CASE REPORT

Congenital Mediastinal Neuroenteric Cyst as a cause of respiratory distress since birth

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Abstract

Mediastinal Neuroenteric Cyst is a rare congenital presentation in infants and is associated with a high mortality rate. It is a very uncommon benign lesion and usually develops from abnormal embryological development of the foregut. Till now, only 106 cases have been reported worldwide. In Pakistan only three cases have been published, with varying presentations. The clinical presentation and age at presentation vary from asymptomatic and coincidental finding on chest x-ray, to limb numbness or early presentation with severe symptoms like those in our case. In fact, it poses an important challenge for paediatricians. We present a rare case with emphasis on clinical presentation and diagnostic criteria.

Keywords: Respiratory Distress; Congenital Bronchogenic Cyst; Foregut Duplication; Congenital Neuroenteric Cyst; Lobar Pneumonia.

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Introduction

The mediastinal neuroenteric cyst is a rare congenital benign cyst of foregut origin. The lesion results from faulty development of certain mesodermal and ectodermal structures during the third week of embryogenesis.[1] Clinical presentation is confusing and diagnosis of such a rare condition often proves challenging for paediatricians. From 1954 till 2021, a total of 106 cases with different clinical presentations have been reported in Medline database.[2] From Pakistan, three cases have been reported in adults with different presentations.[3-5] We present the most commonly encountered clinical presentation and best imaging modality for mediastinal neuroenteric cysts.

Case report

An eight-month-old male infant weighing five kilogram was admitted at the Ayub Teaching Hospital's General Paediatrics ward on February 24, 2020, under MR number 2234221; he was referred through a private clinic.

The patient presented with respiratory distress. On examination, he was severely dehydrated. He had tachypnoea with a respiratory rate of more than 60 breaths per minute, and a heart rate greater than 150 beats per minute. The O_2 saturation with oxygen inhalation was 97%. The baby was febrile with a temperature of 100ºF. Upon auscultation of the chest, a decreased air entry in the right upper lobe and crepitations audible throughout the chest with nasal flaring, intercostal and subcostal recessions was noted. The abdomen was soft, distended and bilateral inguinal hernia was noted. Birth history was uneventful. Past history, since birth, revealed multiple hospitalisations with the same complaints. Total white cell count was $14.2x10^3$/µL (4-$11x10^3$/µL), RBC $4.55x10^6$/µL (4-6 $x10^6$/ µL), Haemoglobin 9.6 g/dl (11.5-17.5g/dl), platelet count was $573x10^3$/ µL (150-$400x10^3$/µL). Serum electrolytes, urea, creatinine, and blood glucose were normal. Abdominal and pelvic ultrasound showed that right sided gut loops were extending till the scrotal sac, with right descended and left retractile testis. Echocardiogram was normal. He was started with treatment of lower respiratory chest infection.

Plain X- rays of the chest, both old and new, were compared and a radiopaque shadow with mass effect was noted in the upper lobe of the right lung. Some rib crowding and displacement of the trachea and mediastinum to the opposite side was noticeable (Figures-1). A right lateral view of plain X- ray of the chest was obtained which showed a mass projecting forwards and having a push effect on the mediastinum. It also seemed to be attached

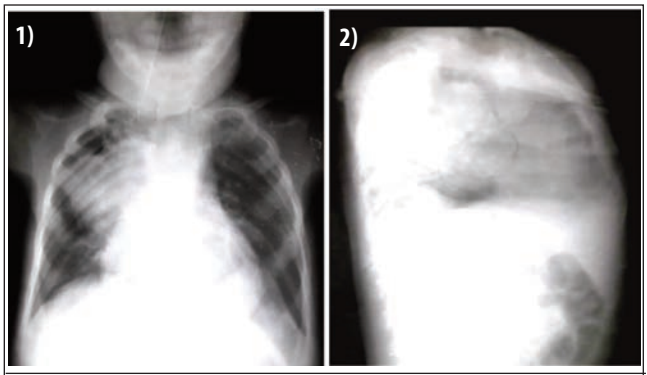

Figure-1: Images 1 and 2: Plain chest X-ray showing a mass in the right upper and middle zone with crowding of adjacent ribs, Plain X-ray chest on lateral view shows a posterior mediastinum mass which is displacing the oesophagus and the trachea forward.

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to the vertebral column. The expert report suggested a posterior mediastinal mass projecting into the right hemithorax with associated vertebral anomalies and peri-hilar early pneumonic patches. A CT Scan of the chest was advised.

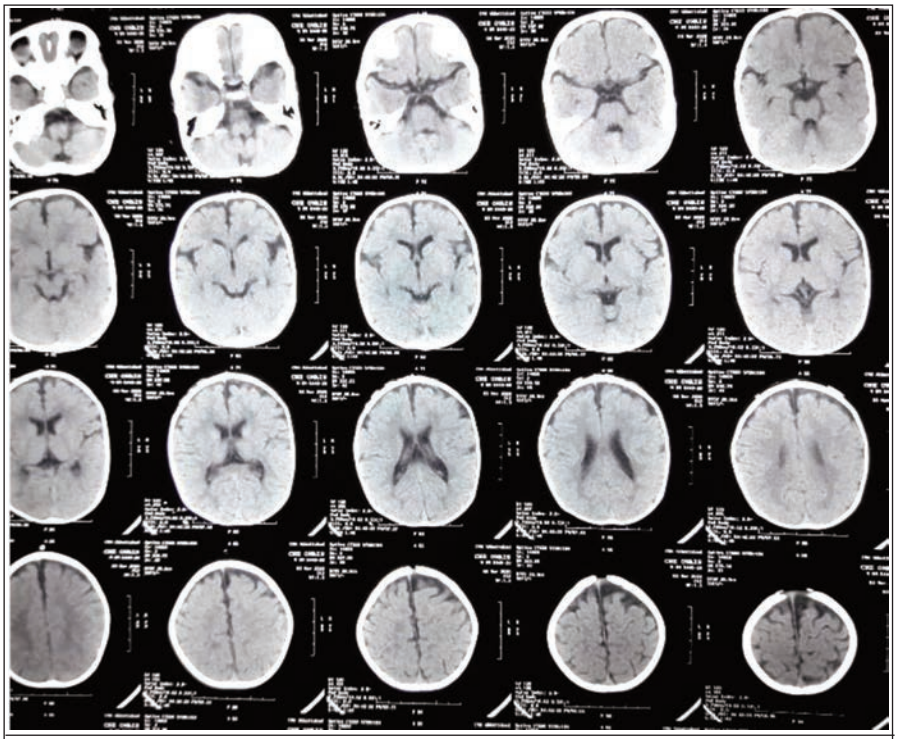

Figure-2: Plain CT scan of brain: Showing normal grey and white matter interface with mild frontal lobe atrophy.

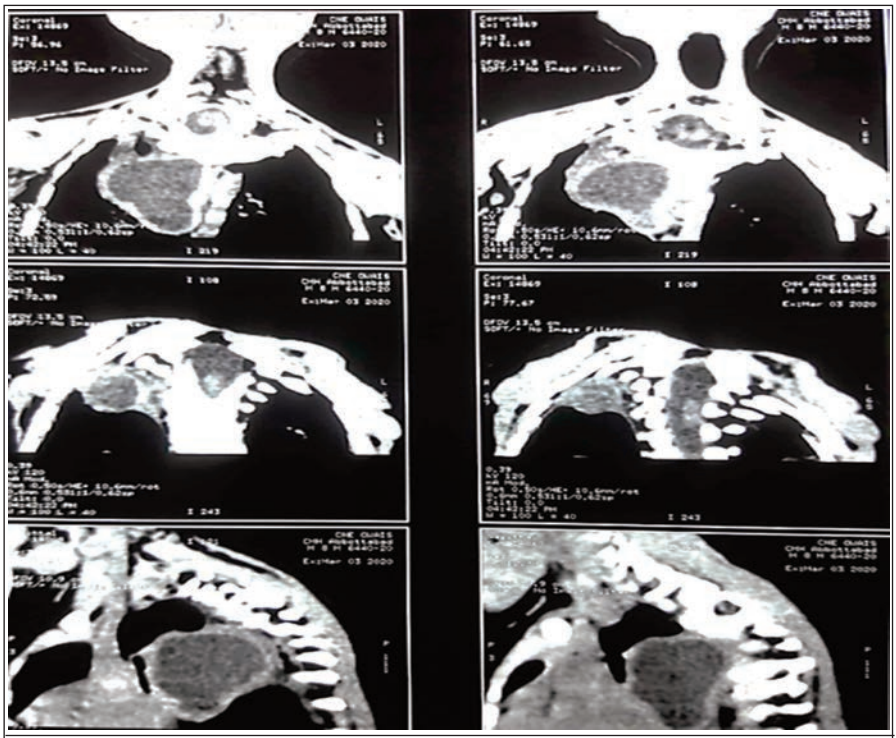

Figure-3: HRCT chest: Showing a large well-defined thick-walled cyst in posterior mediastinum on right side of oesophagus with pressure effects and vertebral anomalies.

Plain CT of the brain and spinal cord showed mild frontal anomaly and a large well-defined thick-walled cyst in the posterior mediastinum on the right side suggestive of foregut duplication cyst, most likely Neuroenteric cyst. Spina bifida with multiple deformed vertebral bodies involving dorsal thoracic vertebrae 1 to 7 and blocked thoracic vertebrae 1 and 2. A widening of central canal at the level of cervical vertebrae 7 likely representing syrinx was seen.

A detailed report of CT of the brain mentioned normal grey and white matter interface with mild frontal lobe atrophy, but no focal lesion, no evidence of mass effect, haemorrhage, SOL, or midline shift. Intracerebral CSF spaces appeared normal. Extra cerebral CSF spaces were dilated in the frontal region and the cerebral sulci were mildly prominent in bilateral front parietal region, suggesting some atrophy (Figure-2). The ventricular chain was normal; the brain stem, cerebellum, both Cerebellopontine angles, and visualised orbits were normal. Posterior fossa contents were normal, and no focal cerebellar lesions were noted. Spinous processes of thoracic spinal vertebrae 1 to 7 appeared unfused with their posterior elements being fused. Vertebral bodies at these levels were deformed with fusion of vertebral bodies of thoracic vertebrae 1 and 2. Focal dilatation of the central canal was also noted at this level, measuring about 1x0.5 cm in size, likely representing Syrinx. Widening of the central canal was noted at the level of cervical vertebrae 1 till thoracic vertebrae 4.

The final report of plain CT of the chest that showed a large cystic area in the right side of midline in the posterior mediastinum having mass effect on bronchus in carina abetting the oesophagus and dilated spinal canal of thoracic region with spina bifida of cervical 7 and thoracic 1 and 2 level,

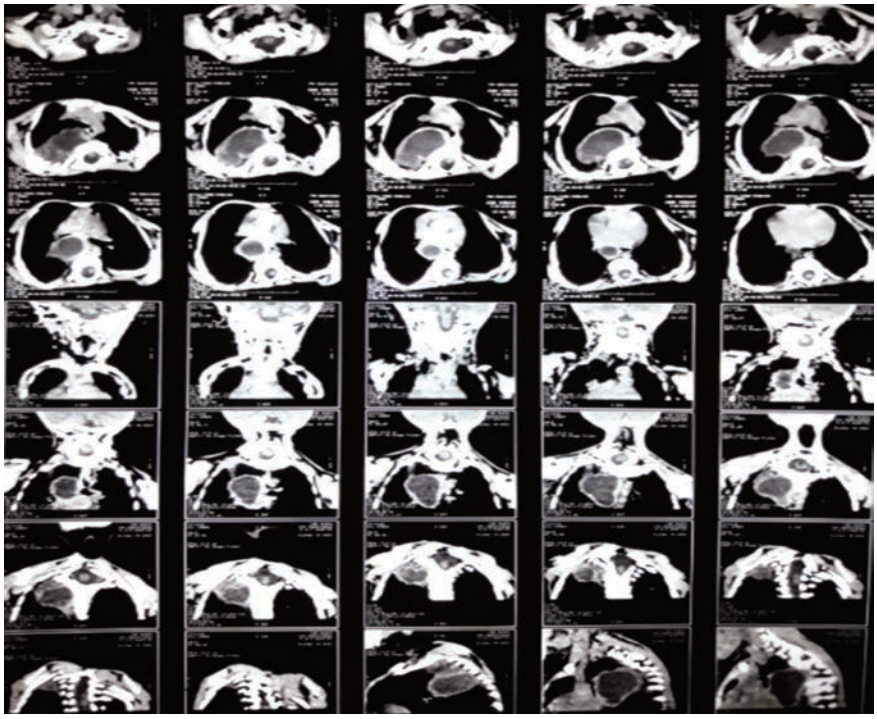

Figure-4: HRCT chest: Showing a large well-defined fluid density area with a thick wall in the posterior mediastinum on the right side of the oesophagus measuring 4.0 x 3.9 x 3.2cms and extending towards the right. Anteriorly, it is compressing the trachea and the right main bronchus near the carina displacing them anteriorly & medially displacing the thoracic mediastinum. Posteriorly it is extending up to the posterior chest wall. A spina bifida with spinal canal diameter at the level of 19mm is present. There is widening of spinal canal at Cervical 2 to Thoracic 4 level.

suggested a differential diagnoses of a foregut duplication cyst, bronchogenic cyst, and mediastinal neurenteric cyst.

An HRCT of chest was done. The HRCT report stated that a large well-defined fluid density area with a thick wall was noted in the posterior mediastinum on the right side of the oesophagus measuring 4.0x3.9x3.2 cms which was extending towards the right. Anteriorly, it was compressing the trachea and the right main bronchus near the carina displacing them, somewhat medially displacing the thoracic mediastinum; posteriorly, it was extending up to the posterior chest wall; the cystic mass was abutting the thoracic oesophagus, however, no communication was noted with the spinal canal (Figure-3). The spinal canal diameter was prominent in the thoracic region with prominent fluid in the canal with maximum diameter of 15mm. A spina bifida was also noted with spinal canal diameter at the level of 19 mm. No herniation of meninges or spinal contents was noted in the subcutaneous tissues. There was widening of spinal canal at Cervical 2 to Thoracic 4 level. The anteroposterior diameter was 1.5 cm. Prominent fluid was noted around spinal cord. (Figure-4).

Right-sided azygous fissure was noted. The great vessels and the cardiac chambers were normal. There was haziness in the lower lobe basal segments. The conclusion drawn was mild front parietal cerebral atrophy, cervical spina bifida occulta with dilation of cervical canal and prominent extra medullary intradural fluid around the spinal cord at cervical and upper thoracic level (cervical 1 to thoracic 4 vertebrae). In addition to fusion of dorsal thoracic vertebrae 1 and 2, no air fluid level or calcifications were seen in the cystic mass. Foregut duplication most likely mediastinal neurenteric cyst was suggested. On consultation the surgery department replied that no surgical intervention at the time was required; the parents were counselled and the patient was discharged on March 11, 2020 after a proper referral was placed. Patient was followed up till surgery , we were informed that unfortunately he had expired before being taken in the operation theatre.

Discussion

In mediastinal neurenteric cysts, the abnormal connection between the primitive ectoderm and endoderm result from a failure of complete separation of the notochord from the foregut. Discovered during the first five years of life, mediastinal neurenteric cysts can be seen anywhere from intracranial to the abdomen but mostly are located in the posterior mediastinum, cervical, and upper thoracic vertebrae.[6] These can be diagnosed antenatally, as early as 18 weeks of gestation.[7] Two thirds of all mediastinal cysts are related to compression of the adjoining structures and obstruction of distal lungs.[8] There are three subtypes of foregut duplication cysts—first, enterogenous cysts lined by intestinal epithelium; second, bronchogenic cysts with respiratory epithelium; and third, the mediastinal neurenteric cysts associated with apparent vertebral anomalies.[6] Their clinical presentation and radiological interpretations are always challenging to the paediatrician.

The clinical presentation of mediastinal cysts depends on its size, shape, and location within mediastinum. An infant with mediastinal neuroenteric cyst becomes symptomatic as the cyst expands due to fluid accumulation and starts compressing the adjoining structures. The signs and symptoms are primarily due to such mass effect. Respiratory symptoms like cough, shortness of breath,

stridor, and distress are more frequent clinical presentations. Besides respiratory symptoms leading to distress, mediastinal mass and spina-bifida form a triad of Neuroenteric cysts.[9]

Another dilemma for the paediatricians is how to diagnose such a congenital mass post-natally. The presence of Meningocele, cystic extension into vertebral foramen or spinal canals without calcification and vertebral involvement in mediastinal area (like the patient under discussion) suggest a mediastinal neurenteric cyst. Moreover, such a neurenteric cyst may present with spina bifida, as in the current case. In other condition, like presence of calcification and absence of vertebral involvement and mass in the same location is more likely to be a bronchogenic cyst. Moreover, a foregut duplication cyst is usually present along the lower third of the thoracic oesophagus with a predilection to the right chest. But they all need CT or HRCT to differentiate, as these can't be differentiated only on anteroposterior or lateral chest x-rays. In the current case, the mediastinal cyst can be characterised as Neuroenteric. We strongly recommend CT scan imaging modality to be used as gold standard for diagnosing posterior mediastinal mass with suspension of Neuroenteric cyst.[10]

It is well established that spinal and neurological complications tend to characterise almost exclusively the neurenteric cysts. Mediastinal neurenteric cysts can be associated with cervical and upper thoracic vertebral anomalies such as scoliosis, anterior spina bifida, hemi vertebrae, butterfly vertebrae or vertebral fusion. The vertebral anomaly is commonly cephalad to the cyst as the oesophagus descends during foetal development.[11]

Surgery is the definitive treatment of these lesions and is generally curative. A patient reported by Neuhauser et al with flaccid paralysis of the lower extremities experienced return to normal function following resection of the mass and severance of its communication with thoracic spinal canal.[12] The condition can cause life-threatening respiratory distress and surgery is essential to improve severe symptoms.[13]

Conclusion and suggestions

Mediastinal neurenteric cysts are a rarity and were noted for the first time in our clinical encounter of children presenting with chest problems. Respiratory symptoms leading to distress, mediastinal mass and spina-bifida can form a triad of Neuroenteric cysts. Among imaging modalities, CT scan is recommended for diagnosis of Neuroenteric cysts with prompt surgical intervention. Not considering these rules of thumb can delay life-saving surgery and lead to serious morbidity and mortality.

Consent: Consent of patients of the patient was obtained to publish the case.

Disclaimer: None.

Conflicts of interest: None.

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References

1. Wilson ES Jr. Neurenteric cyst of the mediastinum. Am J Roentgenol Radium Ther Nucl Med 1969;107:641-6. doi: 10.2214/ajr.107.3.641.
2. Rizalar R, Demirbilek S, Bernay F, Gürses N. A case of a mediastinal neurenteric cyst demonstrated by prenatal ultrasound. Eur J Paediatr Surg 1995;5:177-9. doi: 10.1055/s-2008-1066199.
3. Saleem M, Sadiq J, Rauf A, Shaukat M. Intra-Spinal Enteric Cyst. Pak J Paed Surg 1999;180:40-8.
4. Khadim MT, Asif M, Ali Z. Intraspinal neurenteric cyst--a rare entity. J Pak Med Assoc 2011;61:1143-5.
5. Sagheer S, Amir Humza Sohail AM, Hadi YB, Fatimi S. Enteric cyst in the left posterior mediastinum mimicking a hydatid cyst on chest Computed Tomography scan. J Pak Med Assoc 2016;66:363-5.
6. Setty H, Hegde KK, Narvekar VN. Neurenteric cyst of the posterior mediastinum. Australas Radiol 2005;49:151-3. doi: 10.1111/j.1440-1673.2005.01360.x.
7. Aydin K, Sencer S, Barman A, Minareci O, Hepgul KT, Sencer A. Case report: Spinal cord herniation into a mediastinal neurenteric cyst: CT and MRI findings. Br J Radiol 2003;76:132-4. doi: 10.1259/bjr/60547861.
8. Patel SR, Meeker DP, Biscotti CV, Kirby TJ, Rice TW. Presentation and management of bronchogenic cysts in the adult. Chest 1994;106:79-85. doi: 10.1378/chest.106.1.79.
9. Wilkinson CC, Albanese CT, Jennings RW, Feldstein VA, Goldberg JD, Baehner FL, Farrell JA, Peacock WJ, Harrison MR. Fetal neurenteric cyst causing hydrops: case report and review of the literature. Prenat Diagn 1999;19:118-21. doi: 10.1002/(sici)1097-0223(199902)19:2<118: aid-pd478>3.0.co;2-5.
10. Kouerinis IA, Zografos GC, Exarchos DN, Silimingas NT, Argiriou ME, Manoussaridis JT, et al. A huge posteromedial mediastinal cyst complicated with vertebral dislodgment. World J Surg Oncol 2006;4:56. doi: 10.1186/1477-7819-4-56.
11. Kirwan WO, Walbaum PR, McCormack RJ. Cystic intrathoracic derivatives of the foregut and their complications. Thorax 1973;28:424-8. doi: 10.1136/thx.28.4.424.
12. Neuhauser EB, Harris GB, Berrett A. Roentgenographic features of neurenteric cysts. Am J Roentgenol Radium Ther Nucl Med 1958;79:235-40.
13. Turkyilmaz A, Aydin Y, Erdem AF, Eroglu A, Karaoglanoglu N. Congenital cystic pulmonary malformations in children: our experience with 19 patients. Eurasian J Med 2009;41:15-21.