Original Research Paper

Volume - 12 | Issue - 01 | January - 2022 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Respiratory Medicine

ALPHA - 1 ANTITRYPSIN DEFICIENCY - AN UNDER RECOGONIZED CAUSE OF EMPHYSEMA

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ABSTRACT Alpha 1 antitrypsin deficiency is a hereditary disorder characterized by low circulating levels of AAT. A case of 36 year old male patient, chronic alcoholic presented with acute abdominal pain vomiting & breathlessness, having family history of emphysema .On examination chest was barrel shaped with hyper resonant note on percussion and decreased breath sound on auscultating bilateral lung fields. Per abdomen was tender with voluntary guarding and diminished bowel sounds , later diagnosed to have acute pancreatitis according to raised serum amylase, lipase level& USG abdomen reports. Patient was treated with iv antibiotic and continuous iv fluids. Routine blood investigation, ECG and 2D echo was normal and Patient had respiratory acidosis with hypercapnia on ABGA and CXR shows hyperinflated lung field and flattened diaphragm. HRCT thorax shows hyperinflated bilateral lung parenchyma & paucity of bronchovascular marking in bilateral lower lobe. PFT showed moderate obstructive pattern with poor bronchodilator reversibility. Serum Alpha 1 antitrypsin level was low. Patient was given symptomatic treatment of COPD & later get discharged. There is a large delay between symptom onset and AATD diagnosis.

KEYWORDS :

INTRODUCTION:

Chronic obstructive pulmonary disease is a common, preventable and treatable disease that is characterized by persistent respiratory symptoms and airflow limitation that is due to airway and/or alveolar abnormalities usually caused by significant exposure to noxious particles or gases. Besides exposure, host factors predispose individuals to develop COPD, which include abnormal lung development and genetic abnormalities such as alpha 1 antitrypsin deficiency.

Alpha 1 antitrypsin deficiency is a hereditary disorder characterized by low circulating levels of AAT and is associated by development of COPD often by 3rd or 4th decade and liver disease as the AAT is a secretory glycoprotein produced by liver. Prevalence of emphysema in AATD is 21% [2]. The most common SERPIN A1 mutation associated with AATD are Z & S mutation and vast majority of AATD individual diagnosed with COPD are ZZ homozygotes.

Case:

A case of 36 year old male patient, chronic alcoholic presented with acute abdominal pain, vomiting & breathlessness for less than 1 week. Abdominal pain was dull, boring, radiating to back. Breathlessness was of mMRC grade 2. Patient's father is having a history of emphysema for which he had taken treatment. On examination chest was barrel shaped with hyper resonant note on percussion.

Breath sound on auscultating bilateral lung fields was decreased. Per abdomen was tender with voluntary guarding and diminished bowel sounds, later diagnosed to have acute pancreatitis according to raised serum amylase, lipase level& USG abdomen reports. Patient was treated with iv antibiotic and continuous iv fluids. Routine blood investigation, ECG and 2D echo was normal. ABGA showed respiratory acidosis with hypercapnia so that he was kept on non invasive ventilation for 5 days followed by oxygen inhalation with nasal prongs for 3 days.

DIAGNOSIS & DISCUSSION:

On CXR lung field were hyperinflated and diaphragm flattened. HRCT thorax shows hyperinflated bilateral lung parenchyma & paucity of bronchovascular marking in bilateral lower lobe. PFT showed moderate obstructive pattern with poor bronchodilator reversibility. Alpha 1 antitrypsin level was 0.7 (N= 0.9-2.0). Patient was given symptomatic treatment of COPD with bronchodilators & later get discharged.

CONCLUSION:

Even though COPD cases are more commonly seen in old age patient with smoking history or with history of exposure to occupational dust, it can also be rarely presented in young age group with a genetic component. Any young patient with family history of emphysema presenting with emphysema in HRCT thorax and pancreatitis should be suspected for AATD even though it is a rare case. There is a large delay between symptom onset and AATD diagnosis.

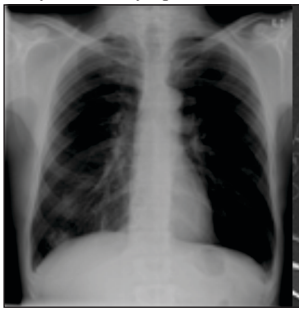

Figure 1: Chest Xray

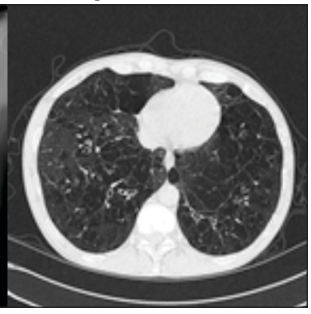

Figure 2: Hrct Thorax

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