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CASE REPORT

Classic Dandy-Walker malformation with gray matter heterotopia, corpus callosal dysgenesis and occipital cephalocoele: A case report

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INTRODUCTION

Dandy-Walker malformation (DWM) lies at the most severe end of the spectrum of cystic posterior fossa anomalies. Several associated intracranial anomalies have been described including corpus callosum dysgenesis and gray matter abnormalities, as were seen in our case.[1] Occipital cephalocoele, a coexistent abnormality in this case, is a relatively remote association and has hardly been reported with DWM.[2] This case gives a comprehensive idea of all the commonly and infrequently described intracranial associations of DWM, all coexisting in the same patient and highlights the significance of sonography as an effective screening tool.

CASE DESCRIPTION AND MANAGEMENT

A 1-day-old female neonate was brought to the Department of Radio-diagnosis for ultrasonography of suspected caput succedaneum. Ultrasonography of the swelling revealed an anechoic cystic lesion in the scalp with intracranial communication through a gap in the occipital bone. Ultrasonography of the posterior fossa, done to ascertain the deeper extent of the lesion, revealed a large posterior fossa cyst communicating with a dilated fourth ventricle with associated vermian agenesis. The cerebellar hemispheres were hypoplastic and winged anterolaterally **(FIGURE 1. I-II)**.

LEARNING POINTS

1. This case reveals the importance of prenatal anomaly scan so that the parents can take informed decision regarding termination as mortality rates in classic Dandy-Walker Malformation are significantly high.
2. The case also highlights the role of Ultrasonography of brain in diagnosing the suspected caput as occipital cephalocoele communicating with a posterior fossa cyst.
3. As a representative case, it draws attention to a plethora of commonly and rarely described intracranial anomalies that should be searched for in a case of Dandy-Walker Malformation.

The findings were consistent with DWM. Complete sonographic evaluation of the brain was done in search of other associated intracranial anomalies demonstrating nodular projections isoechoic to gray matter along the lateral walls of both lateral ventricles with mild hydrocephalus. The splenium of corpus callosum was deficient with sulci radiating peripherally from the expected location of splenium giving a "spoke-wheel" gyral pattern **(FIGURE 1. III-VI)**. The mother had never undergone an antenatal anomaly scan. Magnetic resonance imaging of the neonatal brain was advised for corroboration.

Mmagnetic resonance imaging performed after a month validated the findings of classic DWM with associated periventricular gray matter heterotopia, corpus callosal dysgenesis, hydrocephalus and occipital cephalocoele

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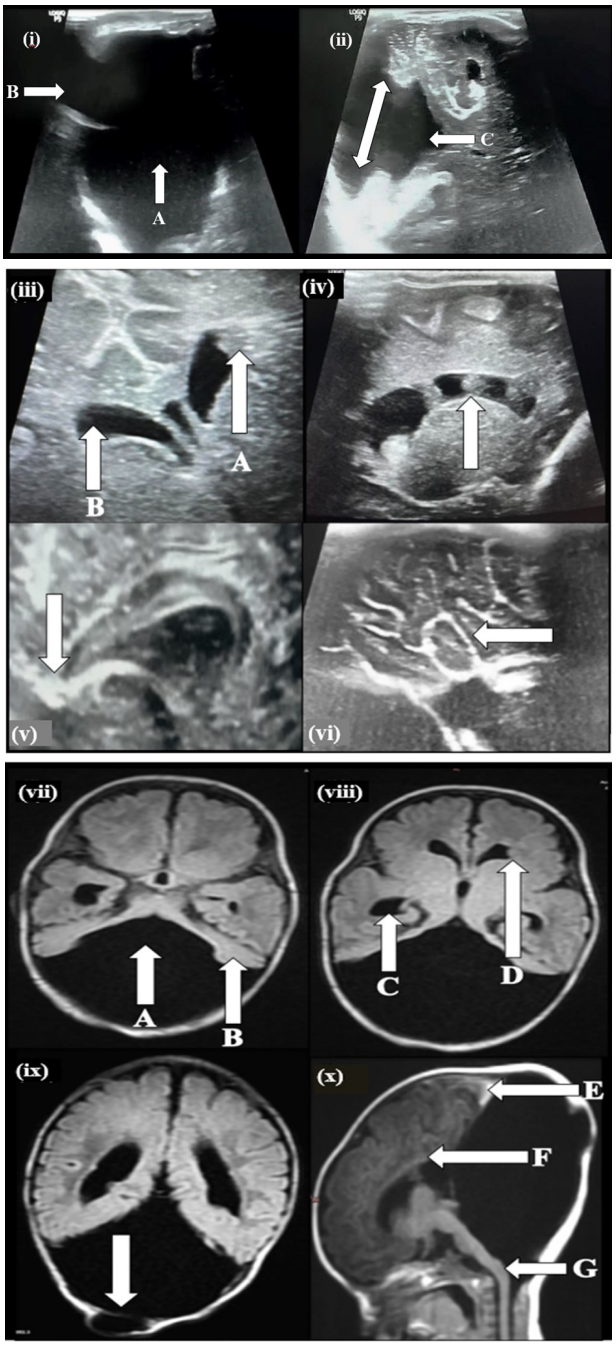

(i) Axial USG scan of posterior fossa with A, a large posterior fossa cyst and B, occipital cephalocoele communicating with the posterior fossa cyst. (ii) Axial USG scan of the posterior fossa showing C, dilated fourth ventricle continuous with the posterior fossa cyst with absence of vermian tissue and winged-out hypoplastic cerebellum. (double-sided arrow).

(iii) Coronal USG scan of neonatal brain showing A, periventricular nodular gray matter heterotopia and B, mild hydrocephalus. (iv) Parasagittal USG scan through the left lateral ventricle showing multiple periventricular nodules (arrow). (v) Midsagittal USG scan ahowing the corpus callosum with deficient splenium (arrow). (vi) Posterior aspect of midsagittal USG image showing radiating gyri and sulci (arrow) in the expected location of the splenium giving a "spoke-wheel" gyral pattern.

(vii) Axial T2/FLAIR image at the level of midbrain showing A, large posterior fossa cyst communicating with a dilated fourth ventricle with vermian agenesis and B, anterolaterally winged out hypoplastic cerebellar hemisphere. (viii) Axial T2/FLAIR image showing C, dilated temporal horn of right lateral ventricle consistent with hydrocephalus and D, nodular projection in frontal horn of left lateral ventricle with signal intensity similar to gray matter suggestive of periventricular nodular gray matter heterotopia. (ix) Axial T2/FLAIR image above the level of third ventricle showing the occipital cephalocoele (arrow) communicating with the posterior fossa cyst. (x) Midsagittal T1-weighted image showing E, elevation of confluence of sinuses by the posterior fossa cyst, F, absent splenium and G, cervicomedullary kink.

FIGURE 1 **Ultrasonographic and magnetic resonance images of brain**

communicating with the posterior fossa cyst. Additional important information obtained from MRI brain was the kink at the cervicomedullary junction, indicating a poor prognosis **(FIGURE 1. VII-X)**. There was progressive increase in hydrocephalus compared to the initial scan.

The patient never underwent any definitive treatment or corrective surgery for hydrocephalus. A follow-up one month later revealed that the baby had respiratory distress and a poor suck reflex. Patient was under symptomatic and supportive treatment (oxygen support and paladai feeding) and was stable. Unfortunately, a subsequent follow up after six months she passed away one week back.

DISCUSSION

Almost all the known intracranial associations of DWM were seen in our case, including the relatively rare occipital cephalocoele and no such case with all these associations in a single patient was found in existing literature. This case is a prototype of classic DWM with a natural course as described classically in literature and highlights the ominous coexistence of cervicomedullary kink. The only limitation of our report is that no definitive treatment or corrective surgery could be undertaken.

DWM is a rare spectrum of congenital anomalies with an incidence of 1 in 30,000 live births with mild female predisposition, as in our case.[3, 4] This spectrum of cystic posterior fossa anomalies comprises of classic DWM, Blake pouch cyst, inferior vermian hypoplasia and mega cisterna magna.[1] DWM is the most severe anomaly in the spectrum with the classic triad of enlarged posterior fossa, upward displacement of the confluence of sinuses with lambdoid-torcular inversion and cystic dilatation of the fourth ventricle.[1] Vermian agenesis can be complete or partial.[5] This classic triad was seen in the index case.

The diagnosis of DWM warrants search for other extracranial anomalies due to syndromic associations like Klippel-Feil syndrome, Aicardi syndrome, Trisomy 18 and PHACE syndrome.[6] Common intracranial associations are corpus callosal dysgenesis, gray matter heterotopia and hydrocephalus.[1] Occipital cephalocoele is a rare association, with only 33 such cases described in literature as of 2022.[2]

Ultrasonography and magnetic resonance imaging done one month apart revealed progressive hydrocephalus in our case. Standard surgical treatment includes ventriculoperitoneal or cystoperitoneal shunts for relief of hydrocephalus.[7] Besides, supportive treatment like oxygen therapy and alternative feeding strategies are required, the only measures employed in our case.

Any couple who had experience of having a baby with DWM, must undergo an anomaly scan because there is a high incidence of such a condition among siblings.

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Author contributions

Manuscript drafting and revising it critically: AM, SS, NP, AM. *Approval of the final version of the manuscript:* AM, SS, NP, AM. *Guarantor of accuracy and integrity of the work:* AM, SS, NP, AM.

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Conflict of interest

We do not have any conflict of interest.

Ethical approval

Ethical approval was not sought because this is a case report. However, informed written consent was obtained from the patient for preparation of this manuscript.

Data availability statement

The data that support the findings of this study are available on request from the corresponding author.

REFERENCES

1. Osborn AG, Hedlund GL, editors. Osborn's Brain Imaging, Pathology and Anatomy. 2nd ed. Philadelphia (PA): Elsevier; 2018. 1186-91. https://books.google.com.bd/books/about/Osborn_s_Brain.html?id=2hvfAQAACAAJ&redir_esc=y
2. Gutierrez F, Ballestero M, Herrera D, Gonzalez C, Cardona A, Mora L. Occipital encephalocele associated with Dandy-Walker malformation: a case-based review. Childs Nerv Syst. 2022;38(9):1683-1688. DOI: https://doi.org/10.1007/s00381-022-05551-2
3. Bosemani T, Orman G, Boltshauser E, Tekes A, Huisman T, Poretti A. Congenital Abnormalities of the Posterior Fossa. Radiographics. 2015;35(1):200-20. DOI: https://doi.org/10.1148/rg.351140038
4. Sreelatha S, Vedavathy N, Sathya P, Hanji N. Dandy-Walker variant: A case report. Sch J Med Case Rep. 2014;2:40-41. DOI: https://doi.org/10.36347/sjmcr.2014.v02i01.016
5. Rohanachandra YM, Dahanayake DM, Wijetunge S. Dandy-Walker Malformation Presenting with Psychological Manifestations. Case Rep Psychiatry. 2016;2016:9104306. DOI: https://doi.org/10.1155/2016/9104306
6. Sule MB, Gele IH, Shirama YB, Abacha M. Dandy-Walker Malformation with an Occipital Cephalocele in an Infant: A Case Report.J Clin of Diagn Res.2020; 14(5):TD04-TD05. DOI: https://doi.org/10.7860/JCDR/2020/43253.13716
7. Mohanty A, Biswas A, Satish S, Praharaj SS, Sastry KVR. Treatment options for Dandy–Walker malformation. Journal of Neurosurgery: Pediatrics PED. 2006;105(5):348-356. DOI: https://doi.org/10.3171/ped.2006.105.5.348