Case Report

Dandy-Walker Variant Associated with Bilateral Congenital Cataract

Abstract

Dandy-Walker Syndrome (D-WS) is a rare disorder with an incidence of 1%–2% of all central nervous system anomalies. The diagnosis can be challenging, especially in the prenatal period. Here, we present an extremely rare case of D-WS associated with bilateral congenital cataracts. A 36 weeks and 6 days old male baby presented with a Dandy-Walker variant associated with bilateral congenital cataract. Ophthalmological examination revealed microphthalmos and congenital cataracts present in both eyes with sclerocornea, iris coloboma, and zone 3 retinopathy of prematurity involving only the right eye. However, the right eye was salvageable. Skull transillumination was negative with no cranial bruit. He was admitted to the neonatal intensive care unit with breathing difficulties, maintained SpO_2 with oxygen through prongs, and noninvasive continuous positive airway pressure for 7 days. He had two episodes of hypoglycemia with hypothermia. There was no significant finding in sepsis evaluation. The abdominal ultrasonography was normal. Echocardiogram was suggestive of patent foramen ovale. Mother's torch panel tested positive for cytomegalovirus immunoglobulin G antibodies. Magnetic resonance imaging brain suggested variant D-WS with dilation of cerebellar fossa and occipital lateral ventricle horn and lack of usual corpus callus structure. Intravenous antibiotics cefotaxime and amikacin were administered along with fluid supplementation. He was shifted to mother feed. The neonate was referred to the pediatric surgery department for further management.

Keywords: *Congenital, Dandy-Walker syndrome, newborn*

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Introduction

Dandy and Blackfan first described Dandy-Walker syndrome (D-WS). It is a congenital brain abnormality that most often affects the fourth ventricle and cerebellum.[1] The Dandy-Walker variant (D-WV) is an extremely rare disorder. The D-WV is described as cerebellar dysgenesis with or without the posterior fossa enlargement and varying cerebellar vermis hypoplasia.[2] The estimated incidence of D-WV is about 1 in 35,000 live births.[3] The inferior vermis is hypoplastic in D-WV, but the fourth ventricle and cisterna magna remain normal in size. Hydrocephalus occurs in about 70%–80% of the cases.[4] It is characterized by the deviation of the inferior cerebellar vermis and communication between a normal-sized cisterna magna and the fourth ventricle.[5] The ultrasonography procedure uses the foramen magnum to help describe the anatomy of the neonatal posterior fossa more precisely. However, this method requires the addition of a diagnostic technique.[6] Prenatal diagnosis of fetal D-WS is incredibly challenging for radiologists, who mainly rely on ultrasonography. Ultrasound during pregnancy can diagnose classic cases of this syndrome. Magnetic resonance imaging (MRI) is preferred use to make a postnatal diagnosis.[7] We reported 36 weeks of old male baby with D-WS, associated with bilateral congenital cataract in the tertiary teaching hospital.

Case Report

A late preterm (36 weeks 5 days) male baby was borne to a primigravida mother by emergency lower segment cesarean section with fetal bradycardia. The baby was conceived through *in vitro* fertilization after 8 years of marriage due to the mother's primary infertility. Mother was 28 years old, diagnosed with genital tuberculosis, having undergone 6 months of antitubercular treatment before pregnancy. She received oral antihypertensive (labetalol 100 mg BD) due to pregnancy-induced hypertension. The mother's quadruple screen was negative, but the antenatal toxoplasmosis,

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rubella cytomegalovirus (CMV), herpes simplex, and HIV screening was not done. Mother had consumed iron and folic acid during pregnancy. At 33 weeks of the gestational age, prenatal sonography showed polyhydramnios with bilateral fetal kidneys displaying 3 mm pyelectasis, intrauterine dichorionic triplets with one viable fetus with 34 mm × 15 mm subchorionic hematoma, and no apparent congenital fetal malformations. No significant family history of consanguinity or congenital malformation. The baby cried immediately after birth. The baby's weight was 2.2 kg, length 52 cm, and head diameter was 33.5 cm. The neonate was dolichocephalic, the anterior and posterior fontanel was wide-open, sutures were broadly spaced, occiput was conspicuous, low-set ears, and the absence of right-side testis.

Ophthalmological examination revealed microphthalmos and congenital cataracts present in both eyes with sclerocornea, iris coloboma, and zone 3 retinopathy of prematurity involving only the right eye. However, the right eye was salvageable [Figures 1 and 2]. Skull transillumination was negative with no cranial bruit. Systemic examination was normal. Because of breathing difficulties, the neonate was admitted to the neonatal intensive care unit, holding on to O_2 for seven days through prongs. Gradually spoon feeds started, neonate with tachypnea (respiratory rate = 70 beats/min), holding on O_2 by blender continuous positive airway pressure, retaining SpO_2-96%. He had two episodes of hypoglycemia with hypothermia. There was no significant finding in sepsis evaluation. The abdominal ultrasonography was normal. Echocardiogram was suggestive of patent foramen ovale. Mother's torch panel tested positive for CMV immunoglobulin G antibodies. MRI brain suggested variant D-WS with dilation of cerebellar fossa and occipital lateral ventricle horn and lack of usual corpus callus structure (colpocephaly) [Figure 3]. Intravenous antibiotics cefotaxime and amikacin were administered along with fluid supplementation. He was shifted to mother feed. The infant was referred to the Pediatric Surgery Department for further management.

Discussion

D-WS is a neuropathophysiological process defined by a posterior fossa cyst, hydrocephalus, and vermian cerebellar hypoplasia.[8] Sutton first published D-WS in 1887, followed by Dandy and Blackfan in 1914. D-WS is more common in females than males.[9] D-WV is described by cystic dilatation of the fourth ventricle and hypoplasia of the inferior vermis in the absence of posterior fossa enlargement.[10] Dandy-Walker malformations and related variants affect an estimated one in every 35,000 live births. These malformations account for between 1% and 4% of the cases of hydrocephalus.[3,4] Nevertheless, others may be caused by chromosomal aneuploidy, Mendelian defects, or environmental exposures, such as congenital rubella or fetal alcohol exposure.[11]

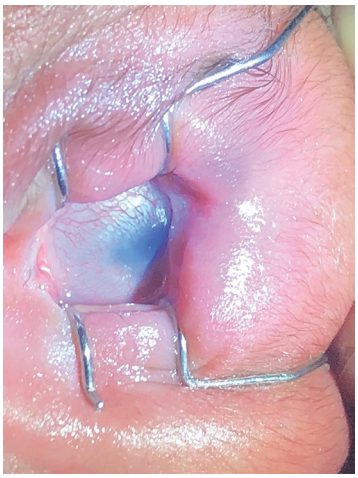

Figure 1: Ophthalmological examination revealed left congenital cataract and microphthalmos

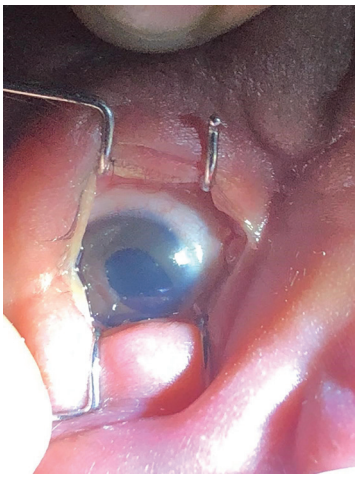

Figure 2: Ophthalmological examination revealed right congenital cataract, colboma of iris, sclerocornea and microphthalmos

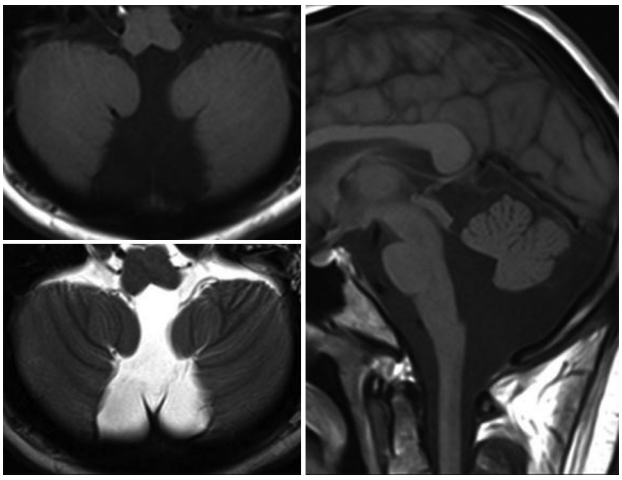

Figure 3: TIW MRI brain suggestive of variant of dandy walker syndrome

In 20% of instances, ophthalmologic manifestations occur. There are many types of vascular anomalies in the retina, including optic atrophy, iris artery hypertrophy,

iris hypoplasia, morning glory disc, congenital cataract, sclerocornea, lens coloboma, and exophthalmos.[12] D-WS is correlated with other anomalies; corpus callosum agenesis and other organ malformations, in 20-30% of the cases such as heart, lung, nasal, digital, or vertebral abnormalities.[13]

Ultrasonography may be used to diagnose hydrocephalus malformations during the prenatal period.[13] However, a definitive diagnosis of the D-WV and Mega cisterna Magna before 18 weeks of gestation is challenging.[14] It can be confirmed after the baby's birth through a computed tomography scan and MRI of the brain.[15]

Hydrocephalus treatment with shunting significantly decreases death rates in newborns. The existence of extra-central nervous system defects has a direct correlation with fetal mortality.[16,17] Medical management is very important before shifting to the patient for a surgical procedure.

Conclusion

Certain associated congenital disabilities are "simple repairs" that can correct quickly; others include staged or multiple surgical operations, and delays in recovery can result in lifelong disease. When dealing with D-WS, it is important to provide a well-coordinated consultation approach by pediatricians, pediatric neurosurgeons, and medical counselors.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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