A Rare Triple Neural Tube Defect- Case Report

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Date of submission : 14th Feb 2023 Date of acceptance: 20th June 2023 Date of publication : 15th July 2023

Abstract

Multiple neural tube defects are a rare entity and it also includes anomalies like encephalocele, dermal sinus, myelomeningocele, and spina bifida. These occur due to defective closure of neural tube at multiple sites. There have been isolated cases of coexistence of triple neural tube defect in same subject and only 9 cases have been reported in available world literature.

Here, we present a rare case of 7 days old boy with three different NTDs- occipital encephalocele, upper cervical meningocele and lumbar myelomeningocele. Type II Chiari malformation, tethered cord, fenestration of posterior falx, hydrocephalus and syringohydromyelia accompanying the NTDs.

Key words: Multiple neural tube defects, Encephalocele, Myelomeningocele, Magnetic Resonance Imaging, Fenestrated Posterior Falx.

Introduction

Neural tube defect is a complex entity comprising multiple anomalies of central nervous system including anencephaly, encephalocele, and spina bifida.[1] CNS anomalies are most common defects encountered at birth.[2] NTD can be classified by neurulation defects occurring as stage 1, 2 and post neurulation defects.[3] Neurulation defects are characterized as absence of skin covering over the defect and include craniorachischisis, myelomeningocele and anencephaly. After neurulation, the ectoderm is separated from neural tube and the defects are usually closed by skin. Multiple neural tube defects is a rare form of the spectrum, occurring in less than 1% of cases in which the defects are at more than one site along the vertebral axis. [4]

Access this article online
Website: https://www.nepjol.info/index.php/NJN
DOI: https://doi.org/10.3126/njn.v20i2.52488
HOW TO CITE
Kumar A, Kumar K. A Rare Triple Neural Tube Defect- Case Report. NJNS. 2023;20(2):79-83.

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ISSN: 1813-1948 (Print), 1813-1956 (Online)

We present a case report of 7 days old child with occipital encephalocele, upper cervical meningocele and lumbar myelomeningocele associated with type II Chiari malformation, tethered cord, fenestration of posterior falx cerebri, hydrocephalus and syringohydromyelia.

Case Report

A 4- day old male child was brought to the department of pediatrics, IGIMS with the complaint of swelling in lower back and nape of neck since birth. The baby was appropriate for gestational age, delivered at full term at goverment hospital and cried immediately after birth. There was no history of abnormal body movement, lethargy, refusal to feed or rash.

On examination, two swellings were noted at occipital region, each measuring approx 2 x 3cm (Fig 1) and 4x 6 cm sized swelling at lumbar region (Fig 2). Bilateral lower limb movements were normal and passing urine and stool normally. Anterior fontanelle was at level. Other systemic examinations were unremarkable. Routine blood investigations were within normal limit.

Magnetic Resonance Imaging (MRI) of brain revealed a CSF containing sac measuring 2.2 x 3 cm containing dysplastic cerebellar tissue through a defect of size 13mm in occipital region in midline (Fig 3). There is also a CSF containing sac measuring approx 2.5 x 3.5 cm through a defect a defect of size 15 mm in posterior element of C1 cervical vertebra in the midline (Fig 3). There is descent of brainstem and cerebellar tonsil in foramen magnum with crowding of foramen magnum and effacement of fouth ventricle causing upstream dilatation of third and bilateral lateral ventricles (Fig 8). Cervicomedullary kink and tectal beaking is also seen (Fig 8). There is fenestration of falx cerebri at high parietal and occipital region with interdigitation of gyri (Fig 7). MRI whole spine showed a

defect of size 9.2 mm in posterior element of L4 vertebra through which distal portion of spinal cord (conus medularis), few cauda equine nerve roots, meninges and CSF are seen herniating in a sac, measuring 6.2 x 5.7 cm (Fig 4). Central canal of spinal cord is dilated with maximum diameter- 5 mm at T5 level opposite to C6 to T11, which appear T2 hyperintense and T1 hypointense suggestive of syringohydromyelia (Fig 6). Spinal cord is low lying, extending below L4 level, suggestive of tethered cord.

Computed tomography (CT) study showed midline bony defect at inferior aspect of occipital bone, posterior element of C1 vertebra and posterior element of L4 vertebra (Fig 5).

Ultrasonography (USG) showed two cystic masses in cervical region, one containing CSF with internal echoes and brain tissue and another only CSF (Fig 1). CSF filled sac containing herniated spinal cord and nerve roots seen at lumbar region (Fig 2).

The child has been successfully operated for lumbar myelomeningocele and will be operated for other defects in due course of time.

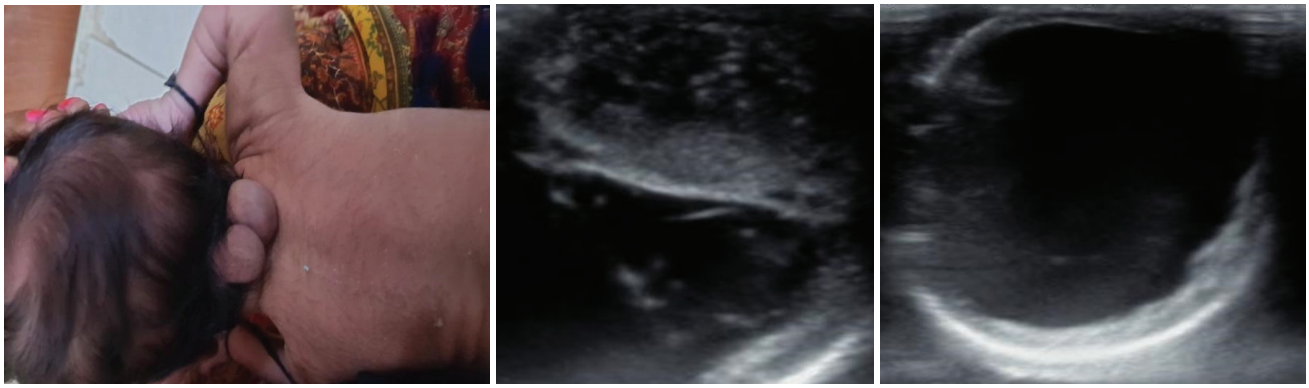

Figure 1: Two swellings in cervical region - occipital encephalocele and cervical meningocele

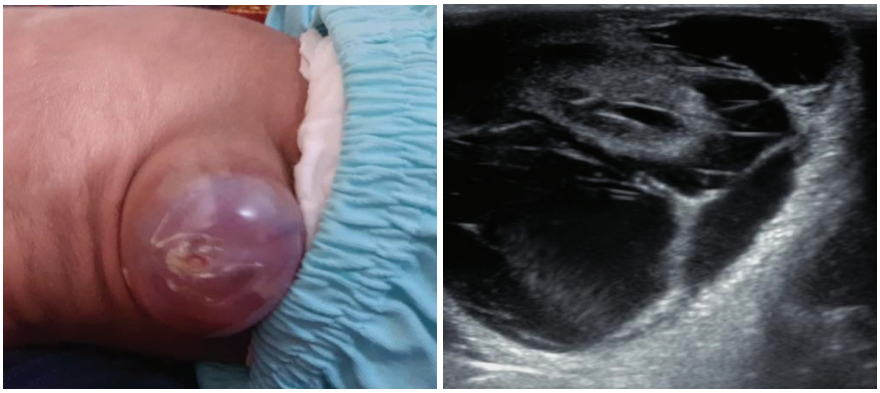

Figure 2: Swelling in lumbar region - lumbar myelomeningocele

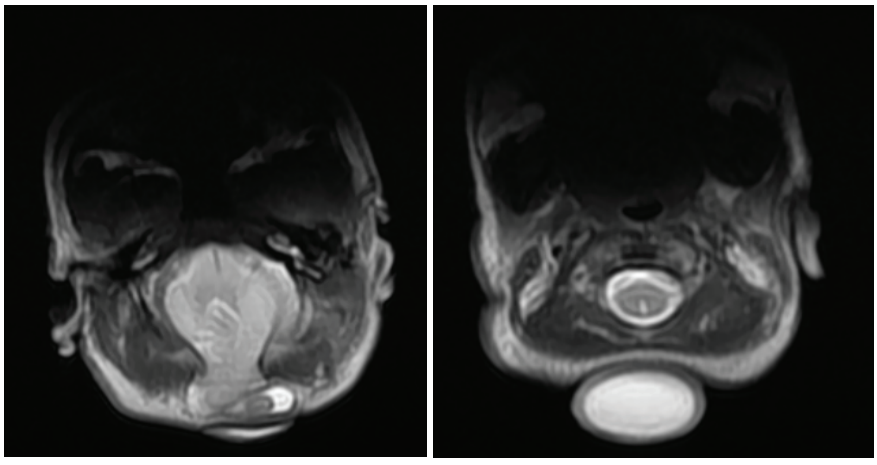

Figure 3: MRI Images: Occipital Encephalocele and cervical meningocele.

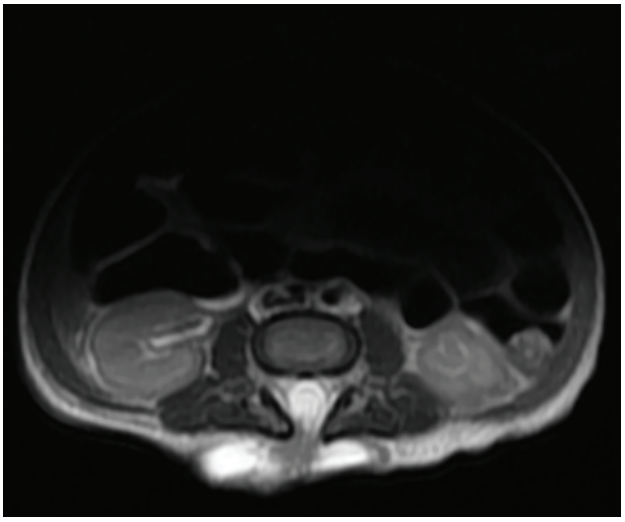

Figure 4 : Lumbar Myelomeningocele

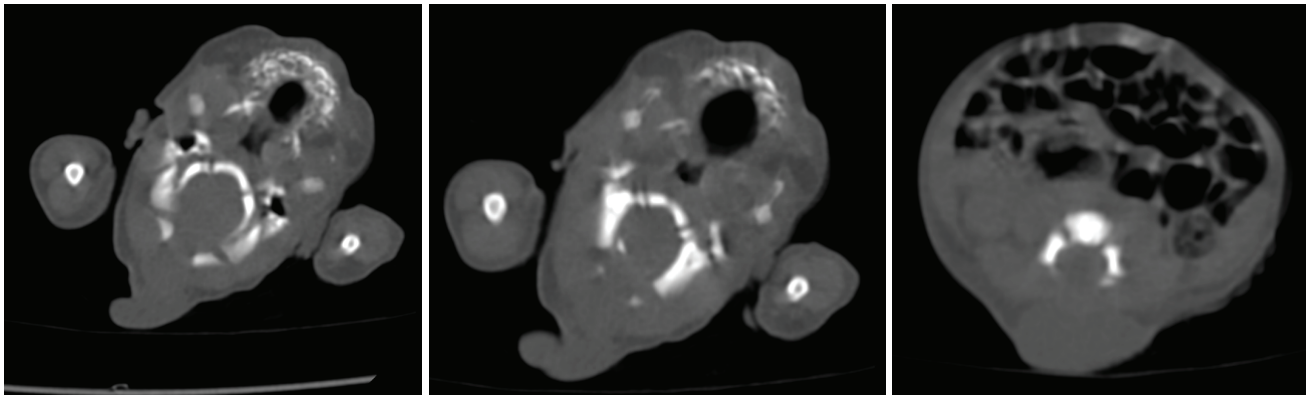

Figure 5: Ct Images Showing Bony Defect In: Occciptal bone, C1 Vertebra and L4 vertebra

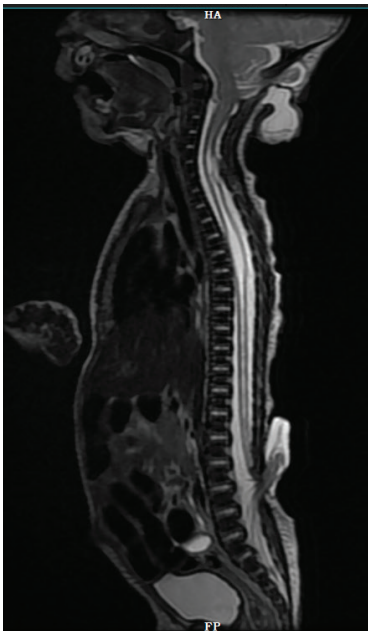

Figure 6: Triple Neural Tube Defect With Syringohydromyelia

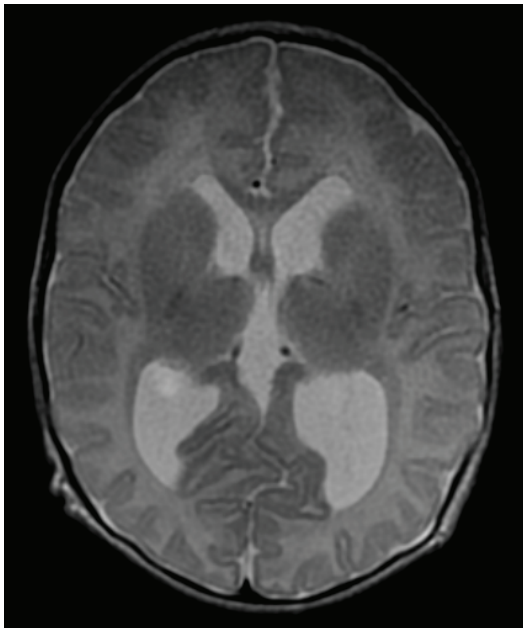

Figure 7: Hydrocephalus With Fenestration Of Falx Cerebri Posteriorly

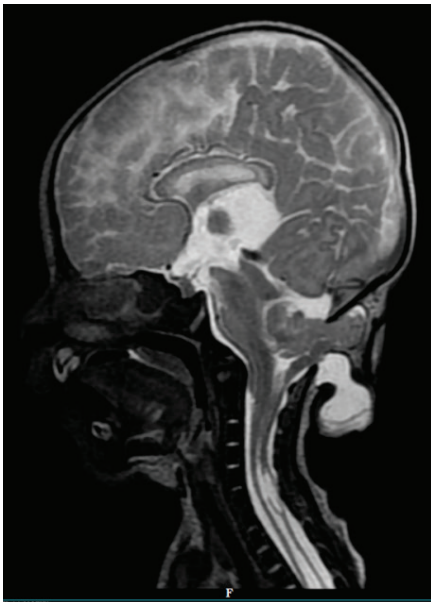

Figure 8: Descent Of Brainstem & Cerebellar Tonsils Causing Crowding At Foramen Magnum, Cervicomedullary Kink And Tectal Beaking

Discussion

Multiple neural tube defects (MNTDs) are extremely rare congenital anomalies reported in literature, most of them being reported from developing countries like India and Africa. The term "multiple neural tube defect" refers to the simultaneous occurrence of defect in single patient with normal neural tissue in between them. Unlike MNTDs, 'Complex' spinal defects are multiple defects that are contiguous in manner.[5]

The prevalence of neural tube defect is 4.5 per 1000 live births in India.[6] The prevalence of neural tube defect is 0.7 per 1000 births in USA, 0.7 per 1000 births in Canada and 1 per 1000 births in Africa.[7]

Etiopathogenesis of Neural tube defect (NTD) is multifactorial comprising gene-gene, gene-nutrients, gene- environment (non-nutrient) interactions.[1] Neurulation occurs at 3-4 weeks of geststion in which neuroectoderm undergoes various changes to form neural tube. The conventional single site theory explains the closure to be zipper like in which there is bidirectional closure. According to this, the defect could be either at anterior or posterior end. However, this failed to explain the causation of MNTDs, therefore the Multiple site neural tube closure theory was postulated by Van Allen et al,[4] who hypothesized that there are five closure sites which proceeds in a sequential manner and any deviation from this sequential closure leads to the development of different combinations of MNTDs.[8] The " re-zippering initiation model" postulated by Mahalik et al. is the most recent and advanced theory which explains the development of any type of neural tube defects including MNTDs in a better way.[9] Deora et al. and Shiferaw et al. agreed to this explanation of development of neural tube defect.[10,5]

MNTDs are extremely rare type of NTDs. Review of MNTDs cases by Deora et al showed that Only 57 cases have been reported in world literature out of which more than 50% cases have been reported from India. 3 major series on MNTDs have been done in India. Encephalocele associations have been more observed in case series of Deora et al. (55%) as compared to previous case series by Ahmad et al (42 %) and Mahalik et al. (30 %).[5]

Adequate intake of folic acids in pre-conception period and in early pregnancy can lead to significant reduction in risks of NTDs.[11] Limited Indian studies are available to assess the folic acid deficiency among pregnant females which shows conflicting prevalence rates ranging from 1.2% to 26.3 %.[12] Clinical trials have shown to reduce the risk of NTDs using folic acid supplementation.[13]

The patients with neural tube defects need to be investigated for other associated congenital anomalies. Creating awareness among general population about NTDs and folic acid supplementation along with vit B12 should be done to prevent children from these NTDs.[5]

Conclusion

Triple neural tube defect are rare. To the best of our knowledge, this is the tenth case of triple neural tube defect and first case of triple neural tube defect associated with multiple other brain and spinal cord anomalies in world literature. The child was operated for lumbar myelomeningocele and the procedure went uneventful. The treating neurosurgeons have kept a gap of two months for operating the occipito-cervical region to avoid any complications.

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