Rare Association of Childhood Bronchiectasis with VACTERL Anomalies; a Case Report

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Abstract

VACTERL association is a sporadic non-random cluster of multi-organ congenital anomalies. It is an acronym that describes the association of vertebral defects (V), anal atresia (A), cardiac defects (C), tracheoesophageal fistula (TE), renal (R) and limb (L) anomalies. Three or more anomalies should be present to make the diagnosis of VACTERL association.Bronchiectasis is an uncommon respiratory disease in childhood. It is often associated with underlying aetiology. Bronchiectasis has been described in association with VACTERL associations extremely rarely.We report an 8-year-old girl with known multiple congenital anomalies including tracheoesophageal fistula, atrial septal defect and imperforate anus presented with recurrent respiratory infections since 06 months of age, who was diagnosed with bilateral bronchiectasis later. The diagnosis of bronchiectasis in association with VACTERL anomalies was made following exclusion of specific aetiology with extensive investigations.This case demonstrates the rare association of bronchiectasis with VACTERL anomalies.

Keywords: *VACTERL association, bronchiectasis*

Introduction

VACTERL association is a recognizable group of multi-organ congenital anomalies that tend to co-exist in a single patient.(1) This was described in the early 1970s. It is a rare disease with an estimated incidence of 1/10000 to 1/40000.(2) VACTERL association is defined as co-existence of 3 or more of following congenital anomalies; vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal and limb anomalies.

Pulmonary involvement in the form of bronchiectasis has been described in association with VACTERL anomalies extremely rarely. (3)

Case report

An 8 years old Sri Lankan Muslim girl was presented to a respiratory disease treatment unit, Teaching hospital-Kandy, Sri Lanka with a history of recurrent episodes of a cough with shortness of breath since 06 months of age.

She was a product of non-consanguineous parents and the eldest of two other healthy siblings.

She had a complicated medical history accompanied by multiple congenital anomalies. She had been diagnosed to have a tracheoesophageal fistula, imperforate anus with any-vestibular fistula and ostium secundum atrial septal defect since birth. Her tracheoesophageal fistula had been corrected at 16 days of age, and imperforate anus had been corrected surgically during the first year. The atrial septal defect was followed up with serial echocardiographic monitoring and noted to be closed spontaneously by the age of 7 years.

She had been suffered from recurrent lower respiratory tract infections occurred with a frequency of once a month approximately. These episodes were characterized by productive type cough, shortness of breath, wheezing and occasionally with fever. They had been managed non-specifically with supportive treatment.

She denied recurrent aspirations or features of gastroesophageal reflux disease. She had been vaccinated age appropriately. There were no similar conditions in her family.

An examination of respiratory system revealed bilateral rhonchi with few coarse crepitations. Her cardiovascular, neurological and abdominal system examinations were within normal limits.

Figure 01

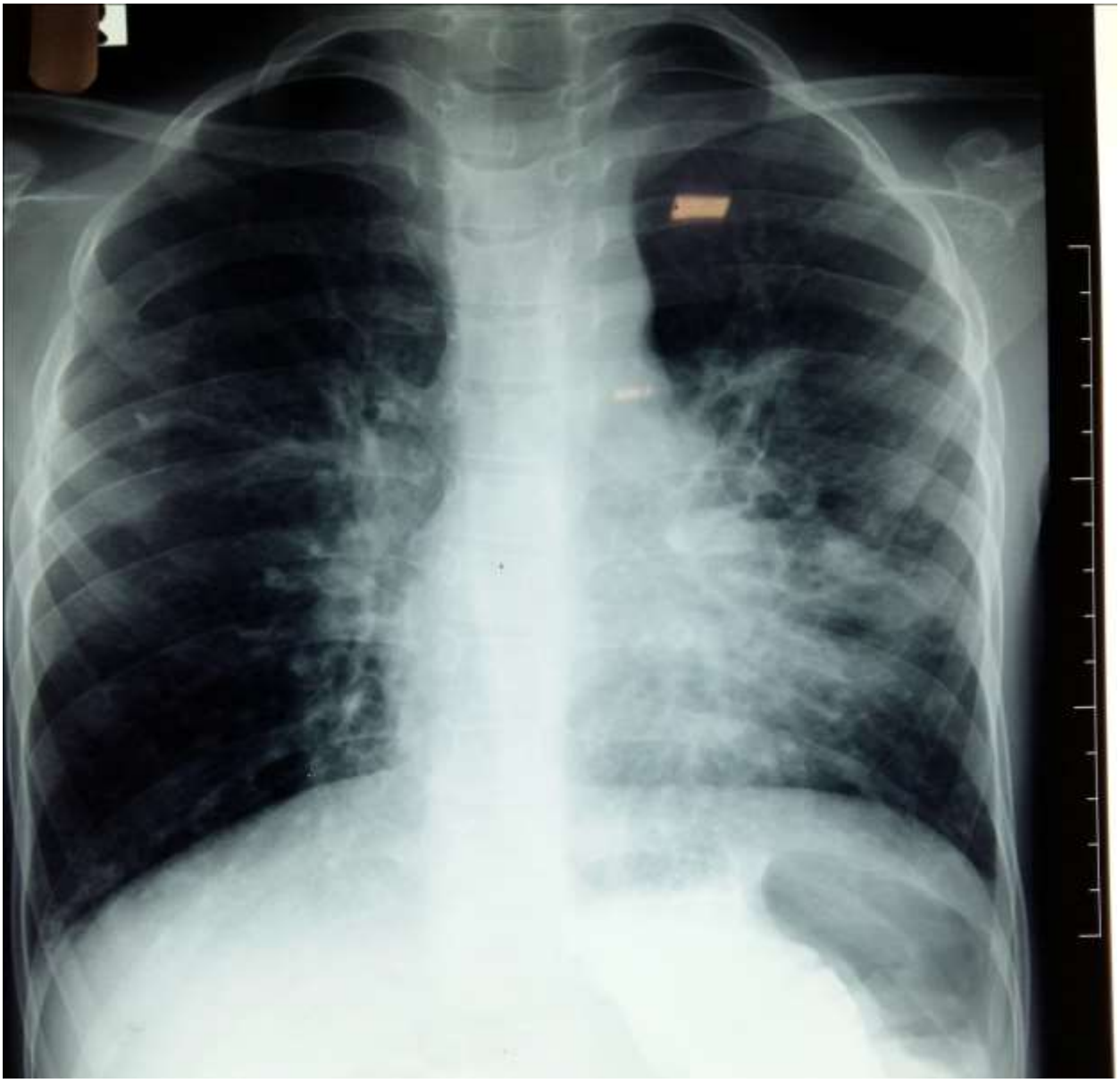

Figure 02

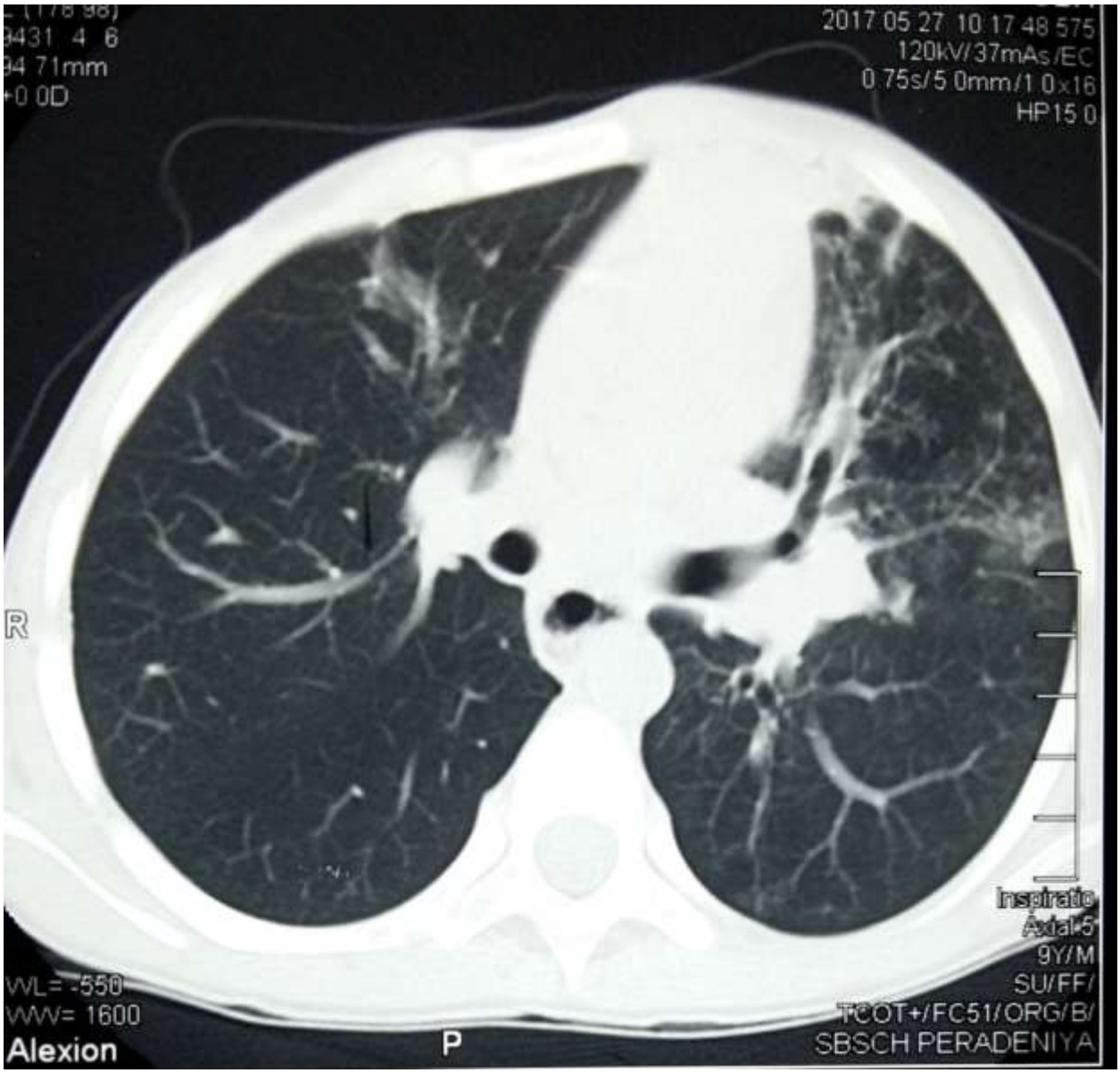
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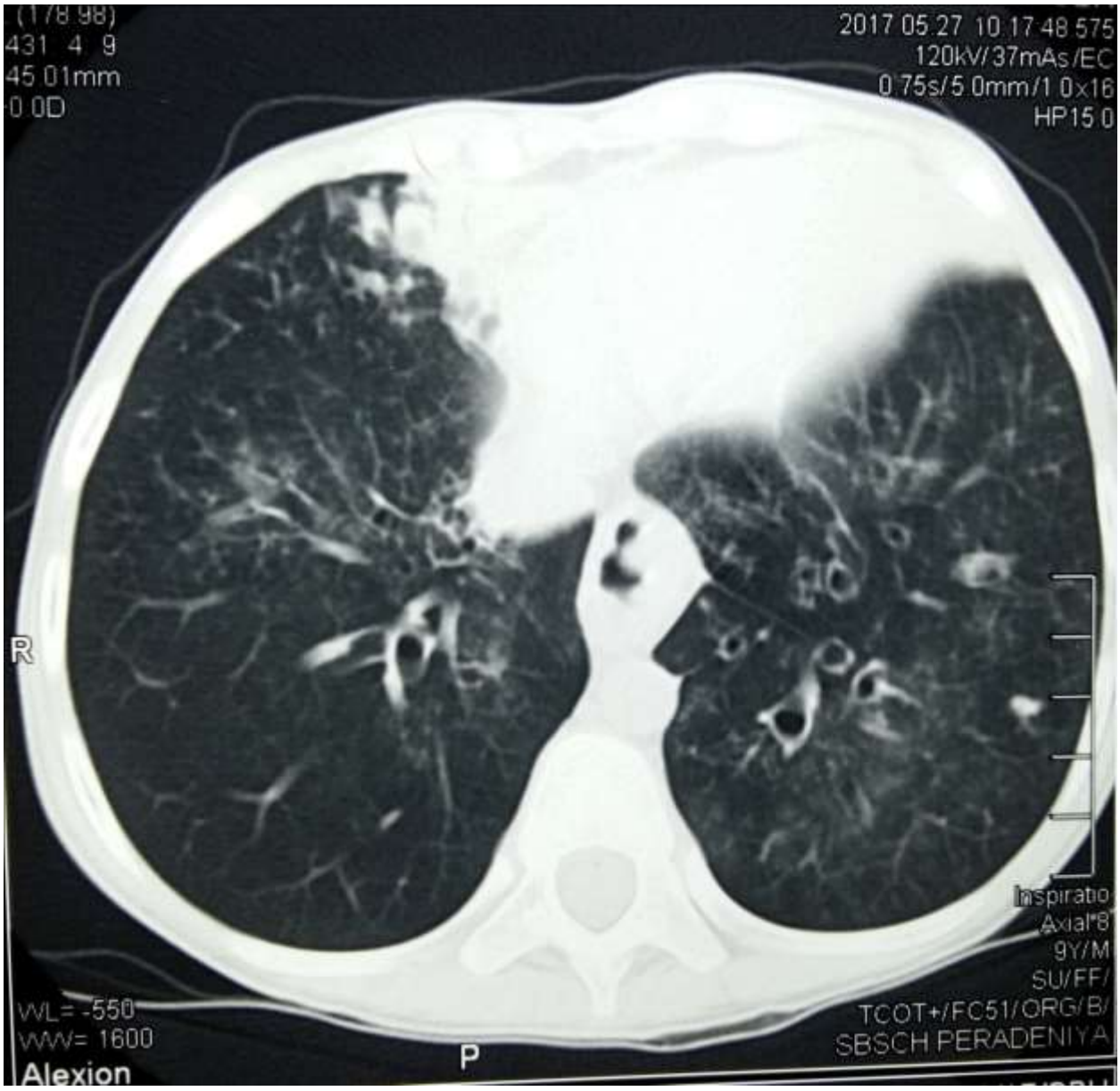
Figure 03



Her chest X-ray showed features of bronchiectasis involving the left lingular lobe. (Figure 01) High resolution computed tomography revealed early bronchiectatic changes involving predominantly right middle lobe and left lingular lobe and to a lesser degree bilateral lower lobes. (Figure 02) Additionally, there was evidence of diffuse pulmonary emphysema and secondary infection. She was extensively investigated for underlying secondary cause for bronchiectasis. She had mild anemia with evidence of beta thalassemia trait. Total white cell count with differential count, platelets, renal functions, liver functions were within normal limits. Investigations for immunodeficiency state including immunoglobulin electrophoresis complement levels, CD3, CD4, CD8, CD19 counts, neutrophil activity, antibody response to tetanus toxoid and human immunodeficiency virus serology did not find a significant abnormality. Eosinophil count and serum immunoglobulin E levels were within normal range. Cystic fibrosis transmembrane receptor mutation was not detected. Alpha1 antitripsin levels were not assigned due lack of resources. Bronchoscopic examination revealed thick secretions within both bronchi. Bronchoalveolar lavage was negative for acid-fast bacilli, Mycobacterium tuberculosis Xpert/RIF test,

tuberculosis culture and fungal studies. Due to the high prevalence of tuberculosis in Sri Lanka, she was treated for presumed tuberculosis for six months according to common clinical practice; however, this did not improve her symptoms. She was managed with supportive treatment including inhaled steroids and bronchodilators, chest physiotherapy and infection prevention strategies.

Even following extensive evaluation, the aetiology for bronchiectasis was not identified in this patient. So, this was considered to represent an extremely rare association of bronchiectasis with VACTERL anomalies.

Discussion

Bronchiectasis is a chronic suppurative lung pathology caused by many aetiologies. The term bronchiectasis describe a permanent dilatation of bronchioles as a result of destruction of muscles and elastic connective tissues.(4)

Bronchiectasis is characterized by chronic productive cough, wheezing, dyspnoea, recurrent chest infections and occasionally haemoptysis. (4, 5, 6) HRCT is the diagnostic investigation of choice. (6) Dilatation of an airway lumen, lack of tapering of an airway toward the periphery, varicose constrictions along airways are considered as specific abnormalities suggestive of bronchiectasis.(5)

Bronchiectasis has many known aetiologies including chronic infections, autoimmune disorders, primary and secondary immunodeficiencies, genetic disorders, ciliary dyskinasia and anatomical derangements as major categories.

Our patient presented with recurrent chest infections since very early in her life and developed bronchiectasis at childhood. This suggests genetic diseases or congenital abnormalities as the most probable cause of her disease. However, investigations failed to identify possible secondary aetiology.

Bronchiectasis has been extremely rarely described to associate with VACTERL anomalies. The exact reason for this association is not known. Possible mechanisms include recurrent aspirations, cardiac anomalies, and vertebral defects. (3)

Conclusion

Bronchiectasis is an uncommon disease in children often due to secondary aetiology. VACTERL anomalies have rarely been described to associate with bronchiectasis. Clinicians should be aware of this association for early diagnosis and optimal management to prevent advanced lung disease.

Abbreviations

HRCT- High resolution computed tomography

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal

Competing interest

The authors declare that they have no competing interest.

Authors' contribution

DM and SN made the clinical diagnosis and supervised the manuscript drafting. ARB drafted the first manuscript, reviewed the literature and involved in direct management of the patient. DM and SN supervised the manuscript drafting.RMDHMR helps writing and literature survey . All authors read and approved the final manuscript.

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