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Case Report

Anterior thoracic myelomeningocele presenting with neck pain: an unusual spinal dysraphism

Rekha Tanwar*, Shibani Mehra, Pooja Gupta, Shashi Kumar Singh

Department of Radiology, ABVIMS and Dr RML Hospital, New Delhi, India

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***Correspondence:**
Dr. Rekha Tanwar,
E-mail: drrekhatanwar91@gmail.com

ABSTRACT

Cervicothoracic myelomeningocele is a rare entity with few cases reported in previous literature of anterior thoracic myelomeningocele. Spinal dysraphism (SD) is a congenital malformation of the spine involving the spinal cord which is characterized by incomplete midline closure of mesenchymal osseous and nervous tissue in the developing fetus. Early diagnosis of myelomeningoceles offers several benefits, including parental counselling, treatment planning, and reducing potential associated complications. MRI is the best modality to study soft tissue morphology of the cord as well as vertebral and discal pathologies. We report a case of 4-year-old female child presented with multiple episodes of neck pain of 1 year duration. The antenatal history was unremarkable except for lack of folic acid intake as a supplement during the pregnancy. There was mild kyphoscoliotic deformity in upper thoracic spine. Radiological imaging was done for further evaluation. MRI revealed herniation of a CSF containing thecal sac ventral to the spine protruding from D1 to D3 vertebral level through a midline defect with neural placode was seen within and tethering of the neural placode. On MRI, the radiological diagnosis of true anterior myelomeningocele was made. Congenital malformations of the spinal cord include a range of abnormalities with varying but distinctive imaging features thereby making MRI the best modality for evaluation and diagnosis. Radiologists and neurologists must both be aware of the wide spectrum of SD and the unusual presentations like neck pain or paraparesis, for early diagnosis and effective management. MRI of brain and spinal cord is the key in detection of anterior myelomenigocele which is an occult form of SD.

Keywords: SD, MMC, MRI T1WI FSE

INTRODUCTION

Spinal dysraphism (SD) is a congenital malformation of the spine involving the spinal cord. The term dysraphism originates from the Greek words dys (bad) and rhaphḗ (suture). It encompasses a range of spinal cord development abnormalities cord occurring between the 2nd and 6th gestational weeks, characterized by incomplete midline closure of mesenchymal, osseous, and nervous tissue in the developing fetus.

SD is the second most common birth anomaly after congenital heart disease, with an estimated worldwide incidence and prevalence of 0.5-2.5 and 1 to 3 per 1000 live births respectively.[1] The socioeconomic status of patients, level of antenatal care, maternal nutrition, genetics, and environment play a significant role in the disparity in global distribution of SD.[1]

The less common variety of SD is Anterior myelomeningocele. The common location of anterior myelomengocele is sacrum, at the level of filum terminale.[2] Only few cases of thoracic location of anterior myelomeningocele have been reported so far in the literature to the best of our knowledge.

SD can be diagnosed antenatally or shortly after birth. However, some cases may not be detected later in life due to a lack of clinical symptoms. Early diagnosis of myelomeningoceles offers several benefits, including parental counselling, treatment planning, and reducing potential associated complications.[3] MRI is the best modality to study soft tissue morphology of the cordas well as vertebral and discal pathologies.

CASE REPORT

A 4-year-old female patient presented with multiple episodes of severe neck pain of 1 year duration. The pain was relieved by the patient holding the neck steady with his hands and compressing for 5-10 seconds. The antenatal and birth history of the patient was unremarkable. The antenatal history was unremarkable except for lack of folic acid intake as a supplement by the mother during the pregnancy. The neurological examination of the child did not reveal any neck rigidity. There was mild kyphoscoliotic deformity in upper thoracic spine (T1-T6) vertebral level with convexity towards right side. The blood work was normal.

Skiagram of cervical and dorsal spine AP and lateral views demonstrated a midline bony vertebral defect in D1, D2 and D3 vertebral bodies. Further evaluation of the spine was performed with NCCT on Siemens 128 slice dual energy CT scan machine. Bodies of D1, D2 and D3 thoracic vertebrae revealed a defect in the midline, with maximum defect size of 14 mm at D2 vertebral level. The spinous process of D2, D3 and D4 vertebra were seen fused together. A cystic fluid attenuation mass was seen herniating ventrally through vertebral defect. Mass did not contain any fat or calcification (Figure 1 A and B).

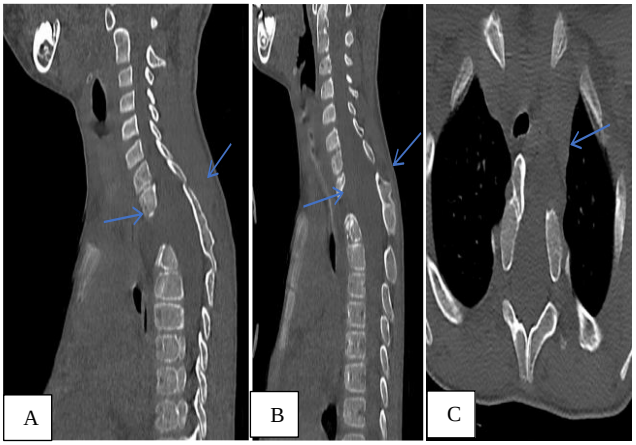

Figure 1 (A-C): NCCT of cervicothoracic spine. Sagittal images-midlinebony defect in D1, D2 and D3 vertebral bodies (blue arrows) with fusion of spinous process of T2, T3 and T4 vertebra (blue arrows).

The child underwent MRI cervicodorsal spine with whole spine screening on a 3 Tesla MR machine (Siemens, Syngo Skyra) for better characterization and evaluation of the cystic mass. Sequences taken were T1 and T2 weighted Fast spin echo, T2 short tau in-version recovery (STIR) and T1 TSE after gadolinium contrast administration.

MRI revealed herniation of a CSF containing thecal sac measuring 2.1×2.3×4.8 cm ventral to the spine protruding from D1 to D3 vertebral level, through a midline defect in these three upper thoracic vertebrae. The thecal sac in the right ventral para-vertebral location was seen extending from D3 to D7 thoracic vertebrae (Figure 2 A and B). T2 hypointense neural placode was seen within this thecal sac tethering of the neural placode to the left antero-lateral wall of thecal was seen, causing stretching of the ventral thoracic cord (Figure 2 C and D). Focal cord edema was noted at D2 level. Short segment syrinx formation was seen at 2 levels, located at C7-D1 level and also at D4-D6 levels. The cystic mass was not causing mass effect on the oesophagus.

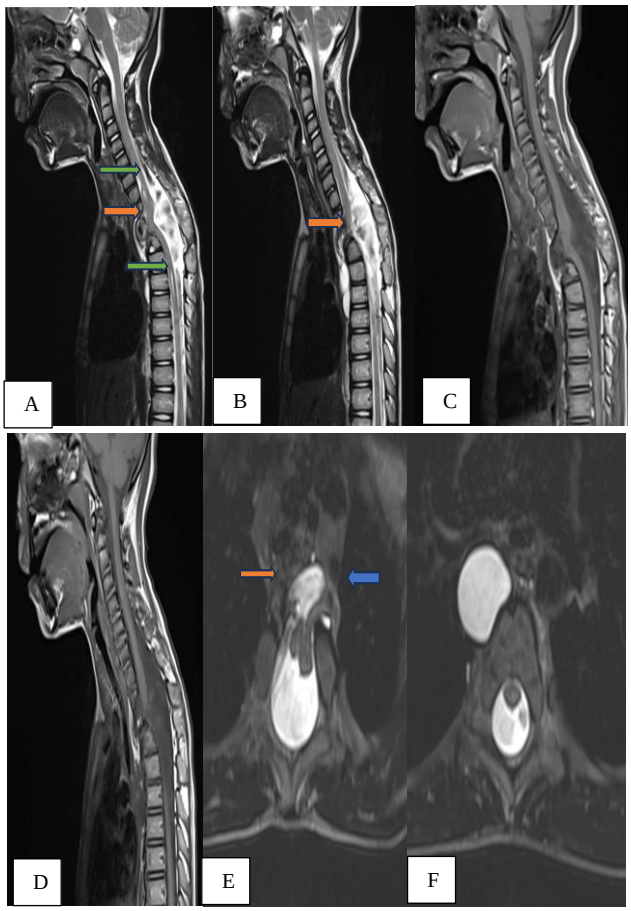

Figure 2: (A-F): T1W and T2WI Cervicothoracic spine MRI in sagittal plane-midline vertebral body defects noted in D1, D2 and D3 vertebral bodies with herniating thecal sac anteriorly and posteriorly. Short segment syrinx is noted at C7-D1 and D4-D6 vertebral levels (green arrows). T2W MR in axial plane. Thecal sac containing CSF is seen in right paravertebral region with the neural elements (Orange arrow). The hypointense neural placode is seen attached to left anterolateral wall of sac (Blue arrow).

Sagittal T1 and T2 sequences of the brain centred at the cranio-vertebral junction were added to look for any associated brain anomaly. The cerebral parenchyma and ventricular system were found to be normal. There were no cerebellar malformations nor any supratentorial anomalies of the corpus callosum. No heterotopic grey matter was detected.

In view of a CSF intensity sac/mass, containing a tethered neural placode within, seen herniating ventrally through a midline vertebral defect in the upper thoracic spine at D2 level on MRI, the radiological diagnosis of true anterior myelomeningocele was made.

DISCUSSION

Myelomeningocele is used to describe herniation of the meningeal sac containing a neural placode within, through a bony defect in the vertebral column. It occurs due to defective primary neurulation at 3-4 weeks of gestation.[1] Approximately 66% of all myelomeningoceles are associated with neurofibromatosis type I or Marfan`s syndrome, with only 22% being non syndromic presentations.[5]

Maternal folic acid deficiency is associated with a two to eight-fold increased risk of the fetus developing myelomeningocele, highlighting the significance of prevention strategies in prenatal care. Women with one child born with myelomeningocele carry a 3-8% increased risk of giving birth to child with similar birth defect in the subsequent pregnancy.[6] However, 95% of myelomeningoceles occur spontaneously in children born to women with no family history of the condition. In the United States of America, fortification of foodstuff with folic acid has had a significant impact on reducing the incidence of children born with myelomeningocele.[7]

In myelomengocele, the herniation of thecal/meningeal sac is commonly seen from the dorsal aspect of the spine and is mostly seen in the lumbosacral region. Ventral or anterior myelomeningoceles are extremely rare and are located in the thoracic or the sacral region.[8] An association with Marfan's syndrome or NF I is described. Clinical presentation can be with neck pain or even paraparesis if the herniating sac causes significant cord compression. Dyspnoea, shortness of breath or dysphagia may occur from compression of mediastinal structures.

For anterior cervico-thoracic myelomeningocele, localization of the defect differentiates a false anterior myelomeningocele from the true type. False variant of myelomeningoceles is associated with neural foraminal enlargement and is more commonly encountered. False anterior meningoceles are not primarily anterior in location, rather are lateral, and dural herniation is seen through an enlarged intervertebral neural foramen. However, true anterior thoracic myelomeningoceles are rare entities with only few cases documented in literature.[2,3,5] True anterior meningoceles have an association with hydrocephalus and Chiari type-II malformation in up to 85% of patients.[4] We did not find associated hydrocephalus and Chiari type-II malformation in this patient. Thoracic myelomeningoceles are more commonly associated with neurofibromatosis type 1, however there were no features of Phakomatoses in our patient.[8]

MRI is the modality of choice for assessment of the cord as well as the vertebra with its posterior elements. MRI is excellent for demonstration of the anatomical relationship of spine, the cord, and the herniating CSF sac with each other, and with other adjacent structures.

In the case being reported, the ventral myelomengocele was not associated with an enlarged neural foramen, instead was seen extending through the midline vertebral defect thereby confirming it to be a true anterior myelomengocele. The location of the myelomengocele was the dorsal/thoracic region in our patient. Cervicothoracic myelomeningoceles are rare and to the best of our knowledge only few cases of anterior myelomeningocele have been reported so far in the cervico-thoracic region.

Purely thoracic anterior myelomeningoceles are even more rare, and this makes the current case truly unusual.

Management

Depending on the size of defect, a laminectomy and intradural repair may suffice for smaller defects while, extirpation of the cystic mass through a right anterolateral thoracotomy, can be done for larger defects. The drawback of thoracotomy is that chest drainage is often needed, and CSF fistulas may develop. Recurrence is also reported.

CONCLUSION

Congenital malformations of the spinal cord include a range of abnormalities with varying but distinctive imaging features thereby making MRI the best modality for evaluation and diagnosis. Radiologists and neurologists must both be aware of the wide spectrum of SD and the unusual presentations like neck pain or paraparesis, for early diagnosis and effective management. MRI of brain and spinal cord is the key in detection of anterior myelomengocele which is an occult form of SD.

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