Dandy Walker malformation and hypertrophic cardiomyopathy

Unusual fatal association

Maher E. Kurdi, *MBBS,* ***Mohammed A. Chamsi-Pasha,*** *MBBS,* ***Saleh S. Baeesa,*** *MBChB, FRCSC,* ***Mohammed M. Jan,*** *MBChB, FRCSC.*

ABSTRACT

تعتبر متلازمة الداندي واكر (DWM) من التشوهات الخلقية النادرة بالدماغ وينتج عنها تضخم في التجاويف الداخلية بالدماغ وضمور بالمخيخ. ومن الممكن حدوث تشوهات أخرى خارج الدماغ و منها عيوب بالقلب. نقدم في هذه الورقة العلمية تقريراً عن حالة طفل مصاب بمتلازمة الداندي واكر DWM مع وجود اعتلال شديد ومتدهور بعضلة القلب المتضخمة والذي نتج عنه هبوط في وظائف القلب. وقد تم تشخيص الحالة في عمر 2 شهر و حدثت الوفاة بعد 5 أشهر من ذلك. ونستنتج من هذه الحالة أن اعتلال عضلة القلب المتضخمة من الممكن حدوثه مع متلازمة الداندي واكر DWM. وبالتالي يجب تقييم وظائف القلب لدى حديثي الولادة المصابين بمتلازمة الداندي واكر DWM للتعرف مبكراً علي إصابة القلب ومحاولة علاجها.

Dandy Walker malformation (DWM) is a rare congenital brain anomaly characterized by cystic dilation of the fourth ventricle and hypoplasia of the cerebellar vermis. Other extracranial anomalies can be associated, including cardiac defects. We report a rare patient with DWM associated with progressive heart failure secondary to hypertrophic cardiomyopathy. He was diagnosed at 2 months of age and died 5 months later. We conclude that hypertrophic cardiomyopathy can be associated with DWM with poor prognosis. A careful cardiac evaluation is needed in all infants with DWM for early recognition of such potentially serious associated cardiac malformations.

Neurosciences 2009; Vol. 14 (4): 368-370

From the Department of Neurosciences (Kurdi, Chamsi-Pasha), King Faisal Specialist Hospital & Research Center, and Faculty of Medicine (Baeesa, Jan), King Abdulaziz University, Jeddah, Kingdom of Saudi Arabia.

Received 16th March 2009. Accepted 6th May 2009.

Address correspondence and reprint request to: Dr. Mohammed M. Jan, Department of Pediatrics, King Abdul-Aziz University Hospital, PO Box 80215, Jeddah 21589, Kingdom of Saudi Arabia. Tel. +996 (2) 6401000. Ext. 20208. Fax. +996 (2) 6403975. E-mail: mmsjan@yahoo.ca

The Dandy-Walker complex is a continuum of aberrant development of the posterior fossa that has been associated with multiple congenital anomalies, radiographic abnormalities, and developmental delay.[1] Dandy Walker malformation (DWM) is rare and characterized by hypoplasia of the cerebellar vermis, cystic dilatation of the fourth ventricle, and enlargement of the posterior fossa. The incidence is 1:25,000-35,000 births with a female to male ratio of 1.5:1.2 Congenital cardiac defects can be associated with this syndrome, and often correlates with poor prognosis.[2] Hypertrophic cardiomyopathy (HCM) is a severe cardiac defect with poor outcome.[3] We describe a rare case of HCM associated with DWM to highlight this rare and serious association.

Case Report. A 2-month-old male infant presented for the first time with shortness of breath and partial motor seizures. He was a product of full term uneventful pregnancy, delivered by cesarean section for breach presentation. His Apgar scores were 8 and 9, at one and 5 minutes. His parents are first-degree cousins. On examination, his heart rate was 180, and his respiratory rate was 45. His head circumference was 43 cm (95th percentile) at birth while his weight and height were along the 50th percentile. He had dysmorphic features in the form of course elongated face with low set ears, hypertelorism, long filtrum, anteverted nares, and short broad hands. His heart sounds were normal, and he had no murmurs. Chest examination revealed bilateral inspiratory crepitations with equal air entry. Neurological examination demonstrated no cranial neuropathy. His tone and power in the upper and lower limbs were normal but brisk reflexes. Investigation included a normal complete blood count and biochemistry profile. An MRI of the brain revealed large cystic dilatation of the fourth ventricle with hypoplastic cerebellum suggestive of DWM (**Figure 1**). Other investigations included normal karyotype (46 XY), negative toxoplasmosis,

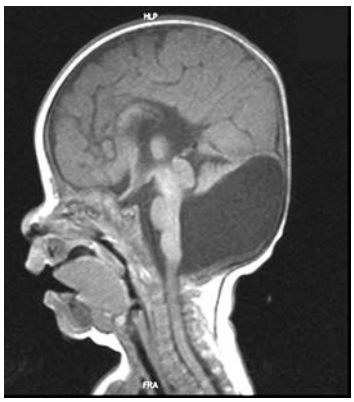

Figure 1 - Sagittal T1-weighted MRI scan demonstrating large posterior fossa cystic dilatation of the fourth ventricle associated with cerebellar hypoplasia and corpus callosal agenesis.

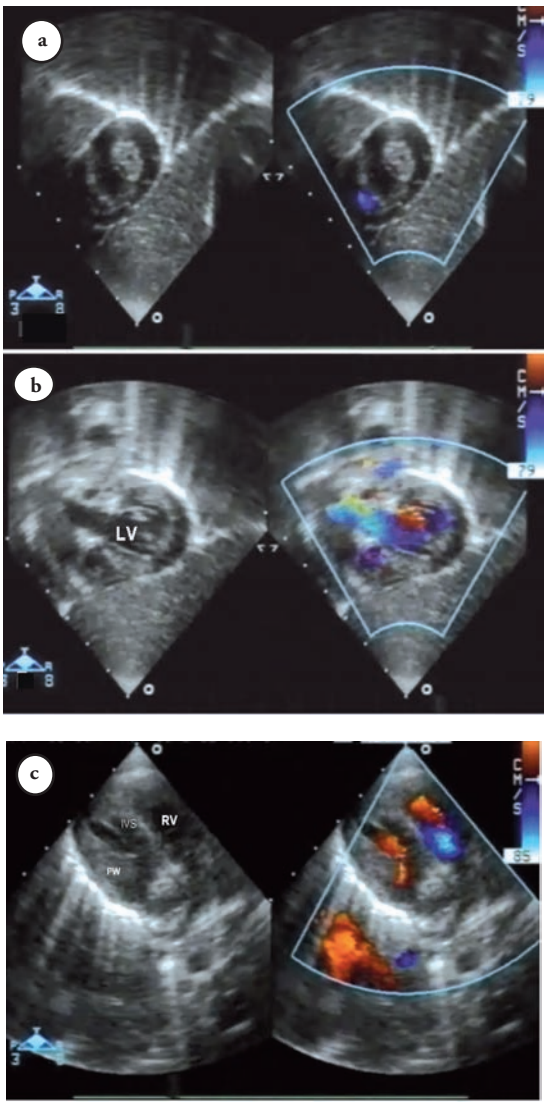

Figure 2 - Echocardiographic scans through subcostal view **a)** demonstrating left ventricular hypertrophy without left ventricular outflow tract obstruction. Parasternal view **b & c)** demonstrating hypertrophic ventricular septum (IVS), posterior (PW) and lateral wall of the left ventricle (LV) RV - right ventricle

rubella, cytomegalovirus, herpes simplex, and HIV (TORCH) screen, and normal metabolic and thyroid profile. Electrocardiogram revealed sinus tachycardia with right axis deviation and bi-ventricular hypertrophy. Chest radiography showed significant cardiomegaly with increased bronchovascular markings. Echocardiography showed concentric left ventricular hypertrophy with normal myocardial contractility (**Figure 2**). The thickness of the interventricular septum and left ventricular lateral wall were 4.5 and 2.5 standard deviations above the mean. There was no left ventricular outflow tract obstruction or other congenital abnormalities. Electroencephalogram revealed multi-focal epileptiform discharges originating from both hemispheres independently. Cysto-peritoneal shunt was successfully performed. The postoperative head circumference decreased to 38 cm after several weeks. He was stabilized on antiepileptic drugs (carbamazepine and levetiracetam), digoxin, and propranolol. Over the next 4 months, he was readmitted several times with recurrent respiratory distress and infection. At the age of 7 months, he suddenly arrested at home after a short period of respiratory distress and was pronounced dead on arrival at the emergency department.

Discussion. The DWM has been classically described as a triad consisting of cystic dilation of the fourth ventricle, complete or partial agenesis of the cerebellar vermis, and an enlarged posterior fossa.[1] The etiology is unknown, but it was thought to be the result of congenital atresia of the foramina of Luschka and Magendie causing dilatation of the fourth ventricle.[2] The DWM may be difficult to identify early due to lack of distinctive symptoms or signs. Progressive macrocephaly secondary to hydrocephalus (70-90%) may result in early recognition.[4] The DWM can be associated with several extracranial anomalies, particularly facial and cardiac defects.[4] In one retrospective series, cardiac anomalies were found in 42% of DWM patients.[5] Reported congenital heart defects included ventricular septal defect, atrioventricular septal defect, atrial septal defect, pulmonary stenosis, and patent ductus arteriosus.[6] Our patient had echocardiographic features of HCM, which were not previously reported in patients with DWM. Pediatric HCM is a rare, but serious cardiac condition.[3,7] The clinical hallmark of HCM is ventricular hypertrophy without an identifiable hemodynamic cause.[8] The HCM is usually idiopathic, however, inborn errors of metabolism, genetic syndromes, and neuromuscular

disorders, are other possible etiologies.[8] Our patient was 2 months old when HCM was diagnosed, and had no chromosomal or metabolic disorder, and no family history of HCM. Many patients with HCM (40%) die with within 2 years of diagnosis.[3,7] The mortality rate is even higher for those presenting during infancy. Congestive heart failure is the most common cause of death.[8] This was the situation in our patient. The DWM by itself is not fatal, however, other associated anomalies contribute to most of the deaths (83%) in these patients.

We conclude that HCM can be associated with DWM with poor prognosis. A careful cardiac evaluation is needed in all infants with DWM for early recognition of such potentially serious associated cardiac malformations.

Acknowledgments. *We gratefully acknowledge Dr. Mai Al-Barakati, Department of Pediatric Cardiology, King Fahad Armed Forces Hospital, Jeddah, Kingdom of Saudi Arabia for providing the cardiac evaluation and echocardiography illustrations for the reported patient.*

References

1. Kumar R, Jain MK, Chhabra DK. Dandy-Walker syndrome: different modalities of treatment and outcome in 42 cases. ***Childs Nerv Syst*** 2001; 17: 348-352.
2. Pascual-Castroviejo I, Velez A, Pascual-Pascual SI, Roche MC, Villarejo F. Dandy-Walker malformation: analysis of 38 cases. ***Childs Nerv Syst*** 1991; 7: 88-97.
3. Nugent AW, Daubeney PE, Chondros P, Carlin JB, Colan SD, Cheung M, et al. Clinical features and outcomes of childhood hypertrophic cardiomyopathy: results from a national population-based study. ***Circulation*** 2005; 112: 1332-1338.
4. ten Donkelaar HJ, Lammens M, Wesseling P, Thijssen HO, Renier WO. Development and developmental disorders of the human cerebellum. ***J Neurol*** 2003; 250: 1025-1036.
5. Sasaki-Adams D, Elbabaa SK, Jewells V, Carter L, Campbell JW, Ritter AM. The Dandy-Walker variant: a case series of 24 pediatric patients and evaluation of associated anomalies, incidence of hydrocephalus, and developmental outcomes. ***J Neurosurg Pediatrics*** 2008; 2: 194-199.
6. Cornford E, Twining P. The Dandy-Walker syndrome: the value of antenatal diagnosis. ***Clin Radiol*** 1992; 45: 172-174.
7. Lipshultz SE, Sleeper LA, Towbin JA, Lowe AM, Orav EJ, Cox GF, et al. The incidence of pediatric cardiomyopathy in two regions of the United States. ***N Engl J Med*** 2003; 348: 1647-1655.
8. Colan SD, Lipshultz SE, Lowe AM, Sleeper LA, Messere J, Cox GF, et al. Epidemiology and cause-specific outcome of hypertrophic cardiomyopathy in children: findings from the Pediatric Cardiomyopathy Registry. ***Circulation*** 2007; 115: 773-781.

Related topics

Erbagci I, Erbagci H, Erbagci Z, Sivasli E, Bekir N. Unilateral nystagmus in an infant with acrocallosal syndrome. ***Neurosciences*** 2004; 9: 315-318.

Chacko A, Koul R, Sankhla DK. Corpus callosum agenesis. ***Neurosciences*** 2001; 6: 63-66.