

A Newborn with Dandy-Walker Malformation and Aortic Coarctation

Dandy-Walker Malformasyonu ve Aort Koarktasyonu Olan Bir Yenidoğan

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Abstract

Dandy-Walker malformation (DWM) is a rare anomaly of the posterior cranial fossa. Concomitant brain or systemic malformations are frequent and can influence the prognosis. DWM is associated with cardiac abnormalities. Recognition of these anomalies are important for diagnosis and accurate surgical management. Association of aortic coarctation with DWM is extremely rare. Here, we report a newborn with DWM and aortic coarctation. Our aim was to emphasize that DWM can be associated with cardiac anomalies. Cardiac congenital defects are often associated with a poor prognosis. These kinds of features should alert the clinician to consider extensive screening in these patients not only for cerebral structures but also for cardiovascular abnormalities.

Keywords

Newborn, Dandy-Walker malformation, aortic coarctation

Anahtar Kelimeler

Yenidoğan, Dandy-Walker malformasyonu, aort koarktasyonu

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Öz

Dandy-Walker malformasyonu (DWM) posterior kraniyal fossanın nadir görülen bir anomalisidir ve beraberinde beyin veya sistemik malformasyonlar sıklıkla eşlik edebilmekte ve prognozu etkileyebilmektedir. DWM'ye eşlik eden kardiyak anomalilerde bulunmaktadır. Bu anomalilerin tanınması, klinik tanı ve uygun cerrahi tedavinin düzenlenmesine yardımcı olur. Aort koarktasyonu ve Dandy-Walker varyantı birlikteliği çok nadir olarak görülmektedir. Burada, DWM ve aort koarktasyonu olan bir yenidoğanı sunuyoruz. Amacımız DWM'nin kardiyak anomalilerle birlikte olabileceğini vurgulamaktır. Kardiyak konjenital defektler genellikle kötü prognozla ilişkilidir. Bu tür özellikler, klinisyeni bu tür hastalarda sadece serebral yapılarla değil kardiyovasküler anomalilere de kapsamlı tarama yapmaları konusunda uyarmalıdır.

Introduction

Dandy-Walker malformation (DWM), a congenital anomaly of the posterior cranial fossa, characterized by the triad of cystic dilatation of the fourth ventricle, enlarged posterior fossa, and complete or partial agenesis or absence of the cerebellar vermis and hydrocephalus (1,2). The malformation occurs in approximately 1 per 25.000 live births, more frequently in girls than in boys (3). Usually, it is diagnosed in the prenatal period or early infancy, rarely in adulthood (3). Systemic anomalies in DWM may include cleft lip and palate, polydactyly,

craniofacial malformations, and cardiac abnormalities (4). Coarctation of the aorta and its associated cardiac anomaly are occasionally seen in DWM (5). Here, we report a newborn with DWM and aortic coarctation.

Case Report

Our case was the second child of non-consanguineous, healthy parents. He was delivered by elective Cesarean section at 39 weeks of gestation. There was no prenatal ultrasound diagnosis. His birth weight was 4600 g (>90th centile), length was 49 cm (75th centile), and occipital frontal circumference was 38 cm (96th centile). Physical examination indicated macrocephaly, frontal bossing, small palpebral fissures, apparently low-set ears, and a flat nasal bridge. During the examination, the patient's blood pressure in the upper extremity was 85/47 mmHg and 67/35 mmHg in the lower extremity. There was a systolic pressure gradient between the upper and lower limbs. His pulse rate was 140 beats/min and regular. A systolic murmur was heard at the right sternal border; pedal and femoral pulses were weak. Other systemic examinations were normal. With a poor general condition and respiratory dysfunction, the patient was intubated and was connected to a mechanical ventilator. Brain magnetic resonance imaging (MRI) showed dilatation of the lateral ventricles and the third ventricle, a thin corpus callosum, hydrocephalus, cerebellar hypogenesis and DWM (Figure 1). Echocardiography and MRI angiography showed a patent ductus arteriosus, moderate aortic valve dysplasia, and severe aortic coarctation distal to the left subclavian vessel (Figure 2). His abdominal ultrasound was normal. The karyotype was normal male 46, XY. To ensure the continuation of the ductus arteriosus patency, prostoglandin E1 infusion was started. His hydrocephalus was treated with a ventricular peritoneal shunt. At postoperative fourth day, sepsis and acute renal failure developed. Later, he developed respiratory dysfunction, multisystem organ failure and had a cardiorespiratory arrest. He died at the age of 9 days. Informed consent was obtained from the parents.

Discussion

The cause of DWM is unknown; it develops early *in utero*, during the embryonic stage (fifth-sixth week



Figure 1. Brain magnetic resonance imaging indicated dilatation of the lateral ventricles and the third ventricle, a thin corpus callosum, hydrocephalus, cerebellar hypogenesis and a Dandy-Walker malformation

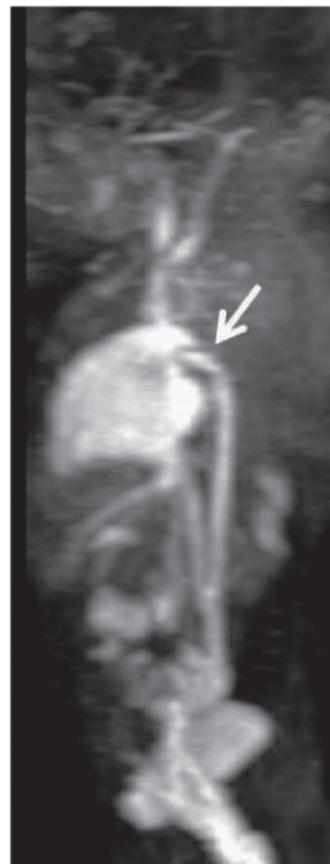


Figure 2. Magnetic resonance imaging angiography showed a patent ductus arteriosus, moderate aortic valve dysplasia, and severe aortic coarctation distal to the left subclavian vessel

post conception) and is described as an error in the development of the rhombencephalon, leading to atresia of the foramina of the fourth ventricle and delayed opening of the foramen of Magendie (6). Various predisposing factors have been reported: viral infections (rubella, cytomegalovirus), toxoplasmosis and exposure to warfarin during the first trimester of pregnancy, maternal alcoholism and diabetes, chromosomal defects and consanguinity, and complicated monochorionic twins, especially in the smaller one (7-9). Systemic anomalies in DWM may include cleft lip and palate, polydactyly, craniofacial malformations, and cardiac abnormalities (4). Coarctation of the aorta and its associated cardiac anomaly are occasionally seen in DWM (5). Sasaki-Adams et al. (10) studied 24 pediatric patients with DWM (10 male and 14 female) and found that cardiovascular abnormalities were the most frequently associated anomaly. Ten of 24 patients (41.7%) had patent ductus arteriosus, ventricular septal defect, atrial septal defect, transposition, hypoplastic right heart, or pulmonary artery stenosis. Also in Turkey, Güven et al. (11) reported a two-day-old male patient with DWM and pentalogy of Fallot.

Congenital cardiac defects associated with the syndrome often lead to poor prognosis resulting in death, and therefore, need a precise prenatal diagnosis (12). Here, we report a newborn with DWM and aortic coarctation. Our aim was to emphasize that a better and ready determination of the neurological malformation would have made the diagnosis of the aortic coarctation simpler and precocious, knowing that DWM can be associated with cardiac anomalies. These kinds of features should alert the clinician to consider extensive screening in patients for not only cerebral structures but cardiovascular abnormalities as well.

Ethics

Informed Consent: Patient informed consent was taken.

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: A.A., S.T., A.B.A., T.Ö., M.K.T., Concept: A.A., S.T., A.B.A., T.Ö., M.K.T., Design: A.A., S.T., A.B.A., T.Ö., M.K.T., Data Collection or Processing: A.A., S.T., A.B.A., T.Ö., M.K.T., Design: A.A.,

S.T., A.B.A., T.Ö., M.K.T., Analysis or Interpretation: A.A., S.T., A.B.A., T.Ö., M.K.T., Design: A.A., S.T., A.B.A., T.Ö., M.K.T., Literature Search: A.A., S.T., A.B.A., T.Ö., M.K.T., Design: A.A., S.T., A.B.A., T.Ö., M.K.T., Writing: A.A., S.T., A.B.A., T.Ö., M.K.T., Design: A.A., S.T., A.B.A., T.Ö., M.K.T.

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