

## WILLIAMS-CAMPBELL SYNDROME- A RARE ENTITY OF CONGENITAL BRONCHIECTASIS: A CASE REPORT IN ADULT

Sukla Mitra<sup>1</sup>, Anadi Roy Chowdhury<sup>2</sup>, Goutam Bandyopadhyay<sup>3</sup>

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**Author details:** <sup>1</sup>Senior Resident, <sup>2</sup>Associate professor, <sup>3</sup>Professor, Department of Pathology, R G Kar Medical College & Hospital, Kolkata-700004

**Corresponding author:** Department of Pathology, R G Kar Medical College & Hospital, Kolkata-700004  
Email: [sukla21feb@gmail.com](mailto:sukla21feb@gmail.com)

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### ABSTRACT

Williams-Campbell syndrome is a rare entity of congenital bronchiectasis due to developmental arrest in the tracheobronchial tree, in which extensive loss of bronchial cartilage is associated with diffuse cystic bronchiectasis; may be unilateral or bilateral. Clinical manifestations start from infancy with subsequent recurrent pulmonary infection leading to respiratory failure. Patients may survive into late adulthood and require lung transplantation. We report such a rare case diagnosed on the basis of clinico-radiological presentation and histopathological examination of the pneumonectomy specimen. A 40 years patient was presented with severe breathlessness and had history of recurrent episode of productive cough with low grade fever since childhood for which he was admitted in hospital and treated symptomatically. Chest roentgenogram revealed right hyperlucent lung, deviation of trachea towards left; while the left lung showed multiple scattered large thin walled cysts along with elevation of left dome of diaphragm. High resolution computed tomography revealed multiple cystic thin walled airways in the left hemithorax, suggestive of bronchiectasis with collapse of left lung and compensatory hyperinflation of right lung along with herniation of right upper lobe to the left.

### INTRODUCTION

Bronchiectasis is the irreversible dilatation of bronchi and bronchioles caused by destruction of smooth muscle and elastic tissue, resulting from chronic necrotizing infections. Williams-Campbell syndrome is a rare developmental disorder of familial occurrence which results absence or deficiency of cartilage in the bronchial walls distal to first divisions of subsegmental bronchi and associated with diffuse cystic bronchiectasis. This uncommon entity should not be confused with congenital bronchiectasis which are those of hereditary conditions, such as cystic fibrosis, primary ciliary dyskinesia or immunodeficiency states; that predispose to subsequent development of bronchiectasis. On chest radiograph large thin walled cysts are found; while high resolution computed tomography (HRCT) scan characteristically shows central, cystic, thin-walled airways that collapse upon expiration. Microscopic studies document, dilated airways having thin walls, absent or deficient cartilage with minimal inflammation.<sup>[1]</sup> Although most cases presented in childhood, some sporadic subclinical cases maybe diagnosed in adults as well.<sup>[2,3]</sup> Here we report such a rare case that had typical clinico-radiological presentation as well as histopathological features of congenital bronchiectasis.

### CASE REPORT

We received pneumonectomy specimen of left lung of a 40 years old male patient from the department of cardiothoracic and vascular surgery of our hospital and retrospectively collected the clinical history and

investigation reports. The patient was presented with severe breathlessness and had history of recurrent episode of productive cough with low grade fever since childhood for which he was admitted in hospital and treated symptomatically with oxygen, mucolytics, bronchodilators and antibiotics. But, he had no history of hemoptysis, joint pain or chronic gastrointestinal symptoms. He smoked five cigarettes per day for last twelve year and had no family history of lung disease. Despite manifestation he declined medical evaluation until the age of 24 year. On admission, he had coarse crepitations with rhonchi and decreased air entry in left lung field and clubbing. His routine investigations were normal and serum was negative for Human Immunodeficiency Virus 1 and 2 antibodies. Sputum was negative for acid-fast bacilli (AFB) on multiple occasions but culture was positive for *Klebsiella* species. Chest roentgenogram revealed right hyperlucent lung, deviation of trachea towards left; while the left lung showed multiple scattered large thin walled cysts along with elevation of left dome of diaphragm (Figure 1A). High resolution computed tomography (HRCT) revealed multiple cystic thin walled airways in the left hemithorax, suggestive of bronchiectasis with collapse of left lung and compensatory hyperinflation of right lung along with herniation of right upper lobe to the left (Figure 1B). Echocardiography was normal.



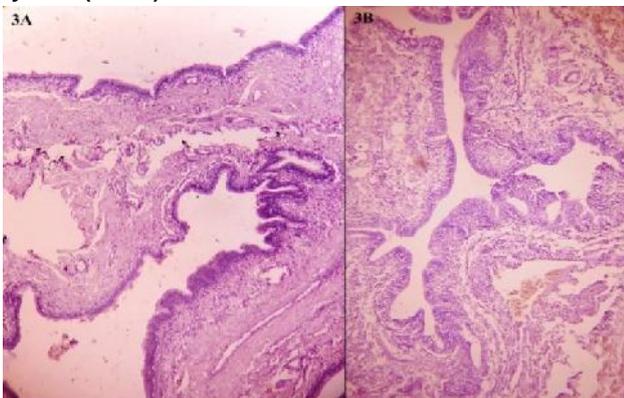
**Fig 1: (A) Chest roentgenogram showed hyperlucent right lung with multiple scattered cystic spaces in left lung field and elevation of left dome of diaphragm (arrow). (B) HRCT shows cystic spaces in left hemithorax suggestive of bronchiectasis with collapse of left lung and herniation of right upper lobe to left.**

Spirometry showed severe restrictive type of lung function and sera was positive for *Aspergillus* allergen specific antibodies (IgE) recently, but it was negative on previous occasions. Other serum immunoglobulins were within normal limits. Culture of broncho-alveolar lavage (BAL) was negative for AFB, but positive for methicilin sensitive *Staphylococcus aureus*.

He underwent left pneumonectomy. Gross specimen revealed a small non-expansile fibrotic lung measuring 12 x 8 x 5 cm and weighing 150 g. The cut section revealed a dilated bronchial system (Figure 2A & 2B). Microscopic findings shows, dilated airways having thin walls with minimal inflammation and deficient cartilage (Figure 3A&3B).



**Fig 2: Pneumonectomy specimen of left lung shows: (A) Pleural surface (B) Cut section shows dilated bronchial system (arrow).**



**Fig 3: Histopathological sections (A) & (B) – dilated, irregularly shaped airway lumen shows absence of cartilage throughout the airway walls with minimal inflammation; Hematoxylin-Eosin stain with magnification 100x.**

## DISCUSSION

Williams-Campbell syndrome is a rare form of familial disorder in which deficiency of bronchial cartilage is associated with diffuse cystic bronchiectasis without other recognized predisposing factors.<sup>[1,4-6]</sup> Cartilage is absent or deficient from fourth to eighth divisions of subsegmental bronchi; while normal amount of cartilage present in first and second order bronchi or other body parts. The cause of this cartilage deficiency is still uncertain.<sup>[1]</sup> Symptoms like cough, dyspnea on exertion, cyanosis and clubbing appears from infancy.<sup>[1,7]</sup> The prognosis varies from rapid clinical deterioration and death in some, while prolonged survival in others.<sup>[4]</sup> The survived patients may developed recurrent pulmonary infections; often present acutely necessitating emergent evaluation and requires lung transplantation.<sup>[8,9]</sup> However, Williams-Campbell syndrome remains a controversial entity, and its congenital origins have yet to be proven beyond question.<sup>[10-12]</sup>

Chest radiograph of this patient shows large thin walled cysts, while HRCT scan characteristically reveals central, cystic, thin walled airways that collapse upon expiration.<sup>[2,13,14]</sup> Microscopic findings shows, dilated airways having thin walls with minimal inflammation and deficient cartilage.<sup>[1]</sup>

In 1960, Williams and Campbell reported five unusual cases of bronchiectasis, with soft, compliant bronchi having deficient cartilage, which dilated and collapsed respectively during inspiration and expiration.<sup>[5]</sup>

The diagnosis of Williams Campbell Syndrome requires an appropriate clinical history, characteristic expiratory collapse of airways and exclusion of other causes of congenital or acquired bronchiectasis such as cystic fibrosis, allergic bronchopulmonary aspergillosis (ABPA), immune deficiency, tracheobronchomegaly.<sup>[7,9]</sup>

The patient was presented with destruction of left lung with cystic changes, but trachea and main bronchi were normal, which rules out tracheobronchomegaly. The differential diagnosis includes ABPA or cystic fibrosis. The antibodies against aspergillus allergen were negative on previous results, but became positive for short duration and that may be due to the result of recurrent pulmonary infection which is a major complication of this entity. Absence of gastrointestinal symptoms during the course of illness ruled out possibilities of cystic fibrosis and lastly, the extent and distribution of the bronchial abnormalities are not consistent with that of cystic fibrosis and ABPA.<sup>[15]</sup>

The case discussed in this article was managed medically for long duration, which developed complications and required pneumonectomy. These scenarios can be prevented by early diagnosis and more importantly by recognizing the existence of these lesions as a separate entity.

## CONCLUSION

Congenital lung disorders are increasingly diagnosed in adults. Williams Campbell Syndrome, although a rare congenital anomaly; it should be recognise as a separate entity and needs to be considered in the differential diagnosis of bronchiectasis.

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## REFERENCE

1. Tomaszefski J F, Dail D H. Aspiration, Bronchial Obstruction, Bronchiectasis, and Related Disorders. In: Tomaszefski J F, editor. *Dail and Hammar's pulmonary Pathology, Nonneoplastic lung Disease vol 1*. Third Edition. New York: Springer; 2008.p: 99-146.
2. Newman KB, Beam WR. Congenital bronchiectasis in an adult. *Am J Med* 1991;91:198-201.
3. Przerwa KB, Bestry I, Gawryluk D, Wiatr E. Case report: Williams Campbell syndrome. *Pol J Radiol* 2009;74:76-78.
4. Jones VF, Eid NS, Franco SM, Badgett JT, Buchino JJ. Familial congenital bronchiectasis: Williams-Campbell syndrome. *Pediatr Pulmonol* 1993;16:263-7.
5. Williams H, Campbell P. Generalized bronchiectasis associated with deficiency of cartilage in the bronchial tree. *Arch Dis Child* 1960;35: 182-191.
6. Davis PB, Hubbard VS, McCoy K, et al. Familial bronchiectasis. *J Pediatr* 1983;102:177-185.
7. Williams HE, Landau LI, Phelan Po. Generalized bronchiectasis due to extensive deficiency of bronchial cartilage. *Arch Dis Child* 1972;47:423-428.
8. Palmer SM Jr, Layish DT, Kussin PS, et al. Lung transplantation for Williams-Campbell syndrome. *Chest* 1998;113:535-537.
9. Wayne KS, Laussig LM. Probable familial congenital bronchiectasis due to cartilage deficiency. *Am Rev Resp Dis* 1976;114:15-22.
10. Hayward J, Reid LM. The cartilage of the intrapulmonary bronchi in normal lungs, in bronchiectasis, and in massive collapse. *Thorax* 1952;7:98-110.
11. Ogrinc G, Kampalath B, Tomaszefski IF Jf. Destruction and loss of bronchial cartilage in cystic fibrosis. *Hum Pathol* 1998;29:65-73.
12. Whitwell E Study of pathology and pathogenesis of bronchiectasis. *Thorax* 1952;7:213-239.
13. Gupta N, Pulinilkunnathil JG, Dixit R, Gupta R. A rare case of congenital bronchiectasis – Williams Campbell Syndrome. *DiagTher Stud* 2012;1(1):1-6.
14. McAdams HP, Erasmus I. Case 4: Williams-Campbell syndrome. *AJR* 1995;165:190-191.
15. Hartman TE, Primack SL, Lee KS, Swensen SJ, Muller NL. CT of bronchial and bronchiolar diseases. *Radiographics* 1994;14:991-1003.