A RARE CASE OF CONGENITAL BRONCHIECTASIS - WILLIAMS CAMPBELL SYNDROME

Gupta N[1], Pulinilkunnathil JG[1]*, Dixit R[1], Gupta R[1]

1. JLN Medical College Ajmer, India

Correspondence
Jacob George Pulinilkunnathil. JLN Medical College Ajmer, India
Email: hipulian@yahoo.co.in

Gupta N, Pulinilkunnathil JG, Dixit R, Gupta R. A rare case of congenital bronchiectasis – Williams Campbell Syndrome. Diag Ther Stud. 2012; 1(1):1 - 6.

ABSTRACT

Williams Campbell Syndrome, also known as Bronchomalacia, is a rare disorder of bronchial cartilage development. Because of this mechanical disadvantage, the patients develop recurrent respiratory symptoms early in the infancy or childhood, representing radiologically as widespread saccular bronchiectasis early in the life. We report such a rare case diagnosed on the basis of clinicoradiological presentation and confirmed on computed tomography.

Key words: Williams Campbell Syndrome, bronchomalacia, saccular bronchiectasis

INTRODUCTION

Bronchiectasis is a condition characterized by the abnormal permanent dilation of bronchi with destruction of the muscular and elastic components of their walls, usually due to acute or chronic infection. While most of the cases of bronchiectasis we come across are probably acquired in childhood because of recurrent lower respiratory tract infections, the condition may rarely result from a gross congenital developmental anomaly (defect in cartilage development) or predisposed by other inherited ultra structural defects e.g. ciliary dyskinesia, cystic fibrosis or due to autoimmune deficiency syndrome e.g. hypogamma-globulinemia. Regardless of the cause, chronic cough and sputum production are the main complaints among such patients. Bronchography, once considered the gold standard for diagnosing bronchiectasis, has now widely been replaced by high resolution computerized tomography (HRCT). The sensitivity and specificity of HRCT exceeds 90% and also lacks the risk and unpleasant invasive nature of bronchography[1].

An extensive but uncommon form of bronchiectasis is Williams - Campbell Syndrome (Bronchomalacia) which basically is congenital bronchi malacia caused by absence of annual cartilage distal to first division of peripheral bronchi. The term is given to a constellation of airway collapse during expiration, bronchiectasis and bronchial cartilage

deficiency[2, 3]. Although the syndrome has been described in children, sporadic and familial cases do occur in adults as well[4].
Here we report such a rare case who presented with typical clinical and radiological features to suggest the diagnosis of Bronchomalacia.

CASE REPORT

A 24-year, unmarried male, presented with history of breathlessness on exertion and recurrent episodes of cough, fever and sputum production since birth. He gave history of recurrent hospital admissions for breathlessness and sputum production, where he was treated symptomatically with antibiotics, oxygen and inhaled bronchodilators. There was no history suggestive of chronic rhino sinusitis He also denied history of joint pains, hemoptysis, seasonal or other atopic symptoms and also gastrointestinal symptoms. He also denied history of exposure to pets or animals in house or in surrounding.
On examination, he was well built but had a BMI of only 18. The patient was tachypnoiec and accessory muscles were working. Clubbing was present but there was no evidence of cyanosis, pallor or lymphadenopathy. His jugular venous pressure was not elevated and pedal edema was absent. Respiratory system examination revealed wide spread bilateral crepitations.
Laboratory investigations revealed a leukocyte count of 8000 /mm^3, polymorphs 68%, lymphocytes 29% and eosinophils 3%. Total eosinophil count 320 /mm^3, Hb 14gm% and ESR 20 mm in first hour. His blood sugar was 83 mg% and his serum was negative for HIV 1 and 2 antibodies. His sputum for acid-fast bacilli was negative on multiple occasions. Sputum sent for fungal and AFB culture did not grow any organisms. Sputum for pyogenic culture grew Pseudomonas which was sensitive to ciprofloxacin and amikacin. Urine complete examination was within normal limits.
His skiagram chest revealed multiple ring shadows in lungs, almost symmetrical in distribution, more apparent in mid and lower zones (Figure 1). His X-ray paranasal sinuses were essentially normal. A contrast enhanced CT scan of the chest revealed bilateral symmetric saccular bronchiectasis with air fluid levels in some of them (Figure 2).
His spirometry showed a mixed obstructive and restrictive defect. Arterial blood gas analysis showed hypoxia and hypocarbia with widened $(A - a)O_2$. His sera were negative for Aspergillus antibodies (IgG and IgM). His total IgE levels were also within normal limits. Total serum IgG, IgA and IgM levels done to rule out hypogammaglobinemia did not reveal any deficiencies.
A repeat CT screening revealed expiratory collapse of the airways with normal trachea and main bronchi. A diagnosis of saccular bronchiectasis, probably congenital in origin was made based on his clinical history traced right from infancy. The patient was managed with oxygen, antibiotics and mucolytics and he gradually improved. A bronchoscopy was planned, but the patient refused for further investigations due to financial constraints and he was subsequently lost on follow up. Our clinical diagnosis of Williams Campbell Syndrome (Bronchomalacia) was based on his clinico-radiological picture and evidence of airway collapse during expiration while CT was performed.

Figure 1. Chest radiograph PA view showing multiple ring shows scattered almost symmetrically in both mid & lower zones.

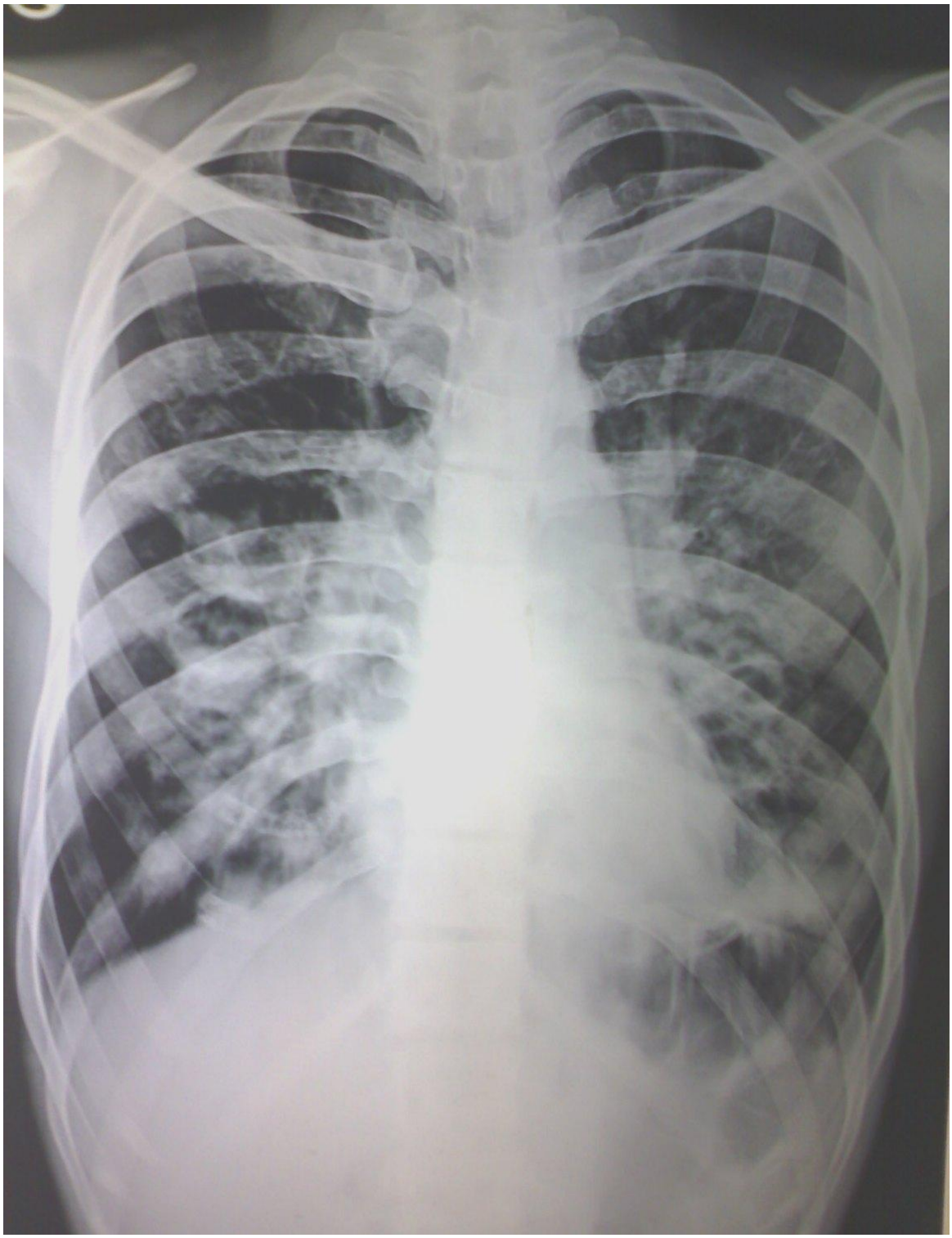
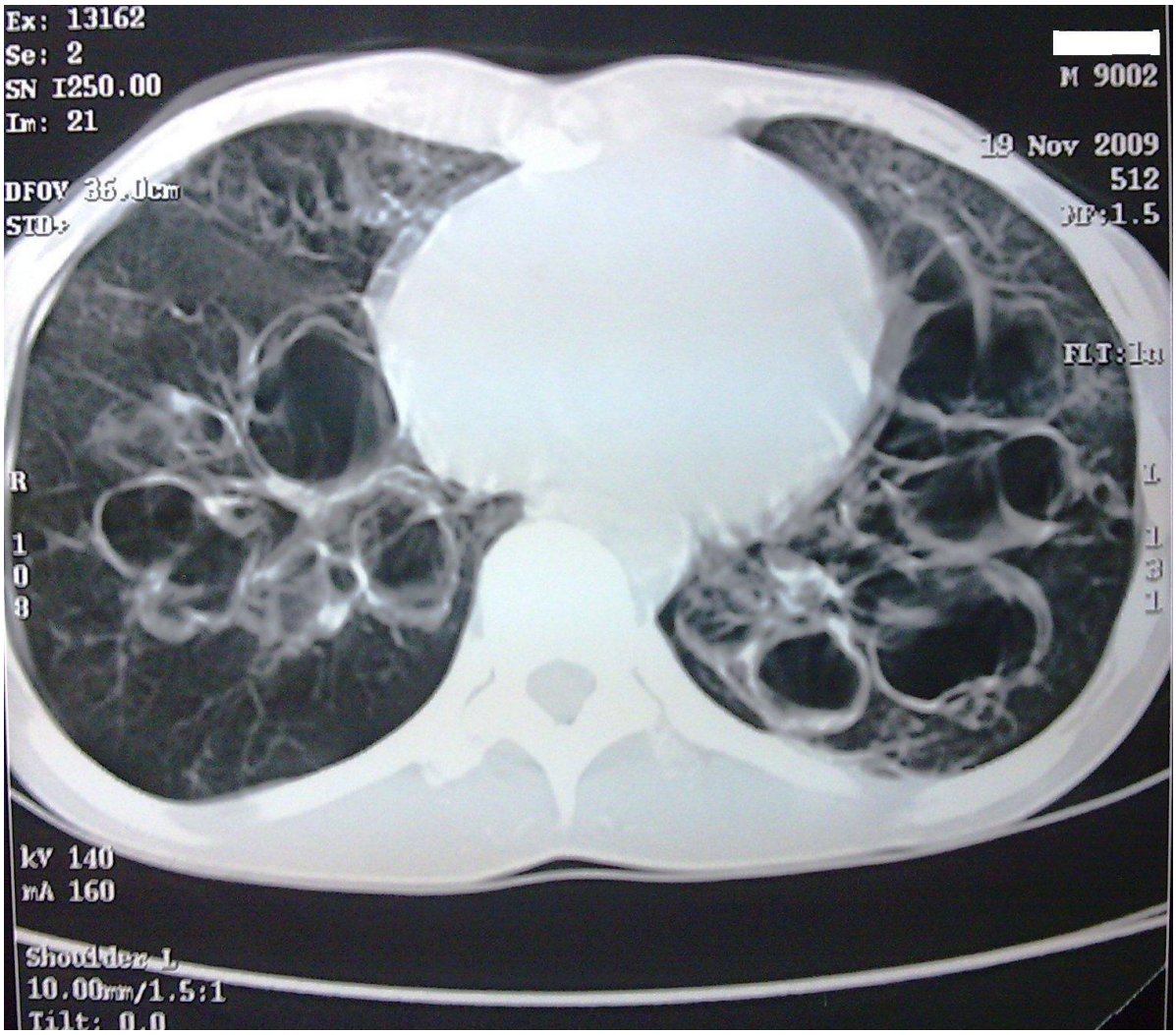


Figure 2. Computed tomogram of chest showing bilateral, symmetrically distributed bronchiectasis of proximal bronchi with sparing of trachea and main bronchi.



DISCUSSION

Williams Campbell Syndrome (Bronchomalacia) is a rare congenital syndrome characterized by defective or completely absent bronchial wall cartilage producing a mechanical abnormality that may contribute to the formation of bronchiectasis[5]. The defect may extend between first to eight generations of bronchi and computed tomography shows a remarkable ballooning expansion of proximal bronchi during inspiration with collapse during expiration originally demonstrated bronchographically[6]. Other anatomic features of the syndrome include absence of destruction of other (non cartilaginous) bronchial wall structures by inflammation and a relatively uniform, bilateral distribution of the process[7]. Familial cases of Williams Campbell Syndrome may occur in siblings, possibly a result of autosomal recessive mechanism, the disorder may also be present sporadically[7,8]. Usually presenting in early childhood, subclinical cases maybe diagnosed as late as in adulthood also[9].

Williams Campbell syndrome was first described by Williams and Campbell et al in 1960 as a rare form of congenital bronchiectasis. They described a case series of 5 children with similar clinico - radiological symptoms. It was proposed that the maldevelopment of cartilage in bronchial tree was responsible for this presentation[5]. An additional eleven cases were later described by Williams et al and his colleagues who then coined the term Williams Campbell syndrome[2]. Commonly described among children, the presenting complaints include recurrent cough, fever and breathlessness. Early development of clubbing because of recurrent suppurative infections and extensive radiological changes of saccular bronchiectasis disproportionate to the degree and duration of infection favors the diagnosis of bronchomalacia. In the initial reports bronchography was used to identify airway collapse. Bronchoscopy with functional maneuvers can also reliably detect the features of expiratory collapse and inspiratory ballooning. Recently Multi Detector - row CT (MDCT) imaging has been able to show non-invasively these features with similar sensitivity to bronchoscopy[10 – 12].

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[2, 7]. Differential diagnosis of this rare condition includes congenital and acquired conditions such as sinobronchial syndrome, cystic fibrosis, allergic bronchopulmonary aspergillosis, immune deficiency, tracheobronchomegaly etc.

Our patient presented with bilateral symmetrical saccular bronchiectasis with normal trachea and main bronchi. Expiratory collapse of the bronchiectatic airways during the performance of CT scan is not a feature of acquired. Further, the proximal extension of bronchiectasis is again most unusual in acquired form of bronchiectasis apart from Allergic Bronchopulmonary Aspergillosis (ABPA). The differential diagnosis thus narrowed to allergic bronchopulmonary aspergillosis or cystic fibrosis, ABPA was excluded by negative tests to aspergillus antibody. Absence of gastrointestinal symptoms any time during the course of illness helped us to rule out a clinical diagnosis of cystic fibrosis in the present case. Hypogammaglobulinemia was also excluded with a normal immunoglobulin A, G and M levels.

No specific treatment is currently available for this condition and the clinical course is variable. Management of acute exacerbations and maintain good bronchopulmonary hygiene is the main stay of management. The patients usually develop respiratory failure and death in early childhood is inevitable. Some children, have a milder course of disease and make it to adulthood. Treatment of such cases is bronchodilators and anti microbial therapy for infections. The disease course is characterized by recurrent respiratory tract infections and gradually respiratory failure sets in. Surgical resection of the bronchiectatic segment has been attempted, but without significant improvement[13]. Bilateral lung transplant has also been tried in an adult patient with WCS, but the patient did not survive more than one year after the surgery[14].

CONCLUSION

We hereby conclude that rare congenital anomalies like Williams Campbell Syndrome should also be considered in the differential diagnosis of bronchiectasis. The characteristic HRCT features along with clinical presentation gives a clue in diagnosing

such cases. However, a definite diagnosis can be made only after detailed workup and excluding the other common causes of bronchiectasis.

REFERENCES

1. Rosen JM. Chronic cough due to bronchiectasis: ACCP evidence – based clinical practice guidelines. Chest 2006; 129: 122S – 131 S.
2. Williams H, Landau L, Phelan P . Generalized bronchiectasis due to extensive deficiency of bronchial cartilage. Arch Dis Child 1972; 47 : 423-28.
3. Mitchell RE, Burg RG . Congenital bronchiectasis due to deficiency of bronchial cartilage (Williams-Campbell Syndrome) : a case report. J Paediatr 1975; 87 : 230-34.
4. Newman KB, Beam WR. Congenital bronchiectasis in an adult (letter). AmJ Med 1991; 91 : 198-201.
5. William H, Campbell P . Generalized bronchiectasis associated with deficiency of cartilage in the bronchial tree. Arch Dis Child 1960; 35 : 182.
6. Watanabe Y, Nishiyama Y, Kanayama H, Enomoto K Kato K, Takeichi M. Congenital bronchiectasis due to cartilage deficiency : CT demonstration. J Comput Assist Tomog 1987; 11: 701 -03.
7. Kenneth S. Wayne, Lynn M. Taussig . Probable familial congenital bronchiectasis due to cartilage deficiency (Williams Campbell Syndrome). Am Rev Resp Dis 1976; 114: 15-22.
8. Jones VF, Erd NS, Franco SM *et al* . Familial congenital bronchiectasis: Williams - Campbell Syndrome. Paediatr Pulmonol. 1993; 16: 263 - 67.
9. Przerwa KB, Bestry I, Gawryluk D, Wiatr E. Case report: Williams Campbell syndrome. Pol J Radiol, 2009; 74: 76 – 78.
10. Gilkeson RC, Ciancibello LM, Hejal RB, Montenegro JD, Lange P. Tracheobronchomalacia: dynamic airway evaluation with multidetector CT. AJR Am J Roentgenol 2001; 176: 205-10.
11. Lee KS, Sun MR, Ernst A, Feller-Kopman D, Majid A, Boiselle PM. Comparison of dynamic expiratory CT with bronchoscopy for diagnosing airway malacia: A pilot evaluation. Chest 2007; 131: 758-64.
12. Zhang J, Hasegawa I, Feller - Kopman D, *et al* . Dynamic expiratory volumetric CT imaging of the central airways : Comparison of standard dose and low dose techniques. Acad Radiol 2003; 10 : 719-24.
13. Jones VF, Eid N, Franco S, et al. Familial congenital bronchiectasis: Williams-Campbell syndrome. Pediatric Pulmonol 1993; 16:263-67
14. Palmer SM, Layish DT, Kussin PS, Oury T, Davis RD, Tapson VF . Lung Transplantation for Williams Campbell Syndrome. Chest 1998; 113 : 534-37.