Case Report

Intrathoracic Myelomeningocele masquerading as Bronchogenic Cyst: A Case Report

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ABSTRACT

Neurofibromatosis is an autosomal dominant disorder that affects multiple organs and systems with different complications. Of the 2 types, Neurofibromatosis type 1, also known as von Recklinghausen Syndrome, is more common (96 %) . In this type, rare intrathoracic meningoceles often occur alongside bone dysplasia. This case has been reported as incidental finding of an intrathoracic myelomeningocele in a known patient of neurofibromatosis type 1 which was mistakenly diagnosed as bronchogenic cyst. This error could have put the patient at risk of incorrect treatment, unnecessary invasive procedure, surgery. It reflects the importance of clinical suspicion, the need for advanced imaging techniques and their correct interpretation.

Keywords: *Intrathoracic myelomeningocele, Neurofibromatosis type 1, Bronchogenic cyst, Magnetic Resonance Imaging*

INTRODUCTION

Intrathoracic meningocele is a rare condition where the meninges is bulged through the intervertebral foramen or a defect in the vertebral body. It is most associated with neurofibromatosis type 1, accounting for 60 to 85% of cases[1]. Patients with intrathoracic meningocele mostly remain asymptomatic in the early stages. However, as this condition progresses, patients may develop shortness of breath or chest tightness or heaviness[2]. Unfortunately, because of the rarity of this condition, and lack of knowledge about the association between patients with neurofibromatosis-1(NF-1) with intrathoracic myelomeningocele and lack of clinical expertise, myelomeningocele may be misdiagnosed as bronchogenic cyst. As a result, patients may undergo unnecessary invasive investigations and surgery which can put the patients at risk of complications and even death. In this article, we present a case of a patient with NF-1 with an intrathoracic meningocele who was misdiagnosed as bronchogenic cyst at multiple institutions. Until now, no such case in Bangladesh has been previously reported.

CASE SUMMARY

A 55-year-old male patient with a history of NF-1 with kyphoscoliosis started to notice occasional chest pain and mild dyspnoea on exertion from April 2024. The patient had a history of skin conditions, such as multiple nodules on his front chest, back and few on his limbs, since the age of 5. An outside CT scan of chest with contrast revealed a large thin-walled cystic density lesion measuring about 12.0 x 9.0 x 10.0 cm in the right upper hemithorax at superior mediastinum (Figure 1). No abnormal enhancement was seen. The cyst adhered medially to trachea & right principal bronchus and extended

laterally to costal pleura below upto transverse fissure. The lesion compressed the adjacent lung parenchyma and great vessels. It was diagnosed as bronchogenic cyst in right upper hemithorax.

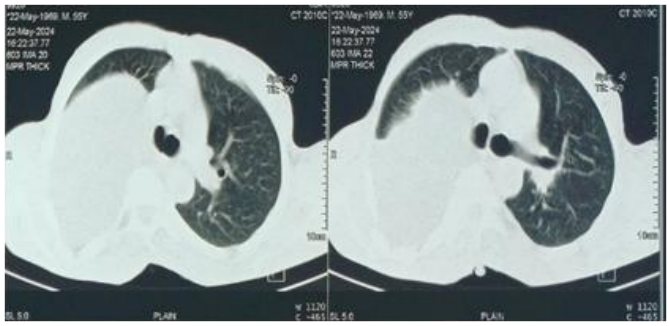

Figure 1:CT scan showing large thin-walled cystic density lesion in right upper hemithorax at superior mediastinum. No abnormal enhancement was seen after contrast injection

Two months later, the patient was referred to a tertiary care institute where his diagnosis of bronchogenic cyst was re-confirmed by chest ultrasound following a chest x-ray (Figure 2), which correlated well with his CT scan. The patient were advised for right uniportal video assisted thoracoscopic surgery (UVATS) for excision of cyst/upper lobectomy, but he refused surgery. The patient was then referred to Evercare Hospital Dhaka with the complaints of right sided chest pain, mild exertional dyspnea and sneezing for about 7 days. On admission, physical examination revealed kypho-scoliosis in upper thoracic level with convexity towards right side. Numerous skin nodules of different sizes were seen over his chest, back, all four limbs and also on scalp (Figure 3).

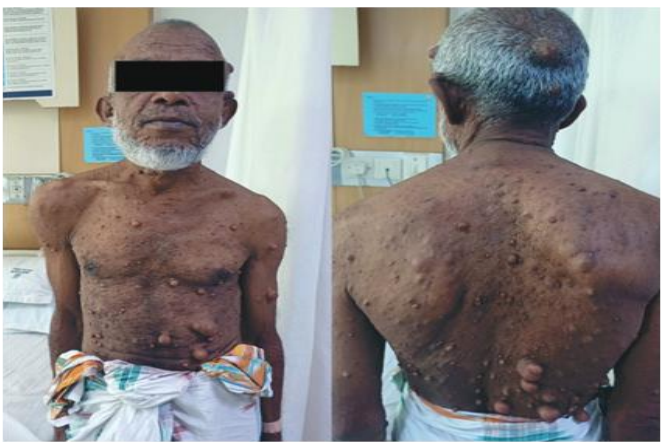

Figure 3: Patient with Multiple neurofibromas on front and back as well as limbs and scalp

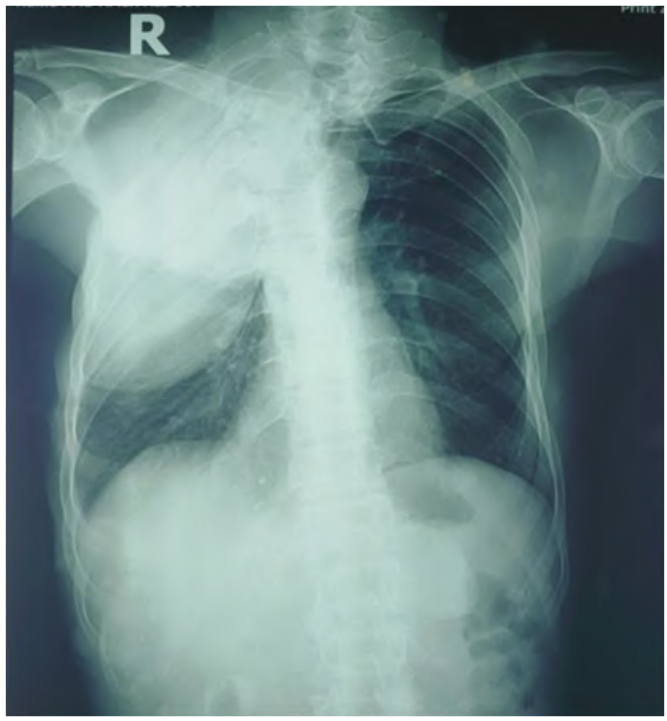

Figure 2: Chest x-ray showing rounded radio-opaque shadow in right upper and mid-zone

He had diminished breath sounds over upper and mid zones of right lung and also dullness on percussion over the aforementioned areas were found on auscultation of his chest. However, no neurological signs or symptoms were observed during the examination. Routine investigation reports were within normal range. Referral was given to cardiothoracic and vascular surgery who after evaluation advised for a second opinion about the CT scan from Radiology.

Radiology advised MRI, which revealed D1 and D2 hemivertebrae with fusion between D1-D2 vertebrae on left side. L arge cystic lesion measuring 20 cm x 8 cm was noted extending from the spinal canal at the aforementioned level into the chest cavity (Figure 4, 5).

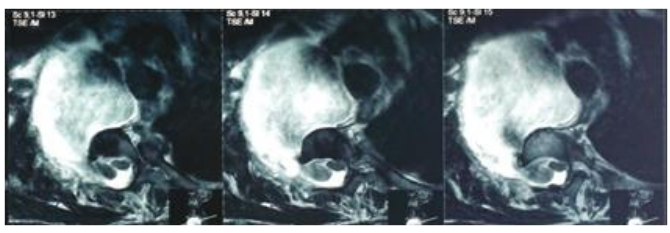

Figure 4: Axial T2 MRI showing CSF filled sac extending from the spinal canal into the chest cavity

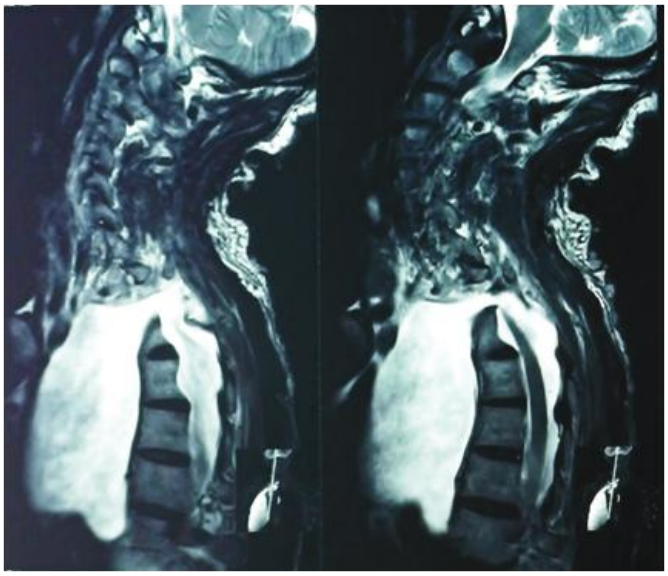

Figure 5: Sagittal T2 MRI showing herniation of cord and CSF filled sac in anterior thoracic region through bony defect

The spinal cord was slightly deviated towards right side. Few nerve fibers were noted within the cystic lesion. Findings were in keeping with large dorsal myelomeningocele due to spinal segmentation fusion anomaly. The patient was reviewed by thoracic surgery, and they advised for surgery. The patient and his attendant were informed of the high risk associated with the procedure. The patient opted for continued observation and follow-up. He was discharged with follow-up advice at OPD basis.

DISCUSSION

Intrathoracic myelomeningoceles associated with neurofibromatosis globally are rare and in context of Bangladesh, they are extremely rare. Until now, no case report on intrathoracic myelomeningocele associated with neurofibromatosis type 1 in the country has been found. The patient did not have any symptoms until he was 55. An intrathoracic myelomeningocele's clinical manifestations are closely related to its size and relationship to surrounding structures, which ranges from back pain, paraparesis from spinal cord injury to shortness of breath and chest pain caused by compression of the lung and mediastinal structures, as was the case for the patient mentioned here. There have been reports of progressive hydrothorax caused by ruptured meningocele as well[3]. Although intrathoracic myelomeningoceles are less associated with neurological deficits compared to lumbosacral ones, they can progress to neurologic deficit, if they are left untreated due to tethering[4,5] . There is often kyphosis due to unrestricted pull of the normally innervated proximal anterior abdominal and intercostal muscles, which may eventually require kyphectomy, to improve lung endurance and functional capacity[6]. Asymptomatic patients can be managed conservatively, while patients with obvious symptoms may require surgery. Surgery is the primary method for fully addressing intrathoracic myelomeningocele in individuals with neurofibromatosis type 1.

CONCLUSION

Detailed history taking and clinical suspicion is essential when treating patients diagnosed as Neurofibromatosis, keeping in mind the association between myelomeningocele, their location and possible relevant clinical features and outcome. Also, selection of relevant advanced imaging techniques and their correct interpretation is of paramount importance as incorrect diagnosis may lead to invasive procedures resulting in complications with potentially fatal outcomes. Our case report is aimed at creating awareness about the existence of such rare conditions and how easily they can be misdiagnosed and the importance of correct interpretation of the advanced imaging tools available to us, by which such conditions can be detected early and can aid in management plan.

DISCLOSURE

Authors have no funding source

INFORMED CONSENT

Informed consent was taken from the patient

ETHICAL APPROVAL

Not applicable

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