Dandy-Walker Syndrome: Case Report

Síndrome de Dandy-Walker: Relato de caso

Joaquim José de Lima Silva[1]

[1] Plastic Surgery Service, Instituto Dr. José Frota (IJF), Fortaleza, CE, Brazil

Rev Bras Cir Plást 2025;40:s00451812332.

Address for correspondence Joaquim José de Lima Silva, Serviço de Cirurgia Plástica, Instituto Dr. José Frota (IJF), Fortaleza, CE, Brazil (e-mail: cirurgiaoplastico@joaquimjose.med.br; jjdelimasilva@gmail.com).

Abstract

Dandy-Walker syndrome (DWS) is a rare neurodevelopmental condition characterized by abnormalities in the posterior fossa of the skull. It has significant clinical implications, including systemic alterations and delayed motor and cognitive development. The current study presents the case of a 3-year-old child with DWS and no hydrocephalus. We discuss the clinical, genetic, and therapeutic aspects of DWS based on a review of the literature from the past 5 years. We aim to elucidate the phenotypic variability and determine the best diagnostic and therapeutic approaches.

Keywords

- brain diseases
- cerebellar vermis
- cisterna magna
- Dandy-Walker syndrome
- hydrocephalus

Resumo

A síndrome de Dandy-Walker (SDW) é uma condição neuro desenvolvimental rara caracterizada por anomalias na fossa posterior do crânio, com implicações clínicas significativas que incluem atrasos no desenvolvimento motor e cognitivo, além de outras alterações sistêmicas. Este estudo apresenta o caso de uma criança de 3 anos diagnosticada com SDW e sem hidrocefalia associada. São discutidos os aspectos clínicos, genéticos e terapêuticos com base em uma revisão da literatura dos últimos 5 anos, visando compreender melhor a variabilidade fenotípica e as melhores abordagens diagnósticas e terapêuticas.

Palavras-chave

- cisterna magna
- encefalopatias
- hidrocefalia
- síndrome de Dandy-Walker
- vérmis cerebelar

Introduction

The first description of Dandy-Walker syndrome (DWS) dates to 1914, as a complex of cerebellar anomalies involving hypoplasia or agenesis of the cerebellar vermis, dilatation of the fourth ventricle, and enlargement of the cisterna magna.[1] This condition represents approximately 4% of congenital brain malformations, with an estimated incidence of 1:25 thousand to 1:35 thousand live births.[2]

The etiology of DWS is multifactorial, involving genetic, environmental, and epigenetic factors. Chromosomal abnormalities, such as deletions or duplications in specific regions (such as 3q24-q27 and 16p11.2), are frequently present.[3]

Maternal exposure to toxins, viral infections (such as rubella and cytomegalovirus), and gestational diabetes are documented risk factors.[4] Recent reports have highlighted the association of DWS with ocular abnormalities, such as microphthalmia and myopia, suggesting a potential relationship between brain and ocular development.[5]

Clinical manifestations vary widely, ranging from asymptomatic to severe conditions with psychomotor delay, seizures, and visual changes, as in the reported case.[6] The presence or absence of hydrocephalus is a critical determinant for clinical management and prognosis.[7]

Advances in neuroimaging, particularly magnetic resonance imaging (MRI), have transformed the diagnosis of DWS. Magnetic resonance imaging provides a detailed evaluation of cerebellar and ventricular structures, aiding in the differentiation between classic DWS, variants, and similar conditions.[8]

received
January 25, 2025
accepted
July 14, 2025

DOI https://doi.org/10.1055/s-0045-1812332.
ISSN 2177-1235.

Thieme Revinter Publicações Ltda., Rua Rego Freitas, 175, loja 1, República, São Paulo, SP, CEP 01220-010, Brazil

The present article aims to discuss a relevant clinical case and review recent advances in the understanding of DWS, focusing on diagnosis, management, and prognostic implications.

Case Report

A 3-year-old boy was brought in by his paternal aunt due to blindness in his right eye and syndromic abnormalities. Family history reveals multiple cases of blindness and hydrocephalus. Physical examination showed microphthalmia in the right eye and significant myopia in the left eye. ►**Fig. 1** shows the patient's face in an anterior view, demonstrating microphthalmia and syndromic appearance.

A skull MRI revealed hypoplasia of the cerebellar vermis, dilatation of the fourth ventricle, and communication with the cisterna magna, but no signs of hydrocephalus (►**Fig. 2**). The scan also showed no skeletal or renal abnormalities.

The ophthalmologist performed a fundus examination, which showed that the patient presented no signs of increased intracranial pressure. As the child had severe myopia, the ophthalmologist prescribed glasses with corrective lenses for both eyes (even the blind one) to improve balance. After ruling out retinoblastoma, it was decided that the patient should remain under regular clinical follow-up, emphasizing ophthalmological management and multidisciplinary therapies. A pediatric neurologist and a psychology team performed a neuropsychomotor and developmental assessment of the child. The tests diagnosed some level of cognitive and learning deficit, and the family received guidance on how to proceed regarding these issues. Family history reported that the child's father underwent a ventriculoperitoneal shunt in childhood and subsequently developed bilateral blindness; the child's mother had bilateral blindness, and the maternal grandfather had blindness in a single eye.

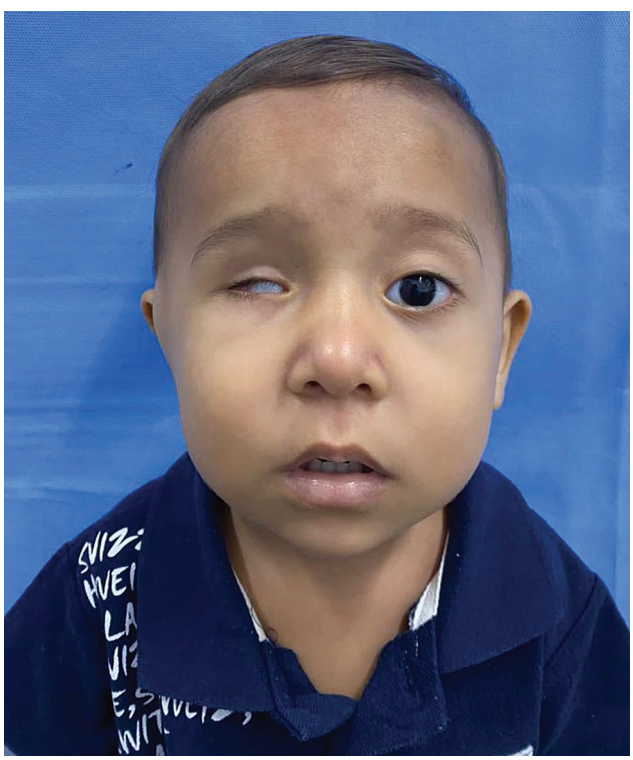

Fig. 1 Image of the patient's face in an anterior view, demonstrating microphthalmia and syndromic appearance.

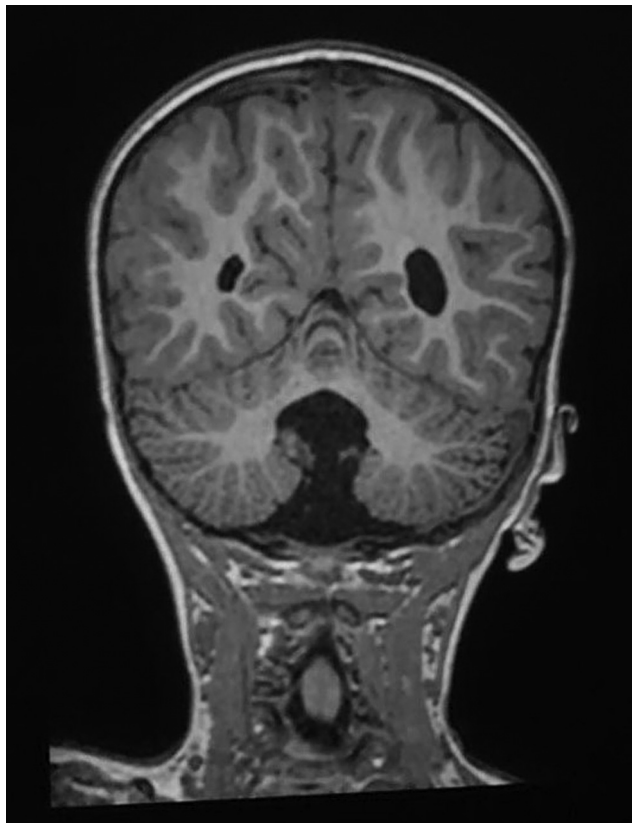

Fig. 2 Magnetic resonance imaging of the skull showing dilatation of the fourth ventricle.

Discussion

Recent studies point to a significant genetic component in DWS, with associations with mutations in genes that regulate brain development (such as *ZIC1* and *FOXC1*).[7] The family history in the reported case reinforces the importance of investigating genetic alterations, especially in contexts of consanguinity or history of congenital malformations.[3]

The literature describes a broad spectrum of clinical presentations, ranging from mild symptoms, such as difficulty with balance, to severe impairments in psychomotor development.[6] The absence of hydrocephalus in the present case is a relevant finding, as it is reported in up to 70% of patients with DWS and usually requires surgical management.[1]

The family history of blindness suggests a potential genetic inheritance, although no specific mutation has been identified to date. In this specific case, it was not possible to evaluate the patient's family members, as they were not located, except for the father, whose medical records were not available. Recent literature indicates the participation of genes involved in brain and eye development, such as *ZIC1* and *ZIC4*, in the pathogenesis of DWS.[5]

Magnetic resonance imaging is the most accurate diagnostic method for DWS.[8] The use of genetic biomarkers, still under development, can improve diagnostic accuracy and identify subjects at higher risk of complications.[9] In the reported case, the absence of clinical signs of hydrocephalus guided the decision for conservative management.

Treatment for DWS depends on the severity of symptoms. Cases without hydrocephalus can be managed with physical therapy, occupational therapy, and psychopedagogical support.[4] Multidisciplinary monitoring is crucial to optimizing the child's development.

In this particular case, the author treated the patient for a crushed finger in the emergency room and followed up for hand trauma at the author's outpatient clinic. Because the child's face had a syndromic appearance, a cranial MRI was requested, and the radiologist suggested the diagnosis.

Early DWS diagnosis is crucial for clinical prognosis and an effective approach to the child's neuropsychomotor development. Prompt identification allows for timely therapeutic interventions, which can significantly improve neurological and functional outcomes. Early intervention is essential to optimize neuropsychomotor development in children with DWS. Early stimulation, physical therapy, occupational therapy, and speech therapy programs should start as soon as possible to address delays in motor and cognitive development. Case studies demonstrate that children who receive early therapeutic interventions have better motor and language skills compared with those who begin treatment later.[10]

Furthermore, regular monitoring by a multidisciplinary team allows for adjustments in therapeutic strategies as the child grows, addressing specific needs and promoting maximum functional independence.[10]

Conclusion

Dandy-Walker Syndrome is a complex condition with wide clinical and prognostic variability. This case illustrates the importance of early diagnosis and a multidisciplinary approach for optimal management. Future studies should focus on genetic and therapeutic advances, enabling a deeper understanding of the condition and improvements in clinical management.[7]

Financial Support
The author declares that he did not receive financial support from agencies in the public, private, or non-profit sectors to conduct the present study.

Clinical Trials
None.

Conflict of Interests
The author has no conflict of interests to declare.

References

1 Pandey S, Bhattarai S, Khatri P. Prenatal ultrasound diagnosis of Dandy-Walker syndrome at 22 weeks of gestation: A case report. J Clin Images Med Case Rep 2024;5(11):3364. Doi: 10.52768/2766-7820/3364
2 Parisi MA, Dobyns WB. Malformations of the Cerebellum and Brainstem. Neuroimaging Clin N Am 2020;30(03):367–380
3 Elsayed LEO, AlHarbi NA, Alqarni AM, Eltayeb HHE, Mostafa NMM, Abdulrahim MM, et al. Chromosome 16p11.2 microdeletion syndrome with microcephaly and Dandy-Walker malformation spectrum: expandinb the known phenotype. Hum Genomics 2020;18(01):95. Doi: 10.1186/s40246-024-00662-0
4 Baidi H, Radi A, Laaraje A, Ourrai A, Hassani A, Abilkassem R. Interstitial deletion of the long arm of chromosome 3, del (3) (q25-q27), in association with Dandy Walker malformation: A case report. World Journal of Advanced Research Reports 2024;23 (03):2239–2244. Doi: 10.30574/wjarr.2024.23.3.2806
5 Aldinger KA, et al. Cerebellar and Ocular Anomalies in Dandy-Walk Malformation: A Case Series. J Child Neurol 2021;36(05): 412–418
6 Spennato P, et al. Ocular Manifestations in Dandy-Walker Syndrome: A Systematic Review. Eur J Ophthalmol 2020;30(04): 678–685
7 Grinberg I, Millen KJ. Genetic Insights into Dandy-Walker Malformation. Genet Med 2022;24(03):567–575
8 Goergen SK, et al. PHACE syndrome: Expanding the phenotype. J Med Imaging (Bellingham) 2021
9 Beyene BG, Abera MT, Zeleka TA, Dhuguma AA. Armored brain as a late complication of CSF overshunting: A rare case report. Radiol Case Rep 2024;20(01):342–345. Doi: 10.1016/j.radcr.2024.10.020
10 Sun Y, Wang T, Zhang N, Zhang P, Li Y. Clinical features and genetic analysis of Dandy-Walker syndrome. BMC Pregnancy Childbirth 2023;23(01):40. Doi: 10.1186/s12884-023-05367-1

