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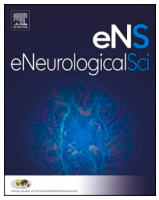

Case report

# Unveiling Dandy-Walker syndrome: A surprising twist in the tale of acute hydrocephalus and Down syndrome child

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ABSTRACT

The correlation between Down syndrome and Dandy-Walker syndrome is an exceptionally uncommon occurrence. To date, only four cases have been documented. All previously reported cases involved individuals under the age of 37 months, with prenatal or birth diagnoses. Additionally, most of these cases displayed a limited life expectancy and experienced poor developmental outcomes. In this report, we present the first-ever instance of an 11-year-old male patient, previously undiagnosed with Dandy-Walker syndrome, who presented with acute intracranial hypertension. Magnetic Resonance Imaging revealed an active hydrocephalus caused by a Dandy-Walker malformation. The patient's condition was effectively managed through the implementation of a ventriculo-cysto-peritoneal shunt. This case highlights the coexistence of Dandy-Walker syndrome and Down syndrome in an asymptomatic young patient. Furthermore, it demonstrates that active hydrocephalus in such cases can be successfully addressed through either endoscopic third ventriculostomy or ventriculo-cysto-peritoneal shunt procedures.

## 1. Introduction

Trisomy 21, also known as Down syndrome [DS], is a genetic condition caused by the presence of an extra copy of chromosome 21. It is associated with various brain anomalies [1]. Many brain anomalies are often observed in individuals with trisomy 21. Individuals with DS typically have smaller brain volumes compared to those without the condition [2]. DS can result in structural anomalies within the brain. Common findings include a smaller and structurally altered hippocampus and alterations in the shape and structure of the ventricles [3]. DS impacts the normal development of the brain, leading to delayed or atypical growth patterns. This affects the formation and organization of neural connections [4]. Trisomy 21 is associated with an increased risk of certain brain disorders. Individuals with DS have a higher likelihood of developing Alzheimer's disease later in life and are at an elevated risk of experiencing seizures or epilepsy [5]. The brain anomalies associated with DS contribute to cognitive and intellectual impairments. Individuals with DS often exhibit varying degrees of intellectual disability, ranging from mild to moderate. Difficulties with learning, memory, attention, and language skills are commonly observed [6]. Understanding the brain anomalies in trisomy 21 is essential for providing appropriate support and interventions to individuals with Down syndrome. Ongoing research aims to further investigate these brain abnormalities to gain insights into their impact and explore potential therapeutic interventions. Dandy-Walker syndrome [DWS] is a congenital brain abnormality affecting the posterior fossa and resulting in developmental and cognitive delays. The coexistence of DS and DWS is extremely rare, with only four reported cases to date, highlighting the infrequency of both conditions occurring together [7–11]. In the present case, we report the first instance of a previously undiagnosed DWS with active obstructive hydrocephalus in an 11-year-old male patient with DS. The patient presented with intracranial hypertension, which was effectively managed through ventriculo-cysto-peritoneal shunting [VCPS].

## 2. Case presentation

An 11-year-old male patient, born to non-consanguineous parents,

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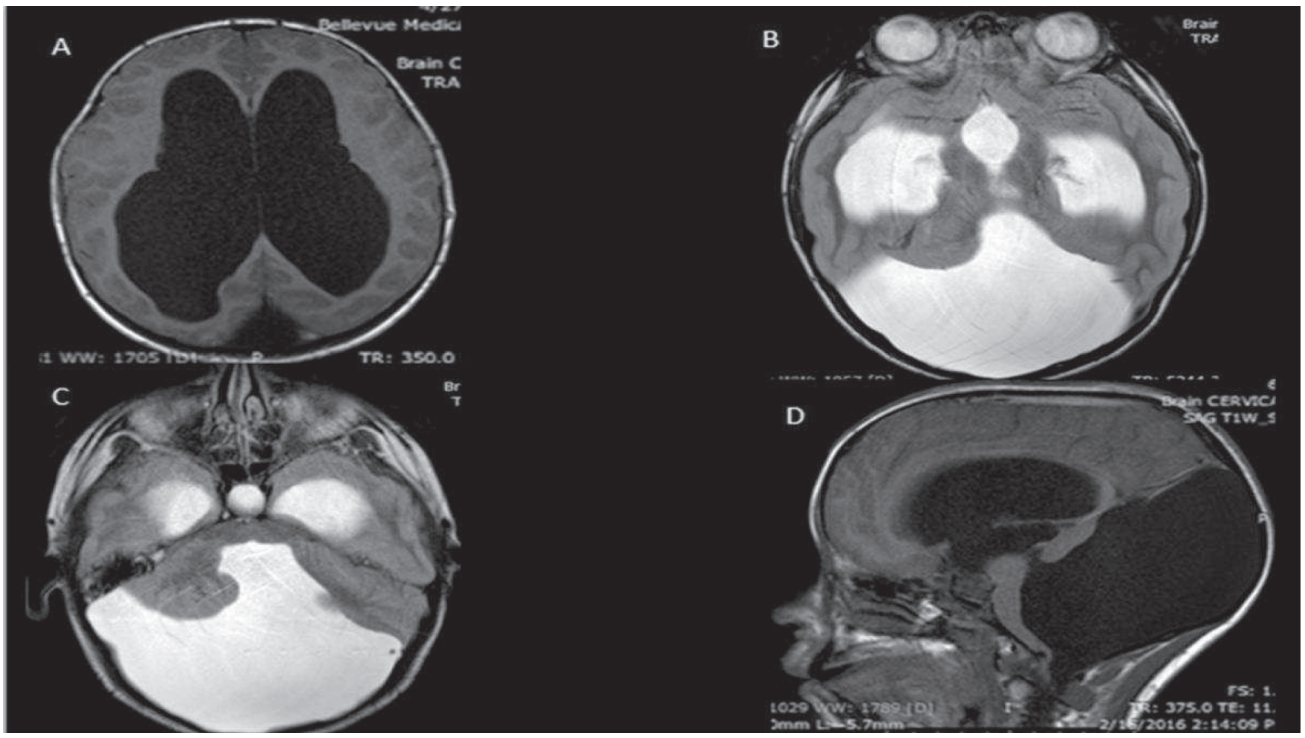


**Fig. 1.** Preoperative MRI.
A: Axial T1 showing lateral ventricular dilatation B& C: Axial T2 showing third ventricular dilatation with Dandy Walker Malformation D: Sagittal T1 showing: the Dandy-Walker Malformation with hypoplasia of the cerebellar vermis, cystic dilation of the fourth ventricle with important enlargement of the posterior fossa.

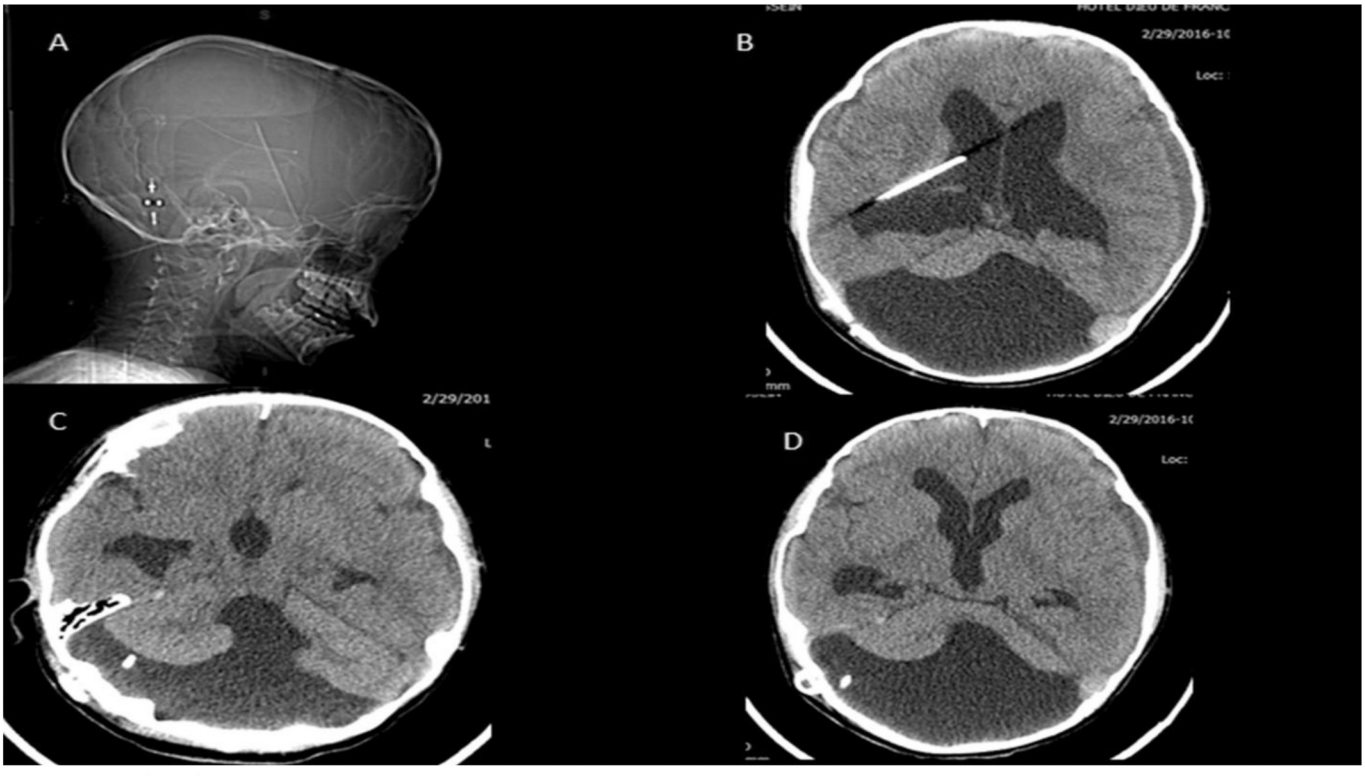


**Fig. 2.** Postoperative CT scanner.
A:Lateral Scout View showing cyst draining catheter connected through a T connector to the ventricular catheter and both connected to an adjustable pressure Sophysa Shunt Valve then to the peritoneal catheter.
B:Axial CT view showing the Right occipital ventricular catheter with major decrease of the size of the ventricles with small bilateral subdural hygromas
C&D: Axial CT view showing the cyst catheter with a major decrease of the size of the fourth ventricule cystic cavity.

**Table 1**
Summary of the English literature reported cases of concomitant Down syndrome and Dandy-Walker Malformation. ICP: intracranial pressure; ILD: interstitial lung disease; PDA: patent ductus arteriosus; ETV: Endoscopic third ventriculostomy; CPS: cysto-peritoneal Shunt.*

| Study | Year of Publication | Age of Patient(s) | Cranial Abnormalities | Management | Other Comorbidities |
|---|---|---|---|---|---|
| Kori et al. [13] | 2022 | 15-month-old premature girl | Dandy-Walker variant with communicating hydrocephalus, and no torcula, tentorium elevation, or posterior fossa expansion | Follow-up due to normal ICP | ILD, PDA |
| Love et al. [11] | 2011 | 37-month-old boy | Asymmetry of the ventricles with excess fluid in the posterior portion of the brain and agenesis of the corpus callosum | Follow-up due to normal ICP | Muscular hypotonia |
| Nigri et al. [9] | 2014 | 19-month-old boy | Tetraventricular hydrocephalus, a posterior fossa cyst and an effacement of his cortical sulcus | ETV | Dysmetric motor pattern |
| Constantini et al. [8] | 1989 | 11-day-old baby | Cystic malformation encompassing the posterior fossa except for the midbrain with obstructive hydrocephalus | CPS | Apathy |
| Nora et al. [14] | 2022 | 3-month-old girl | n/a | Follow-up due to normal ICP | Cardiac defects |

* Data not available.

arrived at the emergency department with complaints of rapidly progressing severe dull neck pain. The pain was partially relieved by assuming an opisthotonic position, but the patient also experienced disabling gait imbalance and dizziness. The patient's medical history included DS and well controlled hypothyroidism with exophthalmia. There were no previous episodes of similar symptoms mentioned. The parents reported that the patient was responsive and energetic.

During the physical examination, the patient was moderately hypotonic but was unable to walk by himself or speak. Opisthotonus (backward arching of the neck and spine) was observed, but there was no fever, nuchal rigidity, or signs of meningism. Before this episode, the patient was fully oriented, and cooperative, and showed no neurological deficits. Upon his arrival at the emergency department, the patient seemed distressed and disoriented. Motor power was 4/5 in the upper limbs and 3/5 in the lower limbs. Laboratory investigations yielded normal results. Fundoscopy revealed bilateral papilledema. A few minutes later, the patient experienced a severe headache, vertical diplopia (double vision), then became somnolent.

An urgent MRI revealed hypoplasia of the cerebellar vermis, cystic dilation of the fourth ventricle, significant enlargement of the posterior fossa, and upward displacement of the lateral sinuses, tentorium, and torcular herophili. Hence, the patient was diagnosed with a Dandy-Walker malformation (DWM). The extension of the fourth ventricle into the foramen magnum caused the displacement of the posterior portion of the brain, resulting in compression of the pons and internal obstruction of normal cerebrospinal fluid (CSF) flow. This led to severe active hydrocephalus and effacement of the cortical sulcus (Fig. 1). Due to signs of severe intracranial hypertension, urgent drainage was deemed necessary. The patient underwent a cysto-ventriculo-peritoneal shunt placement procedure, which involved connecting a cyst-draining catheter to a right occipital ventricular catheter through a T connector. Both catheters were then connected to an adjustable pressure programmable shunt valve and further connected to a peritoneal catheter. Following the surgery, the patient's symptoms resolved. Cystic and CSF cultures showed no evidence of organism growth, and a postoperative CT scan revealed proper positioning of both catheters and a significant decrease in cystic and ventricular size (Fig. 2). Postoperatively, the patient regained full autonomy, and during the 2-year follow-up, remained symptom-free with continued decompression of the cyst. The boy's lack of coordination prevented him from performing tasks like brushing his teeth and getting dressed, which was one of the key qualitative challenges in learning fine motor performance. No further imaging was done during the follow-up period. No emergency department visits were recorded.

## 3. Discussion

Dandy-Walker syndrome (DWS) is a relatively rare condition characterized by cerebellar malformation, including hypoplasia of the cerebellar vermis, dilatation of the fourth ventricle, and an enlarged posterior fossa [12]. The cases reported in the literature are few and a summary of the English literature related to this topic is depicted in Table 1.

Diagnosis of DWS in utero can be challenging as it is often indistinguishable from associated hydrocephalus [12]. DWS is classified into two subcategories: Dandy-Walker malformation (DWM) with posterior fossa enlargement, and Dandy-Walker variant without the latter [11]. The exact cause of DWS is unknown, although maternal viral infections and alcohol consumption have been suggested as potential risk factors [15]. Genetic and chromosomal abnormalities have also been proposed but not yet confirmed [16,17].

DWS can be associated with anomalies outside the central nervous system (CNS), such as cardiac defects, craniofacial abnormalities, gastrointestinal abnormalities, genitourinary abnormalities, respiratory aberrations, and musculoskeletal dysmorphisms [18]. The presence of extra-neurologic abnormalities in DWS is associated with a higher risk of developmental delays [18].

The coexistence of Down syndrome (DS) and DWM is extremely rare, with only four reported cases to date. Constantini et al. described a case of an infant who passed away at two weeks of age [8]. Estroff et al. reported a severely handicapped four-month-old infant with a DWS variant [10]. Love et al. outlined the delayed developmental outcomes in a 37- month-old child diagnosed with both DWS and DS at birth [11]. Nigri et al. detailed the case of a 19-month-old child diagnosed with DWS and DS at birth, who showed positive developmental outcomes [9]. All previously reported cases were diagnosed prenatally or at birth and were younger than 37 months. Most of them had a short life expectancy and poor developmental outcomes.

The main treatment modalities for DWS are cysto-peritoneal shunt (CPS), ventriculoperitoneal shunt (VPS), and cysto-ventriculo-peritoneal (CVPS) shunt. In this case, we present an 11-year-old patient with Down syndrome who presented with acute active obstructive hydrocephalus, which was found to be associated with previously undiagnosed DWS. This is the first reported case of such a presentation in a patient older than 34 months. This raises the possibility of preserved developmental features in some patients with coexisting DWS and DS. Nigri et al. treated obstructive hydrocephalus in their case with an endoscopic third ventriculostomy. In our case, we opted to manage both the cyst and hydrocephalus using a ventriculo-cysto-peritoneal shunt (VCPS), which proved to be an effective strategy. Lin et al. published in 2015 the results of a 12-year follow-up of a 2-year-old boy with DWS who was treated using VCPS [19]. Ultimately, the removal of the shunt was successful as the patient was no more shunt-dependent after the healing of the cyst. Baskin et al. reported that 64% of patients become shuntindependent after a mean follow-up period of 21.4 months [20]. The authors devised a fiberoptic intracranial pressure monitoring system that allowed patients' assessment after the removal of the VCPS [19].

## 4. Conclusions

DWS is a rare disorder seen in pediatric patients; its association with DS is almost uncommon. Asymptomatic coexistence can remain hidden, however, acute exacerbation and active hydrocephalus should be urgently managed by either endoscopic third ventriculostomy or ventriculo-cysto-peritoneal shunt.

## CRediT authorship contribution statement

**Elie Fahed:** Data curation, Conceptualization. **Ali Msheik:** Writing – review & editing, Writing – original draft, Supervision, Software, Methodology, Formal analysis. **Maya Rahal:** Resources, Investigation. **Stephanie Antoun:** Writing – original draft, Visualization, Formal analysis. **Caroline Geagea:** Writing – original draft, Visualization, Validation. **Philippe Younes:** Writing – review & editing, Project administration, Methodology, Conceptualization.

## Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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