History

This is a case of a 26-year-old female, married, Filipino, who consulted due to visions of flickering lights.

The patient was apparently well, until approximately two years prior to consult (PTC), patient had her first episode of headache, described as throbbing, occurring without a trigger, persistent, occipital in location, non-radiating, with a pain scale of 5/10, lasting for a few minutes, aggravated with exertion, and relieved spontaneously. This was associated with nausea and vision of flickering lights. There was no associated head or neck trauma, substance use, fever, vomiting, blurring or loss of vision, photo or phonophobia, extremity numbness or weakness, loss of balance, and altered mental state. In the interim, the headache persisted with the same quality and quantity, occurring once every two months. No medications were taken; no consultation was done.

Approximately one year PTC, the patient, while at work, had a witnessed episode of loss of consciousness. She shouted loudly, fell down, and began to have stiffening extremities followed by jerking movements, with upward rolling of eyeballs. This episode lasted for a few seconds. The patient urinated and was confused after the event. Patient still had no consultation at this time. The episodes then occurred once every two months with the same character and duration, and without particular triggers. Later on, the episodes took place four times in the past month, with the last occurring five days PTC.

Waxing and waning of the headache with photopsia for the past two years and more frequent episodes of tonic-clonic movements for the past month urged consultation at our hospital for evaluation.

Obstetric history revealed that the patient is a G1P1 (1000). She gave birth in 2016 to a term male infant via normal spontaneous delivery, but the child died on the third day of life. The child had findings of underweight, cleft palate, rocker-bottom feet, and polydactyly, and was diagnosed with Potter's Syndrome. Family history revealed that the patient had a maternal uncle with the same physical features as hers and with intellectual disability.

Patient Physical Examination

At the time of consult, physical examination revealed the patient having stable vital signs, with physical findings of a bulging occiput, hypertelorism, down-slanting palpebral fissures, large globular nose, large ear lobules, high arched palate, and clinodactyly (Figure 1). Neurologic examination was unremarkable, apart from the patient having child-like facial features.

Investigations and Diagnosis

The working diagnoses at the initial consult were: to consider a Neurogenetic Syndrome comprising of (a) Seizure Disorder probably secondary to a brain malformation and (b) Neurodevelopmental Delay, probably Intellectual Disability; to rule out electrolyte imbalance, hypoglycemia, and uremia. The physician-in-charge then facilitated a non-urgent evaluation of the seizure episode: cranial computed tomography (CT) scan, electroencephalography (EEG), capillary blood glucose, blood urea nitrogen, creatinine, sodium, and potassium were requested. A determination of mental age was also requested.

Chemistry panel results were within normal range, aside from a finding of mild anemia (Hgb = 110 g/L). CT scan revealed a moderate sized CSF fluid structure in the left retrocerebellar region with associated mild compression of the adjacent bilateral cerebellum, with an impression of retrocerebellar arachnoid cyst versus mega cisterna magna (Figure 2A). EEG revealed no focal epileptiform discharge. Mental age determination revealed an age of 21 years old, as opposed to the chronological age of the patient at 26 years.

Since the CT scan revealed a new brain mass with findings of mild compression, the patient was referred by the physician-in-charge to Neurology service, who then advised hospital admission; Neurosurgery service meanwhile noted no surgical intervention indicated at the time of examination. Discussions regarding the impression and management were facilitated by the physician-in-charge during the time that the patient was in the emergency room.

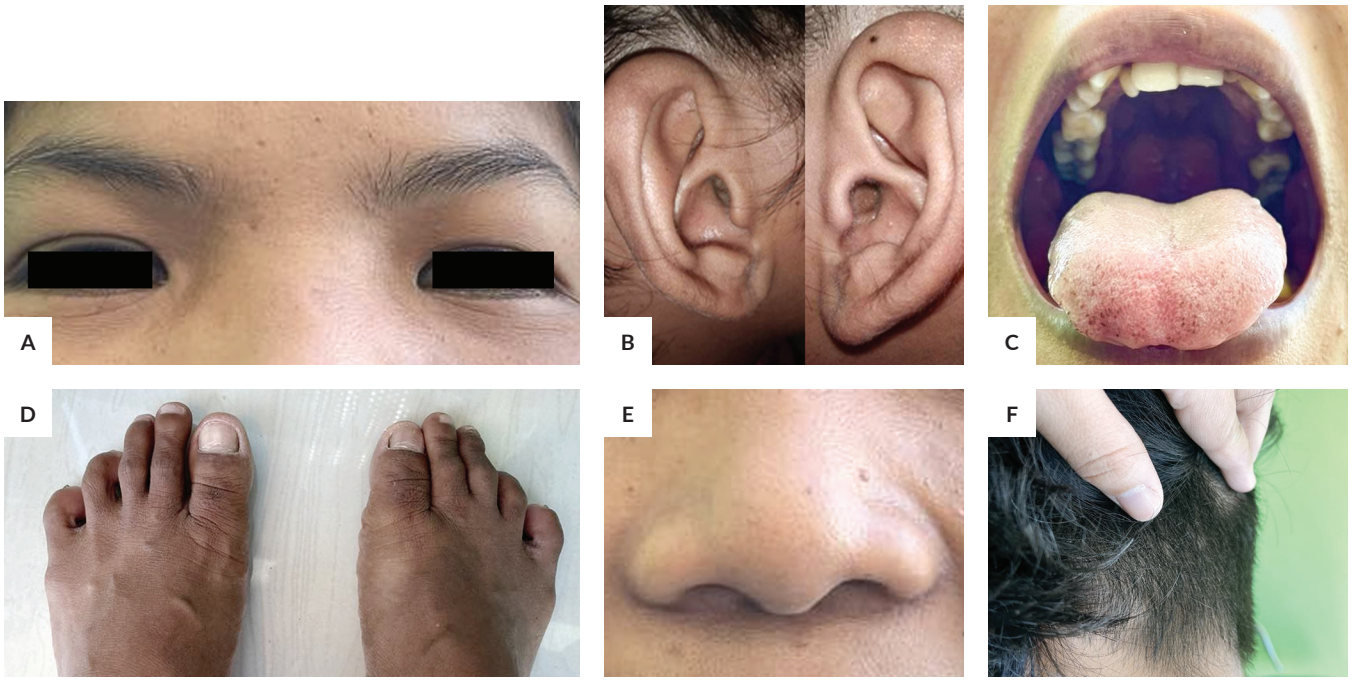

Figure 1. Extra-cranial features of a patient with Mega Cisterna Megna. **(A)** hypertelorism, **(B)** large ear lobules, **(C)** high-arched palate, **(D)** clinodactyly, **(E)** large globular nose, **(F)** bulging occiput.

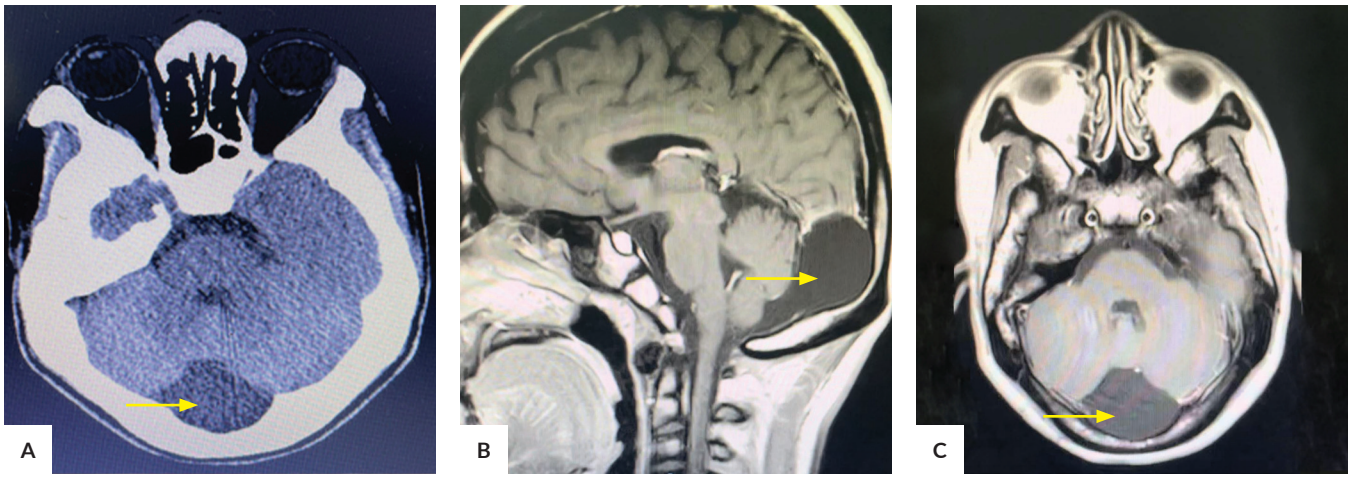

Figure 2. Cranial CT scan, plain, axial **(A)**, and MRI, post-contrast, T1 weighted, sagittal and axial **(B and C)** showing enlarged cisterna magna without associated cerebellar and vermian hypoplasia, and fourth ventricle enlargement *(yellow arrows)*.

Further tests were done while admitted. This included a cranial magnetic resonance imaging (MRI) which ruled out retrocerebellar arachnoid cyst and confirmed Mega Cisterna Magna (Figures 2B and C).

Treatment

The physician-in-charge started the patient on Clonazepam 2 mg one tab at bedtime for the seizure. For the headache, Paracetamol 500 mg, 1 tab every 6 hours, was given. While on admission, coordination with appropriate clinical services was carried out by the physician-in-charge. Clonazepam was discontinued by Neurology service, and the patient was started on Phenytoin 100 mg IV q12, along with Tramadol 500 mg IV q8 as needed for severe headache and ferrous sulfate 1 tablet TID. Phenytoin was eventually tapered to 100 mg BID on the fifth hospital day, and then shifted to Levetiracetam 500 mg BID on the seventh hospital day. Nonpharmacologic intervention by the physician-in-charge included educating the patient and family about the illness through the CEA technique. Patient was discharged on the ninth hospital day.

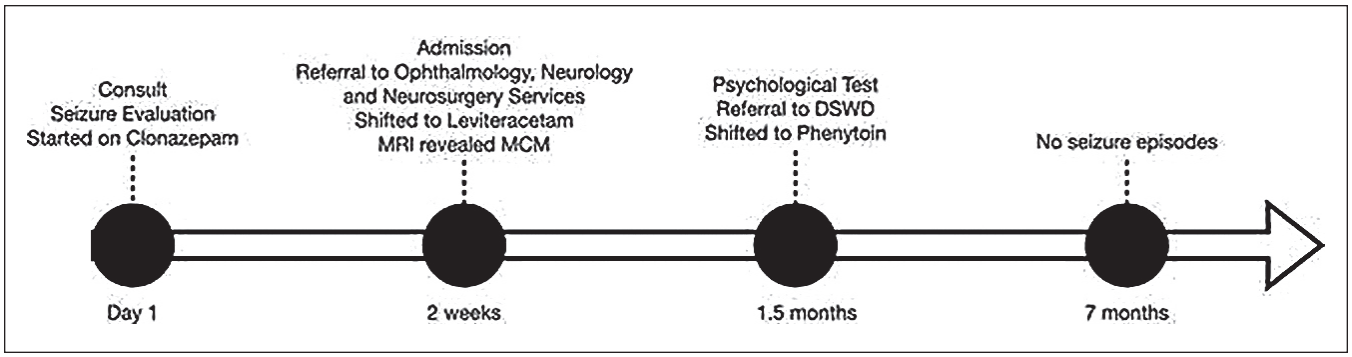

Figure 3. Timeline of patient care.

Outcome and Follow-up

The patient continued to follow-up with the physician-in-charge every month (Figure 3). Patient still had remission of headache and seizure episodes. Levetiracetam was again shifted to Phenytoin 100 mg TID by the physician-in-charge. Phenytoin continued to be tolerated by the patient and at the same time control the seizures. Side-effects of Phenytoin were explained to the patient. Also, referral to Department of Social Welfare and Development (DSWD) and hospital's social service for financial assistance was done. Family meetings were likewise facilitated.

DISCUSSION

The case demonstrates that posterior fossa malformations, which includes MCM and DWM, exhibit diverse clinical presentations, hence patient care should be personalized. Standard seizure evaluation was the approach in the diagnosis of the case as outlined by Gavvala et al. in 2016, which highlights the importance of neuroimaging (i.e., CT scan) and metabolic screening (i.e., hypoglycemia, uremia, and hyponatremia) with adults of new-onset seizures.[17] While the EEG did not reveal epileptiform discharges, MRI confirmed the structural brain anomaly that could explain the recurrent episodes. Treatment, meanwhile, was initially guided by Faulkner et al. in 2016, who recommended Clonazepam for seizure control in neurogenetic syndromes.[18] However, with the evolving clinical picture of a generalized tonic clonic seizure, the standard approach of the use of Phenytoin and Levetiracetam was adopted.[17] The change of medications from Levetiracetam back to Phenytoin underscores the real-world challenges like financial constraints in managing chronic neurological illnesses.

The clinical presentation prompts a deeper consideration of the developmental pathology underlying posterior fossa malformations. Some demonstrated the effects of the rotation of the vermis on the appearance of the fourth ventricle on axial plane images.[16] Without the rotation of the vermis cerebellum (Figure 4A), axial image shows vermian tissue between the fourth ventricle and the cisterna magna. With a small amount of rotation (Figure 4B), axial cuts show some enlargement of the fourth ventricle, but there is still vermian tissue interposed between the fourth ventricle and the cisterna magna. With a large degree of rotation (Figure 4C), there is vermian hypoplasia or aplasia with full communication of the fourth ventricle with the pericerebellar fluid.

Building upon these anatomical observations, researchers proposed that the posterior fossa malformations represent a continuum called the Dandy-Walker Complex: wherein MCM is the most benign to DWM as the most severe form (Figure 5).[16,19,20] However, many still believe that the DWM and MCM cannot be grouped together under DWC based on anatomic features. MCM is characterized by enlarged retrocerebellar space, involuted cyst wall, and without cerebellar hypoplasia; DWM meanwhile is characterized by enlarged retrocerebellar space and with presence of cyst wall and cerebellar hypoplasia. Also in DWM, there is apparently absent vermis with apparent communication of fourth ventricle with very large posterior fossa pericerebellar fluid collection; in MCM, meanwhile, only the cisterna magna is abnormal with >10 mm size on imaging. The controversy of DWC is still found in more recent classifications, wherein DWM was placed under malformations affecting

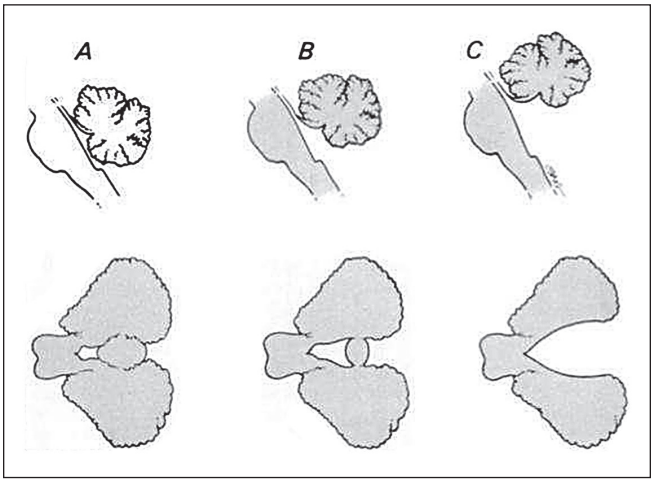

Figure 4. Effect of cerebellar vermis rotation on the development of the fourth ventricle and cisterna magna.[16]

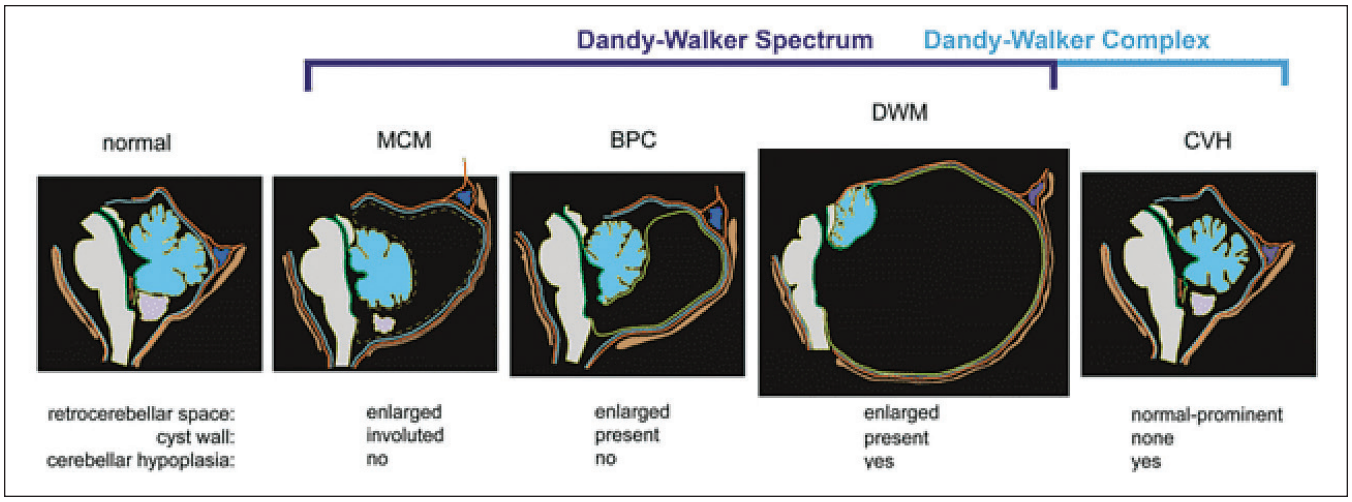

Figure 5. The Dandy-Walker Continuum Classification showing the differences of each entity based on retrocerebellar space, cyst wall and cerebellar hypoplasia. Diagrams represent sagittal magnetic resonance images with anterior left and superior up.[20]

Brainstem – gray, vermis – medium blue, hemisphere – lavender, tela choroidea or cyst membrane – light green, ependyma – dark green, choroid plexus – red, torcula – dark blue, pia – yellow, arachnoid – light blue, dura – orange, bone – brown.

Table 1. Comparison of Features Found in MCM, DWM and Present Case

	Mega Cisterna Magna	Dandy-Walker Malformation	Present case
Cisterna Magna Size	Enlarged, >10 mm	Enlarged, with cystic expansion	Enlarged
Cerebellar Vermis	Intact	Hypoplastic or absent	Intact
Fourth Ventricle	Normal	Enlarged, communicates with cyst	Normal
Extracranial Features	Absent (in isolated cases)	Present (facial and skeletal anomalies)	Present (facie and clinodactyly)
Neurological Symptoms	Often asymptomatic	Hydrocephalus, developmental delay	Seizures, mild cognitive impairment
Prognosis	Good (in isolated cases)	Poor (fetal and early childhood demise)	Guarded (seizure controlled at adulthood)

predominantly the cerebellum and derivatives while MCM was placed in another group of posterior fossa abnormalities with abnormal fluid collections.[21] Other authors, based on imaging, did not include MCM altogether in anomalies with cerebellar hypoplastic or dysplastic changes.[22] Apart from their difference in the anatomy, DWM also presents with extracranial syndromic features, meaning outside the brain abnormalities, such as in the face, extremities, heart, and urinary system. MCM, on the other hand, has been considered just a normal variant of the posterior fossa, as most children with isolated mega cisterna magna develop normally.

The patient reached her adult years having MCM with extra-cranial features of DWM (Table 1). This makes the case a rare one. While present classifications suggest that the Dandy-Walker continuum is unlikely, the mixed presentation could link MCM and DWM together, thereby supporting the notion that DWM and MCM are not separate entities but may share developmental origins.

Like the anatomic classification, the genetic basis for posterior fossa malformations is largely underexplored and complex, especially for the case of this 26-year-old woman with Mega Cisterna Magna and extracranial features suggestive of Dandy-Walker Malformation. There is a high genetic variability among posterior fossa malformations which makes it hard to identify which specific gene is responsible for this case.[1] However, it may be clear that the case is syndromic, based on the case's presentation and the patient's family and obstetric histories. This aligns to the developmental processes associated with posterior fossa malformations.[4] Research on DWM patients elucidated the critical regions for DWM to encompass two adjacent Zinc finger in cerebellum genes, *ZIC1* and *ZIC4* of chromosome 3.[23] Also, case studies on non-isolated MCM have reported MCM in conditions like bilateral coronal synostosis, aplasia cutis congenita, Kleine-Levin syndrome, renal and ear anomalies, and spondylocarpotarsal synostosis, often linked to genetic or chromosomal abnormalities.[24-28] While this is the first recorded case of MCM-DWM phenotype, we can only speculate as to the exact genetic factors at work. A possible genetic link is the *FOXC1* gene, a locus found in animal studies to be associated with cerebellar and posterior fossa malformations including MCM and DWM.[29] Whole-exome sequencing and other advanced genetic technology testing on this locus would be crucial as they can shed light on the pathophysiologic mechanisms underlying the patient's condition. Furthermore, it could demonstrate

the genetic relationship between MCM and DWM, which could bolster the notion that the Dandy-Walker Complex is a genuine continuum. Genetic testing and counseling would also be necessary for the patient and her family due to the potential impact on future pregnancies.

As a number of diseases are now proven to be made up of pathological processes on a continuum and are not just discrete entities (i.e., asthma, Alzheimer's Disease, and micro-macrovascular complications of Diabetes), the existence of this case has potential in directing research on pathogenesis, diagnosis and management of posterior fossa malformations.[30] As seen in genetic studies in diabetic microangiopathy, genetic analysis of this proband is essential.[31] Despite this, clinicians can still cite this case to advise prognosis on patients with more benign conditions in the continuum.

CONCLUSION

This rare case of non-isolated MCM in an adult demonstrated that the Dandy-Walker Complex may have overlapping features and suggests that Mega Cisterna Magna and Dandy-Walker Malformation may be on the same continuum. Despite the presence of clinical signs that point to a more widespread syndrome, thorough genetic testing, including whole-exome sequencing, chromosomal microarray analysis, and karyotyping, was not carried out because of budgetary limitations. This makes it more difficult to pinpoint particular genetic elements that might have influenced the patient's illness. Nevertheless, the concept of the Dandy-Walker Complex as a true continuum needs to be validated and the genetic and clinical spectrum of posterior fossa malformations needs to be further clarified through future prospective studies with larger cohorts and comprehensive genetic analyses.

Informed Consent

Informed consent was obtained from the patient for publication of this case report, including all accompanying images and clinical details. Patient confidentiality has been protected throughout the preparation of this manuscript.

Acknowledgments

The author expresses gratitude to the ISU-COM faculty, the DFCM-CVMC doctors, and Mr. Rey Vincent Villamater for their steadfast support of this work.

Statement of Authorship

The author certified fulfillment of ICMJE authorship criteria.

Author Disclosure

The author declared no conflicts of interest.

Funding Source

None.

REFERENCES

1. Bosemani T, Orman G, Boltshauser E, Tekes A, Huisman T, Poretti A. Congenital abnormalities of the posterior fossa. Radiographics. 2015 Jan-Feb;35(1):200-20. doi: 10.1148/rg.351140038. PMID: 25590398.
2. Nelson M, Maher K, Gilles F. A different approach to cysts of the posterior fossa. Pediatr Radiol. 2004 Sep;34(9):720-32. doi: 10.1007/s00247-004-1253-1. PMID: 15316692.
3. Santoro M, Coi A, Barišić I, Garne E, Addor M, Bergman J, et al. Epidemiology of Dandy-Walker Malformation in Europe: A EUROCAT Population-Based Registry Study. Neuroepidemiology. 2019;53(3-4):169-79. doi: 10.1159/000501238. PMID: 31302658.
4. Kollias S, Ball W, Prenger E. Cystic malformations of the posterior fossa: differential diagnosis clarified through embryologic analysis. Radiographics. 1993 Nov;13(6):1211-31. doi: 10.1148/radiographics.13.6.8031352. PMID: 8031352.
5. Zamora E, Ahmad T. Dandy Walker Malformation. [Updated 2023 Nov 12]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing. 2025 Jan [cited 2025 April]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK538197.
6. Warwick C, Reyes B, Ayoob M, Subit M. Adult diagnosed Dandy Walker malformation presenting as an acute brainstem event—a case report and review of the literature. W V Med J. 2008 Jan-Feb;104(1):25-7. Erratum in: W V Med J. 2008 Mar-Apr;104(2): table of contents. PMID: 18335783.
7. Society for Maternal-Fetal Medicine (SMFM), Monteagudo, A. (2020). Dandy-Walker Malformation. Am J Obstet Gynecol. 2020 Dec; 223(6): B38–B41. doi: 10.1016/j.ajog.2020.08.184. PMID: 33168220.
8. Stambolliu E, Ioakeim-Ioannidou M, Kontokostas K, Dakoutrou M, Kousoulis A. The most common comorbidities in Dandy-Walker Syndrome patients: a systematic review of case reports. J Child Neurol. 2017 Sep;32(10): 886-902. doi: 10.1177/0883073817712589. PMID: 28635420.
9. Ghali R, Reidy K, Fink A, Palma-Dias R. Perinatal and short-term neonatal outcomes of posterior fossa anomalies. Fetal Diagn Ther. 2014;35:108 – 11. doi: 10.1159/000355401. PMID: 24217009.
10. Kim O, Hong S, Jung YM, Seol HJ, Na S, Bae JG, et al. Prenatal Diagnosis and Clinical Outcomes of Isolated Mega Cisterna Magna. JCN. 2024 Oct;39(13-14): 481-486. doi:10.1177/08830738241282037.
11. Patek K, Kline-Fath B, Hopkin R, Pilipenko V, Crombleholme T, Spaeth C. Posterior fossa anomalies diagnosed with fetal MRI: associated anomalies and neurodevelopmental outcomes. Prenat Diagn. 2012 Jan;32: 75 – 82. doi: 10.1002/pd.2911. PMID: 22367673.
12. Forzano F, Mansour S, Ierullo A, Homfray T, Thilaganathan B. Posterior fossa malformation in fetuses: a report of 56 further cases and a review of the literature. Prenat Diagn. 2007 Jun;27: 495-501. doi: 10.1002/pd.1722. PMID: 17367101.
13. Long A, Moran P, Robson S. Outcome of fetal cerebral posterior fossa anomalies. Prenat Diagn. 2006 Aug;26(8): 707-10. doi: 10.1002/pd.1485. PMID: 16764010.
14. Dror R, Malinger G, Ben-Sira L, Lev D, Pick C, Lerman-Sagie T. Developmental outcome of children with enlargement of the cisterna magna identified in utero. J. Child Neurol. 2009 Dec;24(12): 1486-92. doi: 10.1177/0883073808331358. PMID: 19240044.
15. Liu Z, Han J, Fu F, Liu J, Li R, Yang X, et al. Outcome of isolated enlarged cisterna magna identified in utero: experience at a single medical center in mainland China. Prenat Diagn. 2017 Jun;37(6): 575-82. doi: 10.1002/pd.5046. PMID: 28370151
16. Barkovich A, Kjos B, Norman D, Edwards M. Revised classification of posterior fossa cysts and cystlike malformations based on the results of multiplanar MR imaging. AJR Am J Roentgenol. 1989 Dec; 153(6): 1289-300. doi: 10.2214/ajr.153.6.1289. PMID: 2816648.
17. Gavvala J, Schuele S. New-onset seizure in adults and adolescents: a review. JAMA. 2016 Dec 27;316(24): 2657-68. doi: 10.1001/jama.2016.18625. PMID: 28027373.
18. Faulkner M, Singh S. Neurogenetic disorders and treatment of associated seizures. Pharmacotherapy. 2013 Mar;33(3): 330-43. doi: 10.1002/phar.1201. PMID: 23400943.

19. Barkovich A (2012). Developmental disorders of the midbrain and hindbrain. Front Neuroanat. 2012 Mar.6:7. doi: 10.3389/fnana.2012.00007. PMID: 22408608. PMCID: PMC3294267.
20. Hevner R, Millen K, Dobyns W. Dandy–Walker Malformation, Mega Cisterna Magna, and Blake's Pouch Cyst. In: Adle-Biassette H, Harding B and Golden J eds. Developmental Neuropathology, 2nd ed. John Wiley & Sons, Ltd; 2018. pp 141-150. doi: 10.1002/9781119013112.ch13 19.
21. Parisi M, Dobyns W. Human malformations of the midbrain and hindbrain: review and proposed classification scheme. Mol Genet Metab. 2003 Sep-Oct;80(1-2): 36–53. doi: 10.1016/j.ymgme.2003.08.010. PMID: 14567956.
22. Patel S, Barkovich A. Analysis and classification of cerebellar malformations. AJNR Am J Neuroradiol. 2002 Aug;23(7): 1074–1087. PMID: 12169461. PMCID: 8185716.
23. Grinberg I, Northrup H, Ardinger H, Prasad C, Dobyns W, Millen K. Heterozygous deletion of the linked genes ZIC1 and ZIC4 is involved in Dandy-Walker malformation. Nat Genet. 2004 Oct;36(10): 1053–1055. doi: 10.1038/ng1420. PMID: 15338008.
24. Ballard C, Deck J, Iwanaga J, Dumont A, Tubbs S. Bilateral Coronal Synostosis and Mega Cisterna Magna: A Case Report. Cureus. 2022 Jun;14(6): e25717. doi: 10.7759/cureus.25717. PMID: 35812570. PMCID: PMC9261971.
25. Lee E, Park T, Choi Y, Cho E. Familial aplasia cutis congenita associated with mega-cisterna magna. Pediatr Int. 2016 Oct;58(10): 1054-1056. doi: 10.1111/ped.13041. PMID: 27594612.
26. Alqubaty A, Mujlli H, Haroon Z. Rare Case of Kleine-Levin Syndrome with Mega Cisterna Magna. OALib Journal. 2016 Mar;3(3): 1-6. doi: 10.4236/oalib.1102550.
27. Konca Ç, Caliskan B, Tas M. A case with mega cisterna magna renal and ear anomalies: is this a new syndrome?. Case Rep Med. 2013 May;(1): 149656. doi: 10.1155/2013/149656. PMID: 23762068. PMCID: PMC3671267.
28. Assir M, Waseem T. Spondylocarpotarsal synostosis with hydromyelia, mega cisterna magna, and pachydermoperiostosis. Clin Dysmorphol. 2012 Jul;21(3): 144-147. doi: 10.1097/MCD.0b013e3283543224. PMID: 22561843.
29. Aldinger K, Lehmann O, Hudgins L, Chizhikov V, Bassuk A, Ades L, et al. FOXC1 is required for normal cerebellar development and is a major contributor to chromosome 6p25.3 Dandy-Walker malformation. Nat Genet. 2009 Sep;41: 1037–1042. PMID: 19668217. PMCID: PMC2843139.
30. Moynihan R. A new deal on disease definition. BMJ. 2011 May 2011;342:d2548. doi: 10.1136/bmj.d2548.30.
31. Chawla A, Chawla R, Jaggi S. Microvascular and macrovascular complications in diabetes mellitus: Distinct or continuum?. Indian J Endocrinol Metab. 2016 July-Aug;20(4): 546-51. doi:10.4103/2230-8210.183480. PMID: 27366724; PMCID: PMC4911847.