A Case of Newly Diagnosed Dandy-Walker Malformation in an Adult

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CASE DIAGNOSIS

Hydrocephalus secondary to Dandy-Walker Malformation (DWM)

CASE DESCRIPTION

A 56-year-old female accountant presented to our clinic with a 3-month history of bilateral frontotemporal headaches. Her symptoms began insidiously, without any known trauma or inciting event, and have progressively worsened since onset. During our evaluation, she described having 5-7 daily episodes of moderate to severe “throbbing” headaches with ocular radiation. Associated symptoms included nausea, vomiting, and photophobia. Her symptoms were exacerbated by light, noise, and bending over, and relieved by avoidance of these aggravating factors. A trial of tramadol resulted in only mild reduction in the severity of her headaches and worsened her nausea.

Since the onset of her symptoms, she has also experienced dizziness and difficulty maintaining her balance, resulting in a recent fall and numerous near falls. She further noted recent urinary urgency, memory difficulty, upper extremity tremors, pulsatile tinnitus, and generalized weakness. The patient had no prior history of similar complaints. Her medical history was notable for hypertension, glaucoma, and Peters’ anomaly (anterior segment dysgenesis) in her left eye.

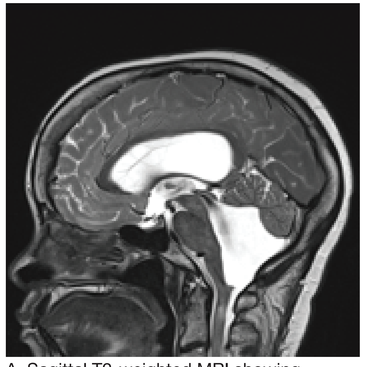
Before she arrived at our clinic, she underwent a neuroradiology studies ordered by her primary care physician, which demonstrated findings consistent with hydrocephalus and elevated intracranial pressure, secondary to a previously unknown diagnosis of Dandy-Walker Malformation. Computed tomography revealed dilation of the lateral ventricles, significant cystic dilation of the fourth ventricle, hypoplasia of the vermis, and superior displacement of the cerebellum. Subsequent magnetic resonance imaging additionally reported anterior compression of the posterior brainstem (Fig. 1). Optic nerve sheath CSF distention and ectasia was also noted.

On physical exam, her blood pressure was 148/87, with otherwise normal vital signs. She demonstrated saccadic eye movements and nystagmus. Motor exam was notable for trace left upper extremity pronator drift. Sensation to light touch was grossly intact. Swaying was observed on Romberg. Her balance was poor, evidenced by her inability to perform a heel, toe, or tandem walk. She required hand-held assistance for ambulation and exhibited a shuffling gait.

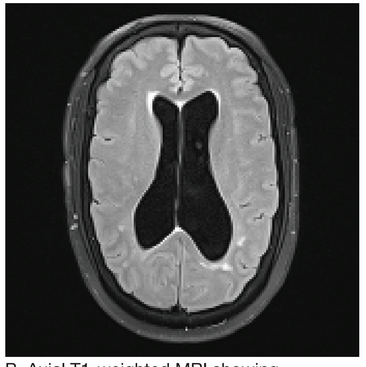
The patient was sent to the emergency department for an urgent evaluation. She was seen by neurology, neurosurgery, and ophthalmology. Her fundoscopic exam was found to be benign, specifically without papilledema. Immediate surgical intervention was ultimately deferred, and she was cleared for outpatient follow-up. Her discharge medications included topiramate, ondansetron, and acetaminophen. At follow up, she was started on acetazolamide and referred to physical therapy for balance exercises. Headaches, dizziness, urine incontinence, and balance all improved.

FIGURE 1.

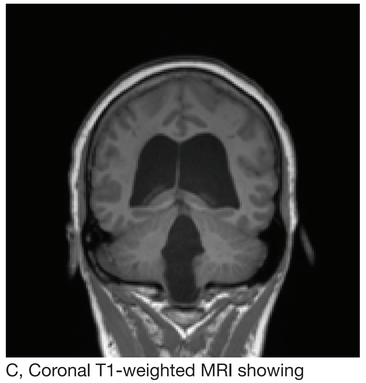
MRI of a 56-year-old female with Dandy-Walker Malformation



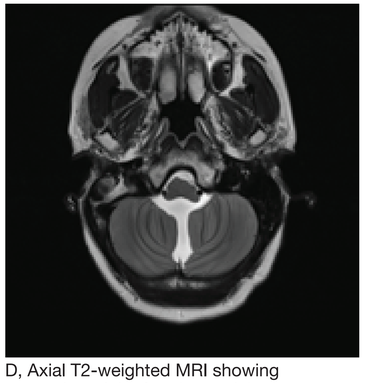
A, Sagittal T2-weighted MRI showing vermian hypoplasia with marked cystic dilation and expansion of the fourth ventricle, displacing the brainstem anteriorly.



B, Axial T1-weighted MRI showing hydrocephalus of the lateral ventricles.



C, Coronal T1-weighted MRI showing enlarged lateral and fourth ventricles.



D, Axial T2-weighted MRI showing expansion of the fourth ventricle.

DISCUSSION

The Dandy-Walker malformation (DWM) is the most common congenital cerebellar malformation, with an estimated incidence between one in 25,000 to one in 30,000 births.[1,2] First described by Dandy and Blackfan in 1914, DWM is classically characterized by a triad of cystic dilation of the fourth ventricle, hypoplasia or aplasia of the vermis, and hydrocephalus.[3,4] The vast majority of cases (80–90%) present in the first year of life with signs and symptoms of hydrocephalus and increased intracranial pressure, notably macrocrania.[2] While DWM is rare, adult cases are far more uncommon.

In this report, we describe a 56-year-old woman who presented to us with a new diagnosis of DWM. Some adult patients, as in this case, present with headache and gait disturbance.[1] Other cases are found incidentally in asymptomatic patients or during autopsy.[5] Additional case reports have identified DWM in patients presenting with psychosis, stroke, mimicking myasthenia, and HIV.[6,7,8,9]

This patient also has Peters’ anomaly, a rare congenital ocular anomaly characterized by anterior segment dysgenesis. Notably, mutations in the forkhead transcription-factor gene (FOXC1) cause abnormal anterior segment development and result in a wide spectrum of glaucoma-associated phenotypes, including Axenfeld–Rieger Syndrome and Peters’ anomaly.[10] In addition to ocular development, FOXC1 plays an important role in cerebellar and posterior fossa development and is one of many mutations associated with DWM.[11] While case reports have separately observed FOXC1 mutations in patients with Peters’ as well as DWM, Peters’ is not commonly seen in DWM and to our knowledge this is the first case report identifying a patient with both phenotypes present.

Treatment for DWM remains individualized and includes posterior fossa craniectomy with cyst excision, ventriculoperitoneal shunt, cystoperitoneal shunt, or ventriculocysto shunt, as described by Torkildsen.[12,13] For this patient, acetazolamide was used to reduce secretion of cerebrospinal fluid from the choroid plexus, thereby reducing hydrocephalus and her symptoms. We recognize that with time, acetazolamide may lose its efficacy, leading to shunt placement.

CONCLUSION

In this case report, we describe a patient with symptomatic DWM, which rarely presents in adulthood. She initially experienced mild relief with acetazolamide but later required definitive treatment with a VP shunt.

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