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Posterior fossa cyst made easy: Case discussion and simplified differential diagnosis

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Abstract
A 28 year old 2nd Gravida mother with 21 weeks of gestation was referred with a 20^{+2} weeks antenatal ultrasound report stating some cystic cranial malformation in fetus. No other structural abnormality noted.

Keywords: Posterior fossa, cystic cranial malformation, fetal CNS

Introduction

Posterior fossa fluid collection encompass panorama of lesions ranging from normal varients to malformations [1]. The common important lesions includes (1) Megacisterna magna, (2) Blake pouch cyst, (3) Vermian hypoplasia, (4) Dandy –Walker malformation, (5) Dandy-Walker Varient, (6) Joubert syndrome and (7) Arachnoid cyst [2]. With recent advances in fetal neurosonography, it is possible to suspect these lesion in the first trimester and reconfirm by a target ed scan in the second trimester.

Case Scenario

A 28 year old 2nd Gravida mother with 21 weeks of gestation was referred with a 20^{+2} weeks antenatal ultrasound report stating some cystic cranial malformation in fetus. No other structural abnormality noted.

Embryology

Fetal nervous system originates from ectodermal neural plate which forms neural tube. Enlargement of neural tube forms 3 primary vesicles (Prosencephalon, Mesenchephalon and Rhombencephalon) by 7-8 weeks and 5 secondary vesicles (Telencephalon, Diencephalon, Mesencephalon, Metencphalon, and Myelencephalon) by 9 weeks.[2] Floor of Rhombencephalon represent 4th ventricle. Tissue forming choroid plexus indent roof plate and divide it into anterior and posterior membranous area. Posterior membranous area expands caudally and forms Blake pouch cyst which later get fenestrated forming cistern magna which communicate freely with 4th ventricle and sub-arachnoid space [3]. Non- fenestration in the region of foramen of magendie leads to persistence of blake pouch cyst. Late fenestration of same results in megacisterna magna [4]. Cerebellar development starts at 4 weeks of gestation and it continue till 2nd year of life [5]. Fusion of cerebellar hemispheres happens craniocaudally forming cerebellar vermis at around 4-9 weeks [6]. The final development of vermis occurs between 16-19 weeks. Mostly cerebellar and vermian agenesis was related to malformative and disruptive causes and had association with mutation in PTF1A gene in chromosome 10 [7]. Joubert syndrome is due to defect in the gene controlling mesenchymal neuroepithelial signalling pathway affecting the decussation of superior cerebellar peduncles and giving the characteristic "Molar tooth appearance" in MRI brain [3, 8, 9]. Arachnoid cysts are benign leptomeningeal duplication cyst [5]. One third cases are reported in posterior fossa but there is no communication with ventricular system.

Diagnosing the posterior cysts

The basic views to evaluate posterior fossa are transcerebellar, sub-occipitobragmatic and saggital views [10].

Axial trancerebellar plane- it is at slightly lower level than transventricular plane and allows visualisation of frontal horns of lateral ventricle, cavum septum pellucidum, thalami, cerebellum, and cistern magna [10].

Coronal transcerebellar plane- also known as occipital plane, passes through posterior fontanel and allows visualisation of occipital horn of lateral ventricle, interhemispheric fissures, both cerebellar hemisphere and vermis [10].

Sub-occipitobragmatic view- allows visualisation of fourth ventricle and vermis [10].

Saggital plane- mid saggital plane allows visualisation of corpus callosum in its entirety, cavum septum pellucidum, cavum vergie, cavum septum interpositi, brain stem, pons, vermis, and posterior fossa [10]. Utilisation of saggital plane with transvaginal approach gives better results [11].

Significant associations

Dandy –Walker malformation

Hydrocephalus is associated with 55-60% of cases of DWS, and almost half may require shunting. The syndromes associated with DWS are Piere robin sequence, Smith-limli-opitz syndrome, Senior Loken syndrome, Meknes syndrome, Coffin-sitis syndrome, Ehler Danlos syndrome, and neurocutaneomelanosis. Gastrointestinal anomalies were observed in 21% cases. Its

association is noted with triploidy and trisomies 13 and 18 in 66% cases [12, 13]. Commonly associated genetic conditions are PHACE syndrome, Ritscher-Schnizel, Cranio-cerebello- cardiac, and Meckel Grueber [4, 14, 15]. Infections with causative association are Cytomegalovirus and Rubella. Commonly associated Cranial anomalies are aqueductal stenosis, Schizencephaly, brainstem dysplasia, dysgenesis of corpus callosum, lumbosacral meningocoele and cephalocele [16, 17]. Prevalance of chromosomal, Central nervous system (CNS) and extra-CNS anomalies are 31.3%, 60.9% and 42.6% [18].

Blake pouch cyst

Usually isolated finding, in the presence of other soft marker 5% association is reported with trisomy 21 [19].

Megacisterna magna

Mostly isolated finding, 10% cases has association with ventriculomegaly [19]. Rate of associated CNS and extra –CNS anomalies are 12.6% and 16.6% respectively [20].

Arachnoid cyst

Also an isolated finding, however few cases had association with trisomies 12, 18 and corpus callosum agenesis [19].

Joubert syndrome

It is having association with syndromic retinitis pigmentosa, Dekaban-Arima syndrome, COACH syndrome, Senior- Loken syndrome, Varadi – Papp syndrome and nephronopthisis [21, 22].

Antenal diagnosis and management

Table 1

Lesion	Prevalence	Ultrasound findings	Investigations	Follow up	Prognosis	Recurrence
Arachnoid cyst [23].	1 in 100	1. Eccentrically located cystic lesion displacing cerebellum laterally.	1. Fetal neurosonogram 2. Target scan for fetal anomaly	4weekly May -↑/↓in size. -Mode and delivery timing to be modified as per presence of hydrocephalous	Good Out of 12 cases followed upto 5 years of age only 1 had visual impairment and autism. [24].	Unknown
Blake pouch cyst [19].	1 in 1000	1.Normal CM,fourth ventricle and vermis 2.Mild vermian rotation with BV angle<30* 3. Key hole sign in transcerebellar view.	1.Fetal neurosonogram 2.Target anomaly scan 3. Chromosomal microarray is recommended in non-isolated cases.	4 weekly By 24-26 weeks, 50% cases had spontaneous resolution.	Normal neurodevelopment in 90% cases [25]. Abnormal cognitive outcome ranges 0-5%.[26]	No increased risk If associated with trisomies 1% risk.
Megacisterna magna [19].	1 in 5000	1.Transcerebellar view-CM>10mm 2. Vermis normal	1.Fetal neurosonogram 2.Target anomaly scan 3.Fetal MRI	4 Weekly	Normal neurodevelopmental outcome [20, 23]. Developmental delay was noted in 8-13.8% patients [26, 27].	No increased Risk
Joubert syndrome [21].	1 in 100,000	1. Agenesis of cerebellar vermis. 2. Batwing or umbrella type fourth ventricle. 3. Molar tooth appearance.	1.Fetal neurosonogram 2.Target anomaly scan 3.Molecular testing 4. Carrier screening	4 Weekly	Neurological impairment-noted in 100% as ataxia,hypotonia and oculomotor disturbances, 50% hyperactive, and 25% aggressive [28].	Autosomal recessive- 25% risk X-linked recessive- males are affected. 50% females are carrier.
Dandy-Walker malformation [23].	1in 30,000 4-12% cases of infantile hydrocephalo-us	1.Ventriculomegaly 2.↑ CM 3.Defect in cerebellar vermis 4.Elevated tentoriu,m 5. BV angle>45*	1.Fetal neurosonogram 2.Target anomaly scan	Terminat-ion of pregnanc-y if detected<20 weeks	Unfavourable 40%- die 58.2% had cognitive deficits and varied from 0-100%.[26] Incidence of motor delay was 30.4% [26].	1 -5% depending on etiology.
Vermian hypoplasia [29].	1 in 30,000	1. Open fourth ventricle. 2. Normal CM 3.BV angle -30-45*	1.Fetal neurosonogram 2.Target anomaly scan	4 Weekly	Mostly normal neurodevelopmental outcome [26]	1-5%

*BV angle – A line drawn along the dorsal aspect of brain stem and another line along ventral aspect of cerebellar vermis, the resulting angle is brainstem vermis BV angle.

First Trimester Evaluation of Posterior Fossa

-Anotomy of posterior fossa can be evaluated in 11-13+6 weeks scan. The reference points are fourth ventricle, cistern magna, and trans-cerebellar diameter and evaluation of intracranial translucency [19].

-Absence of choroid plexus of fourth ventricle in first trimester scan is suggestive of chromosomal defects and posterior fossa abnormalities. However, it is difficult to differentiate various posterior fossa lesion in first trimester, a detailed mid-trimester anomaly scan is recommended to establish the diagnosis [31].

Conclusion

With the evolving ultrasound techniques, visualisation and evaluation normal and abnormal fetal CNS is increasingly possible. Hence, ultrasound should be the first diagnostic modality for evaluation of fetal CNS. Those with CNS abnormalities suspected on first trimester scan should undergo detailed anomaly scan in second trimester. Regarding the role of MRI, its almost as efficacious as fetal neurosonogram with greater accuracy for predicting cortical developmental defects.

Recommendations

1. Incorporation of fetal CNS evaluation in 11-13+6 weeks scan.
2. Detailed neurosonogram with foetuses suspicious of having CNS malformation.
3. Detailed second trimester anomaly scan for all.
4. Use of fetal MRI as and when required if detailed fetal central nervous system assessment is required for diagnosis and management counselling.
5. Multidisciplinary approach involving geneticist, maternal-fetal medicine specialist, pediatrician, pediatric neurosurgeons.
6. Use of molecular testing whenever indicated.
7. Option of termination of pregnancy, antenalt and postnatal evaluation and intervention should be advised.

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