Giant Intrathoracic Myelomeningocele in Von Recklinghausen Disease Mimicking Pleural Effusion: A Case Report

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Abstract

A large intrathoracic myelomeningocele, a saccular protrusion of the meninges through a dilated intervertebral foramen or a bony defect of the vertebral column was diagnosed in a 57-year-old male patient showing clinical features of neurofibromatosis-1 (NF-1). Due to rare entity of this disease at first it presented as pleural effusion seen in initial ultrasound.

On detailed investigation it was revealed as a giant intrathoracic myelomeningocele.

Keywords : *Myelomeningocele, Von Recklinghausen disease, spinal dysraphism*

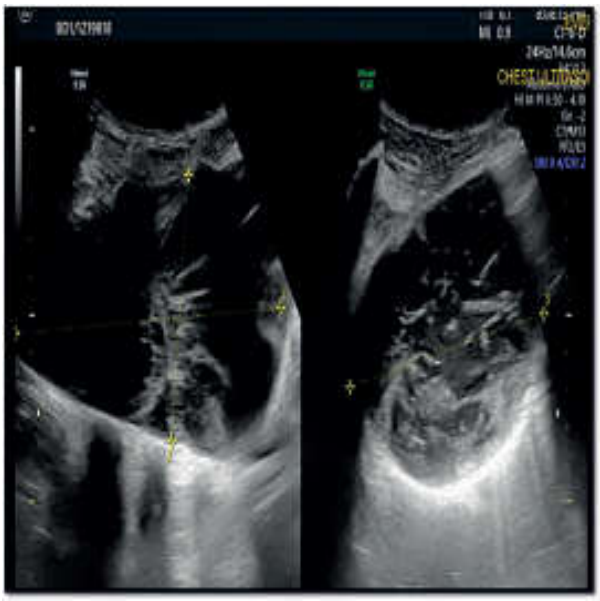

Figure 1: *USG of chest*

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Case Report

A 57 years old non diabetic normotensive male, A known case of neurofibromatosis got admitted in EHD in June 2024 through emergency with the complaints of right sided chest pain for two months and mild exertional dyspnea for same duration. On admission, he was examined thoroughly, and vesicular breath sounds were found in the chest. All relevant investigations were done. Chest USG showed-Right sided loculated and free moderate pleural effusion with passive collapse and consolidation (Figure 1)

CHEST Xray showed -large well defined dense lesion in right hemithorax with scoliosis. (Figure 2)

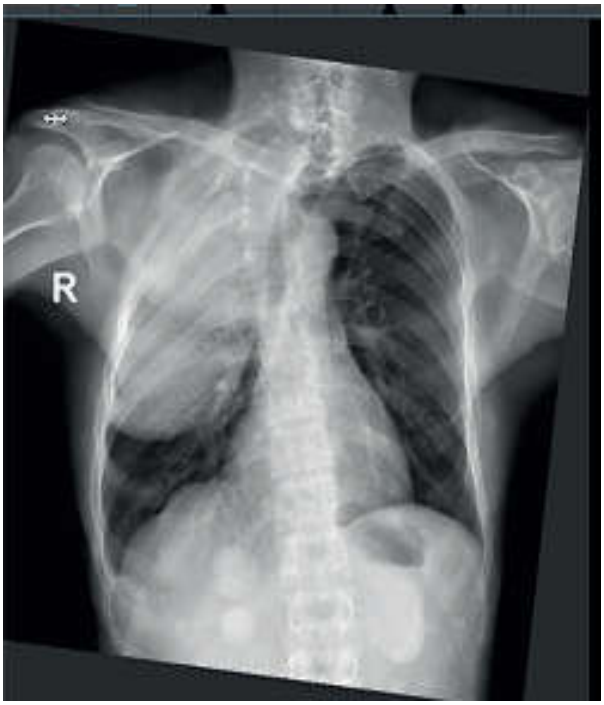

Figure 2: *CHEST X-ray*

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CT scan of chest showed- large well defined dense lesion in right hemithorax with scoliosis (Figure 3a,3b)

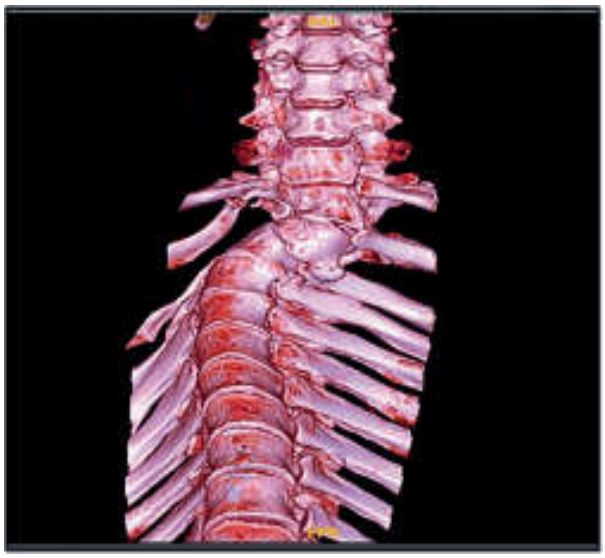

Figure 3 a

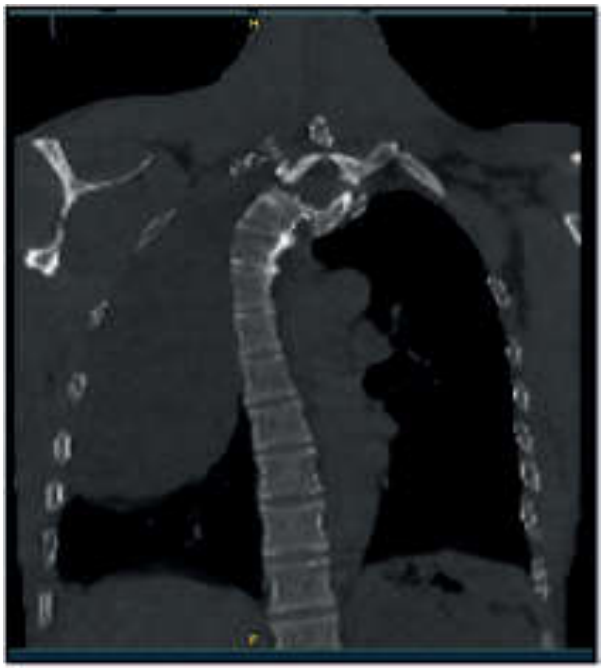

Figure 3 a

MRI of dorsal spine was performed without contrast and showed- Kyphoscoliosis in upper dorsal levels with convexity towards right side. D1 and D2 hemivertebrae noted with fusion between D1-D2 vertebrae on left side. Large cystic lesion measuring 20 cm x 8 cm is noted extending

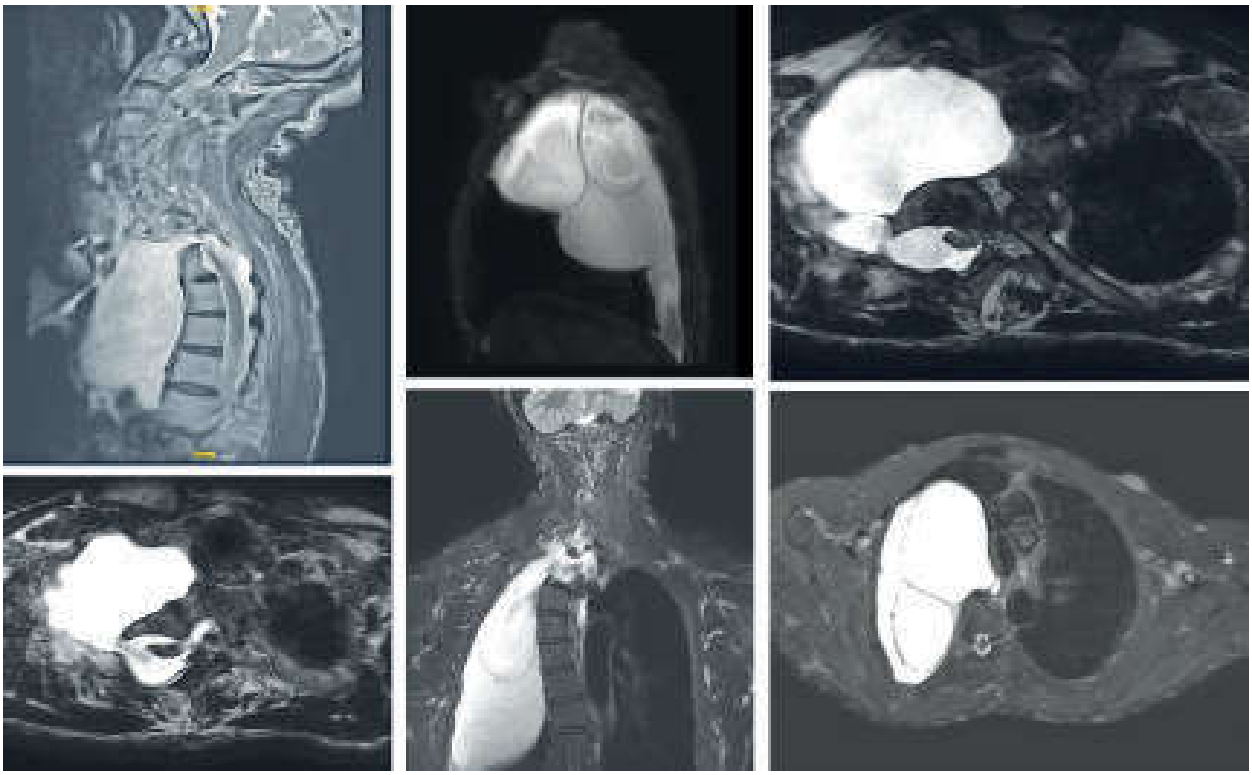

Figure 4 a, b, c, d, e, f: MRI of dorsal spine without contrast showed- large dorsal myelomeningocele due to spinal segmentation fusion anomaly. Multiple enhancing lesions in the visible skin – neurofibromas.

from the spinal canal at this level into the chest cavity compressing the right lung. Spinal cord is slightly deviated towards right side. Few nerve fibers are noted within the cystic lesion. Two septa are noted within the cyst which demonstrate minimum enhancement. Mild meningeal enhancement is present in the adjacent spinal canal. Multiple enhancing lesions are noted in the visible skin – neurofibromas.

Discussion

An intrathoracic meningocele is a relatively rare disease, 60 to 85 % of all thoracic meningoceles are associated with neurofibromatosis type I (NF-1) [1] An intrathoracic meningocele is a cystic formation of the posterior mediastinum, originated by a saccular protrusion of the meninges in the thoracic cavity through the intervertebral foramen pathologically dilated, or by a bone defect in a thoracic vertebra.[2] The accepted etiopathogenesis is the dural dysplasia in patients with neurofibromatosis and enlargement of the intervertebral foramen.[1] Diagnosed intrathoracic meningocele is an uncommon complication of neurofibromatosis type 1[3] although more patients might have small intrathoracic meningoceles without symptoms. Huge meningocele can interfere with lung expansion and can cause respiratory distress. Initially, patients with intrathoracic meningocele may not exhibit any signs or symptoms. However, as the disease progresses, some patients may experience chest tightness or breathlessness due to compression of the lungs. Unfortunately, due to the rarity of the condition and a lack of expertise, the meningocele may be misdiagnosed as pleural effusion. As a result, patients may undergo unnecessary drainage of a mass pleural effusion, putting them at risk of complications and even death. [4]. In this article, we present a case of a patient with NF-1 combined with huge intrathoracic meningocele with NF-1 who was misdiagnosed as pleural effusion.

In a case series, Nanson et al.[6] reported that about 70% of intrathoracic meningoceles were associated with NF1 and scoliosis, referring to the cluster of these disorders as a three-fold syndrome. In our case as well, the patient was known to have NF1 and presented with a giant intrathoracic meningocele and significant scoliosis[6] interestingly, spinal deformity is commonly noted in NF-1. Scoliosis is prevalent in approximately 10% of NF-1 patients, with involvement most commonly observed in the lower cervical and upper thoracic spine.[7] Scoliosis can be either idiopathic or dystrophic. Additionally, rib penciling, vertebral scalloping, severe apical wedging and rotation, and enlargement of foraminal and paravertebral tumors are frequently observed in neurofibromatosis.[8]

Conclusion

This case report showcases a patient with large intrathoracic myelomeningocele which presented as pleural effusion. Multidisciplinary imaging approach played a great role here. Even proper history taking and clinical information about neurofibromatosis of this patient gave an addition clue for association of other pathologies.

Acknowledgements

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