Challenges in the pre- and post-natal diagnosis of posterior fossa cysts: A case report and review of historical evolution of descriptive terminologies

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ABSTRACT

Background: Radiological diagnoses of posterior fossa cystic abnormalities during antenatal and postnatal periods pose significant challenges as they may have similar early imaging features. Some of the frequently described entities are arachnoid cysts and Dandy-Walker malformations. Blake's pouch cyst is relatively underdiagnosed. The main aim of the study was to explore these diagnostic challenges in the context of various descriptive terminologies and their prognostic implications.

Methods: We illustrate this through our case, where fetal magnetic resonance imaging (MRI) at 36 weeks gestation showed small right cerebellum without hydrocephalus or hemorrhage. Possible differential diagnoses included Dandy-Walker malformation or posterior fossa malformations, facial hemangiomas, arterial anomalies, cardiac and eye anomalies, sternal clefting, and supraumbilical raphe.

Results: Postnatal sonography noted posterior fossa cyst without hydrocephalus in a normal term infant, who went on to develop symptomatic hydrocephalus by 15 weeks. Computed tomography brain scan confirmed large subtentorial posterior fossa cyst and extensive internal hydrocephalus. Despite emergent ventriculoperitoneal shunt insertion, head circumference continued to rise. MRI scan showed persistent cyst. Subsequently, infant underwent endoscopic fenestration of the cyst with balloon septostomy and now has an age appropriate developmental profile.

Conclusion: There is considerable discordance between antenatal and postnatal neuroimaging findings as highlighted in our case. Diagnostic conundrum here was whether this was an arachnoid or Blake’s pouch cyst. Differentiating between posterior fossa fluid collections is crucial for management, prognosis, and parental counseling. Close postnatal follow-up is essential to avert complications due to acute hydrocephalus.

Keywords: Arachnoid cyst, Blake pouch cyst, Hydrocephalus, Posterior fossa, Retrocerebellar cyst

INTRODUCTION

A range of fetal posterior fossa malformations (PFM) has been reported in one in 5000 live births. [20] The posterior fossa is located between the tentorium cerebelli superiorly and foramen magnum inferiorly. Cerebellum is one of the first central nervous system structure to form, but the last one to attain maturity and linked to brain stem development. It is susceptible to many insults because of this wide developmental period. Both these structures develop from the mesencephalic-metencephalic complex. The roof plate of metencephalon forms the cerebellum...
and the floor plate becomes pons. The roof the fourth ventricle is gradually closed by the developing vermis and is complete by 18 weeks of gestation.[18]

In recent times, advances in neuroimaging techniques, genetic sequencing, and etiological research have led to precise definition and better classification of congenital abnormalities of posterior fossa.[2] The most frequently described entities include arachnoid and retrocerebellar cysts, mega cisterna magna (MCM), Dandy-Walker malformation (DWM), Dandy-Walker variant, and inferior vermian hypoplasia (IVH). There are infrequent reports of Blake pouch cysts (BPCs) as this is underdiagnosed. Clinicians are less aware of its existence as a distinct entity among the posterior fossa cystic abnormalities.[8] All of the above can become differential diagnoses of each other as they may all include varying degrees of abnormalities of cerebellar vermis, subarachnoid cisterns, cerebellar hemispheres, and surrounding meningeal structures.

Doherty et al.[9] created a practical diagnostic classification based on neuroimaging, which can provide information with high clinical relevance. A precise diagnosis helps to determine the inheritance pattern, risk of recurrence of a particular anomaly, involvement of other organs such as liver and kidney, and developmental prognosis. Malinger et al.[14] classification of cystic and noncystic PFM is a widely used multiplanar evaluation of posterior fossa structures. The term Dandy-Walker variant is absent in this format which certainly eliminates some diagnostic and prognostic challenges. Through the presentation of our case, we highlight this ongoing diagnostic uncertainty and related issues.

CASE DESCRIPTION

A 29-year-old woman had an obstetric anomaly scan at 19+4 weeks of gestation, which noted a low posterior placenta and recalled for a review scan. Following the detection of a posterior fossa anomaly, the patient was referred to fetal medicine specialists for further evaluation with an magnetic resonance imaging (MRI) scan and antenatal counseling. Pregnancy was otherwise unremarkable with no other significant family history.

In the fetal MRI scan at 36 weeks gestation, the right cerebellar hemisphere appeared small with prominence of extra axial fluid. Transcerebellar diameter (TCD) was 38 mm (<1st centile). There was suspected mild vermis dysmorphism with a height of 28 mm (>99th centile). The lateral, third, and fourth ventricles were normal in size and shape with normal cavum septum pellucidum. Major intracranial vascular flow voids were grossly unremarkable with appropriate myelination. There was no evidence of acute or old hemorrhage [Figure 1]. Suggested etiological differentials included mild cerebellar hypoplasia and possible mild vermian abnormality with prominent extra-axial fluid spaces overlying right cerebellum. This could be clastic or syndromic in association such as PFM, facial hemangiomias, arterial anomalies, cardiac and eye anomalies, sternal clefting, and supraumbilical raphe. The other less likely diagnosis would be a large arachnoid cyst compressing the right cerebellum.

Parents were counseled regarding various possible outcomes as the differential diagnoses were unclear based on the available information from the fetal MRI scan. Fetal karyotyping was normal. A female infant was born by normal delivery at 39+2 weeks gestation. Birth weight was 3070 g (9–25th percentile) and occipitofrontal circumference (OFC) was 34 cm (50th percentile). The infant had no dysmorphic features and had a normal neurological examination. Figure 2 shows cranial ultrasound scan on day 4 of life. The fourth ventricle appeared satisfactory and appearances were not in keeping with DWM. The suggested diagnoses were retrocerebellar arachnoid cyst and/or an MCM and MRI brain scan was planned when clinically appropriate.

An outpatient follow-up appointment was scheduled in 8 weeks, with health visitor monitoring in the interim. COVID-19 pandemic restrictions during that period impacted follow-up. In retrospect, there were no parental concerns until about 12 weeks of age when they began to notice large head. The infant was then referred to the hospital with increasing head circumference at 15 weeks of age. OFC was 48.8 cm (99.6th percentile) with bulging fontanelle and sun setting

**Figure 1:** Fetal magnetic resonance imaging coronal 1 image showing large fluid signal intensity structure occupying much of the right side of posterior fossa with the displacement of the right cerebellum.
appearance of eyes. The infant was otherwise clinically and hemodynamically stable. Figures 3 and 4 show computed tomography brain appearances which were suggestive of congenital posterior fossa arachnoid cyst causing compression of adjacent structures with extensive internal hydrocephalus. Maximal transverse diameter of third ventricle was 32 mm. Following transfer to neurosurgical unit, the infant had an emergency right frontal ventriculoperitoneal (VP) shunt insertion and Bactiseal ventricular catheter. The infant did not have an MRI brain scan before this surgery. In hindsight, given the importance of the cyst configuration in relation to the hydrocephalus, this would have been informative. OFC continues to track above the 99.6th percentile. Follow-up MRI brain and cervical cord scan 4 weeks later noted only mild decompression of the supratentorial ventricular system with VP shunt in place [Figures 5 and 6].

The neurosurgical team carried out an endoscopic fenestration of the cyst, which likely grew and secondarily extended to an area near the pineal region. The previous burr hole was extended laterally after disconnection of the reservoir and ventricular catheter. A 6 mm endoscope was introduced after corticotomy and lateral ventricle was visualized. The large cyst was identified at the foramen of Munroe, which was fenestrated with scissors and balloon septostomy. The ventricular catheter and reservoir were replaced. Postoperatively, there was some resolution of up gaze palsy. Another MRI scan a month later showed the persisting large posterior fossa cyst with new bilateral subdural collections overlying both cerebral hemispheres. This might relate to over shunting of cerebrospinal fluid (CSF) [Figure 7]. There could be some difference of opinion as to the necessity of the VP shunt or whether endoscopic fenestration with or without a ventricular access device would have been more appropriate in this case.

The infant’s OFC is tracking above the 99.6th percentile and she has minimal motor skills delay and up gaze palsy. Otherwise, her developmental profile is age appropriate.

Figure 2: Cranial ultrasound image with fluid echogenicity structure occupying most of the right side of posterior fossa with a compressed right cerebellum.

Figure 3: Computed tomography brain coronal image showing dilated lateral and third ventricles and hydrocephalus.

Figure 4: Computed tomography brain sagittal showing that the posterior fossa fluid density structure has enlarged and fourth ventricle is now compressed. Findings consistent with new obstructive noncommunicating hydrocephalus secondary to an enlarging cystic lesion.
DISCUSSION

Our case describes the clinical course of a prenatally detected posterior fossa cyst and its postnatal outcome. Although some sinister pathologies were discussed during counseling, none of these was confirmed later. In this context, we elaborate on the commonly encountered PF cysts and the role of neuroimaging as an invaluable tool in identification as well as prognostication.

Imaging modalities

Ultrasonography is the best initial imaging modality to evaluate fetal posterior fossa with fetal MRI providing excellent additional diagnostic clarity.[7] Fetal MRI with midline T1 or T2W sequences can measure the size of the posterior fossa, the shape and size of vermis, the morphology of fourth ventricle, and brainstem. Fetal MRI has a sensitivity and specificity of 85–100% if performed after 20 weeks.[12,27] It is important to know that the roof of the fourth ventricle is not visible in the fetal MRI.[2] TCD and vermian height are important biometric markers for cerebellar assessment.[3]

COMMON DIFFERENTIAL DIAGNOSES OF POSTERIOR FOSSA CYSTS

DWM

DWM is the most common PWM with sporadic occurrence and low future recurrence risk of 1–5%.[9] It can be associated with hypoplastic left heart syndrome, tetralogy of Fallot, renal anomalies, and short limbs. Such systemic involvement has a poor outcome.[9] The key neuroimaging features of DWM are (1) hypoplasia or agenesis of cerebellar vermis, which is elevated and upwardly rotated, (2) dilatation of cystic-appearing fourth ventricle, which may fill the entire posterior fossa, and (3) high torcular (upper posterior fossa perimeter). This structure is not to be seen as posterior...
fossa subarachnoid space or mistaken for an arachnoid cyst. In addition, the cerebellar hemispheres are typically displaced anterolaterally with a normal morphology and size. Hydrocephalus is present in 90% of cases of DWM and may appear before birth or late with different prognostic value.[20] Many times cases diagnosed as DWM are due to disruptive process rather than a true malformation and are depicted as “variant, complex, or spectrum.” Now, these terms are considered nonspecific and it is preferable to use better anatomic descriptors of the abnormal findings.[22] Although these features were not present in the fetal MRI of our case, this was not explicitly stated. Postnatal sonography ruled out a DWM, which enabled the clinician to project a more favorable outlook regarding prognosis to the family.

MCM

MCM can be an incidental finding where the cisterna magna measures more than 10 mm on midsagittal images. This measurement is arbitrary as some consider it up to 12 mm as normal.[4,26] The vermis and fourth ventricle are normal with focal enlargement of subarachnoid space in posterior and inferior areas of posterior fossa. MCM freely communicates with the fourth ventricle and cervical subarachnoid space, so there is no hydrocephalus. Even though MCM causes less mass effect, it may be difficult to differentiate it from a retrocerebellar arachnoid cyst. This has no recurrence risk in subsequent pregnancies and has normal neurodevelopmental outcome.[4,26] MCM was also cited as a possible differential in the postnatal ultrasound scan of our patient.

Arachnoid cyst

When CSF is trapped between duplicated layers of the arachnoid membrane, it forms a fluid-filled cyst called arachnoid cyst. The location can be in the posterior fossa in about 10% of cases in children. They can be retro or lateral cerebellar, above vermis, or anterior to the brain stem.[1] The cysts are usually well-defined extra-axial cyst or fluid collection. Neuroimaging shows them as isointense relative to CSF in all sequences. Arachnoid cysts do not communicate with the fourth ventricle or the subarachnoid space, but it can cause mass effect on adjacent structures as either displacement or flattening. The fourth ventricle may look flattened or effaced. There are no recurrence risk or associated anomalies. If arachnoid cysts are detected after the neonatal period, sometimes scalloping of the occipital bone can be seen. An arachnoid cyst in the posterior fossa with obstruction of CSF flow can present with macrocephaly, signs and symptoms of raised intracranial pressure, and developmental impairment. They can be incidental findings that remain asymptomatic.[6,13] Based on the fetal MRI findings, an arachnoid cyst was considered as a least likely diagnosis in our case, but not so in the subsequent neuroimaging.

Isolated IVH

In isolated IVH, a small vermis partially covers the fourth ventricle. Some authors refer this as Dandy-Walker variant, which is problematic for accurate genetic counseling. In isolation, this has no recurrence risk and has a favorable prognosis in majority of cases. It is worth noting that if this occurs with other genetic conditions such as Joubert syndrome, there is a likelihood of recurrence. Problems with fine motor and receptive language skills are reported with vermian hypoplasia. Prenatal diagnosis is possible after 18–20 weeks of gestation, although ultrasound diagnosis is unreliable. Fetal MRI can delineate partial absence of inferior vermis. Rest of the vermis is normal along with normal cerebellar hemispheres and fourth ventricle. Posterior fossa anatomy and size are otherwise normal.[13,25]

BPC

Blake's pouch is a normal embryonic structure, which arises from the posterior membranous area on the roof of the primitive rhombencephalon. This area enlarges around 4–5th week of gestation and directly communicates with the fourth ventricle. Later, the Blake's pouch fenestrates to form the foramina of Luschka and Magendie, which is vital for normal CSF flow in the ventricles.[8] The smaller foramina of Luschka is formed by the 4th month of gestation after the formation of larger foramen of Magendie. When there is nonperforation of the latter, Blake's pouch does not regress, leading to enlargement of the fourth ventricle and supratentorial ventricular system. Later, when the foramen of Luschka opens, some equilibrium of CSF flow from ventricles is established. As this opening is much smaller than that of foramina of Magendie, ventricles remain enlarged with compression. Therefore, this does not cause underdevelopment of cerebellum and vermis.[16,17] BPC happens sporadically and can undergo spontaneous resolution in utero and postnatally, with no recurrence risk in subsequent conceptions.[18,19]

The radiographic findings of BPC include retrocerebellar position of the cyst, morphologically normal vermis, cystic dilatation of the fourth ventricle, and some degree of compression of cerebellum.[4,17,26] Neonates can present with ventriculomegaly and hydrocephalus with persistent mass effect needing decompression. Prognosis is good in the absence of complications related to neurosurgery.[4,17]

Endoscopic third ventriculostomy is an accepted form of treatment modality in symptomatic BPC.[4,26] Cyst fenestration alone may not be adequate to establish CSF pathway.[27] Diversionary shunt is sometimes required to treat obstructive hydrocephalus, although it has associated complications of infections or over drainage.

Table 1 summarizes the typical neuroimaging findings of common PF cysts described above.
**HISTORICAL EVOLUTION OF DESCRIPTION OF POSTERIOR FOSSA CYSTS**

The literature provides us with fascinating insights into the evolution of various descriptive terminologies for posterior fossa anomalies. For instance, MCM was thought to be due to cerebellar atrophy and the term was used to describe any retrocerebellar CSF space with normal vermis and cerebellum.\(^{[17,25]}\) Similarly, Blake's cyst was thought to be the same as retrocerebellar arachnoid cyst or even MCM.\(^{[20,23]}\) As more data emerged from MRI studies, many of these anomalies including DWM were lumped together as a continuum of developmental anomalies of the posterior fossa.\(^{[3]}\)

Cornips and colleagues\(^{[9]}\) were the first to specifically report on the clinical presentation and treatment of BPC. In their series of six patients, all had radiological evidence of tetraventricular hydrocephalus. Tortori-Donati\(^{[26]}\) defined persistent BPC as an independent entity within the DWM and gave a classification based on embryopathogenesis. There is debate about this conclusion of linking BPC with Dandy-Walker spectrum, as the fourth ventricle does not communicate with the subarachnoid spaces in the midline. A few held the opinion that BPC has no clinical significance unless it causes mass effect due to hydrocephalus or associated with other cerebral or cerebellar malformations.\(^{[8,26]}\)

Paladini et al.\(^{[19]}\) proposed sonographic criteria for prenatal diagnosis of BPC based on normal anatomy and size of vermis with some rotation and a normal cisterna magna. It is also possible in some cases to visualize the choroid plexus on the superolateral margin of the roof of the cyst. Nelson et al.\(^{[17]}\) noted similarity in appearance of BPC to arachnoid cysts. Like Paladini et al., they considered a diagnosis of persistent Blake's pouch when there is a displacement of the choroid plexus under the inferior surface of the vermis to extend along the superior cyst wall.

Studies by Blaicher et al.\(^{[6]}\) and Patel et al.\(^{[21]}\) appear to favor the concurrence of fetal and postnatal MRI, while Limperopoulos et al.\(^{[12]}\) disagree with this conclusion. In their 5-year series of PF cysts from a tertiary center, there was not a single case of Blake's cyst. Data collected from postnatal MRIs carried out between 1 week and 15 months of age showed complete agreement in only <60% of cases. In some cases, additional and more extensive findings were noted after retrospective evaluations as were in our case.\(^{[12]}\)

**Vermis and Ventriculomegaly**

Recently, there is increasing focus on accurate description of vermis when evaluating PF fluid collections in fetal and postnatal imaging. Moderately raised vermis can be due to vermian hypoplasia or DWM, or it could be due to persistent Blake pouch.\(^{[11,23]}\) In cases of small or flattened vermis, the term IVH was used. New embryological data show that vermis develops from ventral to dorsal rather than superior to inferior. This has led to many authors questioning this nomenclature and postulating that these were likely Blake pouch remnants.\(^{[23]}\) An infravermian BPC can result in massive ventriculomegaly with variable enlargement of the fourth ventricle. Nonrotation of vermis is also a clue to the diagnosis of BPC, where it can be differentiated from DWM. Similarly, an arachnoid cyst will not typically cause elevation of the tegmentovermian angle and does not freely communicate with the fourth ventricle.\(^{[8,10,28]}\)

The high level of persistent parental stress even with normal neurodevelopmental outcomes was a consistent finding in more than a third of cases in a study by Skreden et al.\(^{[24]}\) This emotional burden needs anticipatory guidance, support, and reiterates the importance of continued improved fetal diagnosis and better prognostication.

**CONCLUSION**

The inconsistencies in the classification of cerebellar and posterior fossa cystic anomalies are well documented in literature. Complex and protracted embryological development of cerebellum contributes to increased false positivity rate of many anomalies. There is increasing focus on accurate description of choroid plexus and vermis, especially in the setting of ventriculomegaly, based on current embryological data. In our case, like many other reported case series, there was inconsistent nomenclature.
with listing of broad differential diagnoses during prenatal period. There was adequate parental counseling with discussions on possible diagnoses and favorable and unfavorable developmental predictions. Even with retrospective evaluation, it could not be concluded whether this was a Blake's pouch cyst or a retrocerebellar arachnoid cyst requiring emergent neurosurgical intervention.

Prenatal diagnosis, outcome prognostication, and parental counseling are fraught with difficulties. Information from fetal MRI is a valuable aid as there is a wide variation in the neurological prognoses based on nature of the anomaly. Better understanding of the nuances of normal brain development along with improved tissue resolution of fetal MRI is essential to translate this to patient care. Deriving at an accurate diagnosis has significant clinical relevance and may have an impact on decisions regarding the course of the pregnancy and postnatal neurodevelopmental outcomes.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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