Case Report

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Dandy Walker variant with agenesis of corpus callosum diagnosed late prenatally by foetal ultrasound: a case report

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Introduction and importance: Dandy Walker variant is an intracranial disorder involving variable hypoplasia of cerebellar vermis without posterior fossa enlargement. An anomaly scan performed at mid second trimester has good sensitivity and specificity for detecting foetal congenital anomalies. Despite that, some cases like the authors' might go undiagnosed due to normal biometric parameters for that gestational age and may be detected later in intrauterine life.

Case presentation: A primi-gravid mother underwent sonographic evaluation at 20 + 4 weeks of gestation that revealed all foetal parameters within normal limits. Only at 31 + 2 weeks of gestation, a posterior fossa cyst communicating with forth ventricle was detected. Foetal MRI done at 8 days of life (DOL), confirmed these findings and diagnosis of Dandy Walker variant with agenesis of corpus callosum was made.

Clinical discussion: Although the chances of a CNS anomaly is exceedingly low when foetal metrics like head circumference, atrial width and Cisterna Magna are within normal limits, some cases like the authors' may develop anomalies later in the intrauterine life which may lead to delayed diagnosis of the cases. Thorough performance of anomaly scan involving a multiplanar approach may help in prompt diagnosis of foetal anomalies.

Conclusion: The risks of development of posterior fossa anomalies can exist even after second trimester scan, Clinicians should be aware of this possibility and assess the posterior fossa at repeat scans done later in intrauterine life. Early diagnosis can provide an option to couples of the termination of pregnancy which is complicated when detected later in the intrauterine life.

Keywords: case report, corpus callosum, dandy walker variant, dandy walker, ultrasonography

Introduction

The Dandy Walker complex includes a group of congenital intracranial disorders: Dandy Walker malformation, Dandy Walker variant and Mega Cisterna Magna[1]. These disorders include variable hypoplasia of cerebellar vermis with or without posterior fossa enlargement. Dandy Walker variant mostly involves partial hypoplasia of cerebellar vermis and a variable degree of obstruction to the forth ventricle, without posterior fossa enlargement[2].

The reported incidence of Dandy Walker malformation and its variants is 1 in 35 000 live births[3]. Dandy Walker variants are frequently associated with other congenital anomalies, bringing about corresponding cardiac, neurological, gastrointestinal or genitourinary complications in the children born to the defect[4]. Agenesis of corpus callosum is such associated congenital anomaly that occurs in about 20% cases of Dandy Walker variants and may result in intellectual impairment of such children[4–6]. Due to uncertain prognosis of an infant born with Dandy Walker variant, its prenatal recognition is necessary[7].

We present a case of an infant with Dandy Walker variant who was under regular antenatal check-ups and no significant findings were present on anomaly scan. But, late at 31 + 2 weeks of gestation, a deformity of Dandy Walker spectrum was suspected which was later confirmed by foetal MRI. This case report is in line with CARE guidelines[8].

HIGHLIGHTS

- Dandy Walker variant is a rare intracranial disorder involving variable hypoplasia of cerebellar vermis without posterior fossa enlargement.
- Many developmental anomalies can be detected through an anomaly scan performed at second trimester; however, high resolution ultrasound, proper guidelines and stringent practice are necessary to avoid missing detectable cases.
- Due to uncertain prognosis of an infant born with Dandy Walker variant, its prenatal recognition is necessary.
- Children with Dandy Walker variants may suffer from a variable degree of disability, thus a multidisciplinary approach is essential for management.

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Case presentation

A 26-years-old primi-gravida with no significant past medical and surgical history presented to the Gynae-Obs OPD for regular antenatal check-up during the third trimester. In the ultrasonography (USG) scan, a single live intrauterine foetus corresponding to 31 + 2 weeks of gestation FW: 1823 gm, cephalic presentation, cardiac activity (144 bpm), umbilical artery systolic/diastolic ratio = 1.9 (forward diastolic flow) and AFI = 8.6 cm was seen. Notably, there was an anechoic collection with enlarged retro cerebellar cerebrospinal fluid space (maximum diameter = 16 mm) in the posterior fossa of the brain which was communicating with the fourth ventricle. However, hydrocephalus was not detected. Based on these sonographic findings, a provisional diagnosis of Dandy Walker spectrum disorder was made. This was later confirmed by an MRI of the brain which was performed 9 days after birth.

Diagnostic assessment

The USG scan done in the third trimester showed the aforementioned anomaly. However, an anomaly scan done prior; routinely at 20 + 4 weeks of gestation didn't show any impression. The biparietal diameter (BPD) and femur length (FL) corresponded to the gestational age. Cerebellar hemispheres, trans-cerebellar diameter, cisterna magna, nuchal fold thickness were assessed under axial view of the trans-cerebellar plane. Trans-cerebellar diameter was found to be 18.3 mm, Cisterna Magna measured 6.3 mm, and Nuchal fold thickness was 2.9 mm which were normal for the gestational age. Cerebellar hemispheres were also appreciated as normal.

A repeat scan performed at 31 + 5 weeks of gestation was consistent with that of latest scan, anechoic collection indicating posterior fossa cyst measuring 3.4 × 1.7 cm in posterior fossa, communicating with the fourth ventricle. MRI of the brain, done at 8 days of delivery (DOL), showed slightly dilated and parallely oriented bilateral lateral ventricles, high riding third ventricle communicating with the subarachnoid space. Stenosis of the proximal part of the aqueduct was noted, which warranted the patient for development of Hydrocephalus. Prominent cisterna Magna was visualized. Moreover, corpus callosum was not visualized. (Fig. 1) Hypoplastic inferior cerebellar vermis was noted. (Fig. 2) In light of these radiological evidences, the case was diagnosed as Dandy Walker variant with concomitant agenesis of corpus callosum.

Management

Since no evidence of hydrocephalus and any other indications requiring a CS were found, the baby was planned for a normal vaginal delivery. The patient presented to the ER with labour pain at 39 + 3 weeks of gestation. A thick meconium stained liquor was seen during vaginal delivery. The baby didn't cry after birth. APGAR scores assessed at 1 and 5 min were 5/10 and 7/10, respectively. Positioning and stimulation, suctioning, bag and mask ventilation were done in an attempt to provoke a cry in the newborn and a weak cry was evident. The baby was then shifted to the NICU.

Following initial resuscitation and stabilization, the neonate was closely monitored in the neonatal NICU. Three episodes of seizures occurred within 24 h after birth in form of jerky movements of all the limbs. Injection Midazolam 0.4 mg IV stat was given, but seizures were persistent. The seizures subsided only when Injection Levetiracetam 60 mg IV was given over the duration of 20 min.

Long-term follow-up and neurodevelopmental assessments were planned to evaluate the neonate's progress and identify any

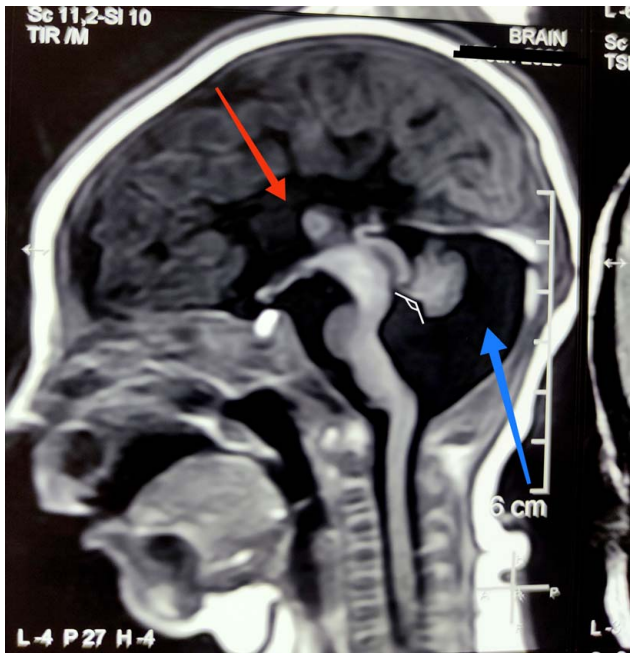

Figure 1. T1 weighted Magnetic Resonance Imaging (MRI), sagittal view of the brain; red arrow designating absence of corpus callosum, the angle showing increased angle of fastigium and blue arrow showing a prominent cisterna magna without posterior fossa enlargement.

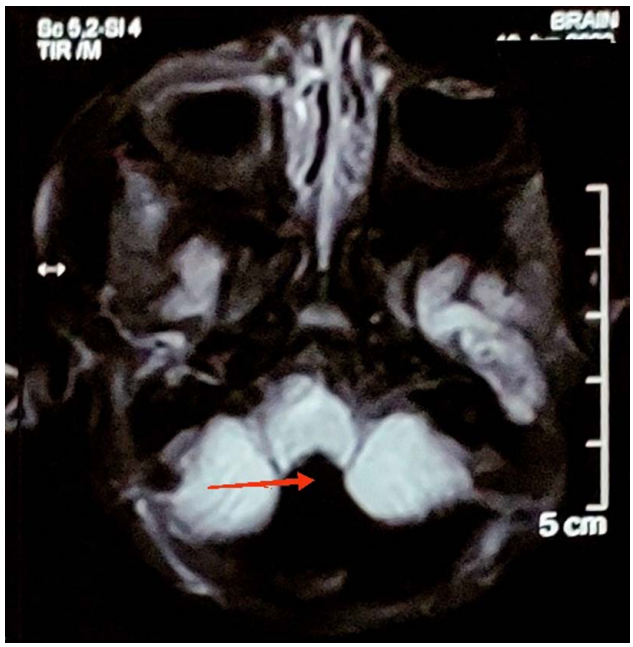

Figure 2. T1-weighted MRI, axial view; red arrow showing hypoplastic inferior cerebellar vermis.

developmental delays or neurological deficits associated with Dandy Walker Malformation and agenesis of the corpus callosum. Neurologic consultation reported that it was extremely likely that the patient would have special needs; with an unknown range of disability. Multidisciplinary care involving neonatology, neurology, and paediatric rehabilitation would be essential in providing optimal management and support for the neonate's neurodevelopmental needs. Ultimately, the neonate was discharged from the hospital with a follow-up schedule, medications and appropriate counselling to the parents.

Follow-up

The patient is currently under follow-up every month at our institution. As of the most recent follow-up, a delay in developmental milestones including uttering words, rolling etc. has been noted. However, there is no visible macrocephaly as of now.

Discussion

Dandy Walker variant falls under a rare group of intracranial disorders, the Dandy Walker complex. They involve variable hypoplasia of cerebellar vermis with or without posterior fossa enlargement[2].

Since the 1990s, ultrasound scan has been a key radiological tool for performing antenatal scans. Hu and colleagues in their cohort study found that detection rate of CNS anomalies in first, second, third and late third trimester were 32%, 22%, 25% and 16%, respectively. These statistics suggest a crucial role of routine USG scans for detection of foetal anomalies[9]. The IUSOG guidelines recommends a transabdominal examination to be performed in two planes in routine practice; the transventricular plane and the transcrebellar plane. According to the guidelines, when satisfactory results are obtained under sonographic evaluation in measurements of head circumference, atrial width and cisterna magna, the risk of CNS anomaly is exceedingly low and no further examinations are indicated[10]. Our case is a low risk pregnancy and at 20 + 4 weeks of gestation, trans-cerebellar diameter of 18.3 mm, a normal atrial width and span of cisterna magna 6.3 mm were found which were within normal limits for that gestational age. Thus, it was not possible to establish a suspicion of a posterior fossa anomaly at that stage. WHO recommends at least 8 antenatal visits during pregnancy. Due to limitations in accessibility of resources, the number of visits are often inadequate. Following routine anomaly scan, the patient reported to the institution at 31 + 2 weeks of gestation where the posterior fossa anomaly was detected. At this point, the gestational age was deemed too advanced to contemplate termination of pregnancy. USG scans were performed by the same radiologist at both instances.

Leung and colleagues recommend in their paper the minimal requirements of a routine mid-trimester anomaly scan. The minimal biometric parameters recommended are biparietal diameter (BPD), head circumference (HC), abdominal circumference (AC), and femur length (FL). In the anatomy of the head, the following are to be assessed: Intact cranium cavum septi pellucidi, Midline falx thalami, cerebral ventricles and cerebellum, cisterna magna. The data regarding sensitivity and specificity of an ultrasound scan depends upon various parameters including medical practice, guidelines, local circumstances, experience of sonographers and resolution of ultrasound machines[11]. Akinmoladun *et al.*[12] report a 93.5% specificity of prenatal ultrasound anomaly screening program. Leiroz *et al.*[13] in a similar study report sensitivity and specificity of the scan to be 61.1% and 96.3%, respectively. These data suggest a good specificity but fair sensitivity.Hu *et al.*[9] also suggest that 5% of CNS anomalies are missed even after routine ultrasound scans. Improvement on aforesaid parameters may help improve the sensitivity as well as specificity of ultrasound scan.

Sasaki-Adams and colleagues report in their case series of 24 patients that 41.7% of patients had cardiac anomalies, 33.3% had neurological, 20.8% had gastrointestinal and some had orthopaedic anomalies. Three out of 24 patients died. Similarly, Stambolliu *et al.*[14] in their systematic review of 187 cases reported the cases to have chromosal abnormalities, cardiac deformities and diseases of ear and eye. These large studies suggest that the children born to the defect are prone to many anomalies. In our case, during a 6 month follow-up, the child is reported to have delayed developmental milestones. The child is not able to roll on his own at this age. The mother reports the child being unable to utter words or syllables. These developmental delays may be due to the absence of corpus callosum in the patient. However, there is no finding suggestive of hydrocephalus yet. The MRI scan done at 8 DOL suggested stenosis of aqueduct, thus the child is expected to have macrocephaly later in the life.

Has *et al.*[15] report in their review of 78 cases that a high proportion of parents were in a consanguineous marriage (44.8%); however, our patient is the first born child arising out of non-consanguineous marriage. Sun *et al.*[16] showed that DWS was associated with chromosomal aberrations, especially aneuploidy. Many case reports of DWS with complications of chromosomal abnormalities have been published in the past. However, only ZIC1 and ZIC4 genes of chromosome 3q24 have been identified as candidate genes in recent years[17]. Prenatal genetic testing and karyotype analysis is recommended but was not done in our case.

When a posterior fossa cyst is detected, absence or hypoplasia of the cerebellar vermis is a key finding for distinguishing between a Dandy Walker malformation and an arachnoid cyst. Moreover, for the differential diagnosis, we should bear in mind that Dandy Walker malformation it is usually accompanied by other intracranial anomalies like agenesis of the corpus callosum[18]. Our case had partial hypoplasia of the inferior cerebellar vermis with concomitant agenesis of corpus callosum. A MRI scan was performed to confirm the diagnosis of our case.

Surgical option for management of hydrocephalus remains ventricular drainage and/or posterior fossa drainage[19]. As aqueductal stenosis was present on MRI scan of the brain, an impending hydrocephalus has been anticipated which will be managed by a multidisciplinary approach. As of now, the parents of the child have been counselled at follow-up sessions regarding the proper care of the infant.

Conclusion

In conclusion, Dandy Walker variants should be diagnosed prenatally by a foetal ultrasound at anomaly scan. Multiplanar evaluation are essential to be performed at routine practice for a detailed evaluation to rule out such anomalies, facilitate its early diagnosis and possibly provide the couple with an option to terminate the pregnancy.

Ethical approval

Not applicable.

Patient's perspective

The patient party were anxious when they were first communicated about the foetal condition given that the anomaly scan of the foetus was reported to be normal. With proper counselling, the parents have now accepted the situation.

Consent

Written informed consent was obtained from the patient's informant for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor- in- Chief of this journal on request.

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Author contribution

S.P.: conceptualization, visualization, project administration, supervision, writing—original draft, writing—review and editing. S.K.P.: conceptualization, visualization, project administration, writing—original draft, writing—review and editing. R.S.: visualization, project administration, investigation, writing-review and editing. S.R.: resources, methodology, writing—original draft, writing—review and editing. R.Z.: visualization, resources, formal analysis, writing—review and editing.

Conflicts of interest disclosure

The author declares no conflict of interest.

Research registration unique identifying number (UIN)

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Data availability statement

Not applicable.

Provenance and peer review

Our paper was not invited.

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