Journal of Academic Research
Vol 30, Issu Special Issue in Medical Sciences, 2026 "71-78"

Severe Unilateral Cystic Bronchiectasis in a Child with Down Syndrome and Prior Congenital Diaphragmatic Hernia Repair: A Case Report

Mustafa El-Ahmer, MD[1 2], Wesam Elsaghayer, MD[1 3], Ahmed Aniba MD 1 2, Eman Alabani, MD[2], Fatma Kannah, MD[3]

[1]. Faculty of Medicine, Misrata University, Misrata, Libya
[2]. Department of Pediatric Surgery, Misrata Medical Center, Misrata, Libya
[3]. Department of Pathology, Misrata Medical Center, Misrata, Libya

elahmermustafa@yahoo.com

Article information

Key words

Bronchiectasis, cystic bronchiectasis, Down syndrome, pediatric pulmonology, congenital diaphragmatic hernia, pneumonectomy.

Received: 16-04-2026

Accepted: 19-04-2026

Published: 09-06-2026

Abstract

Background: Congenital bronchiectasis is a rare but serious condition in the pediatric population and may be more challenging to diagnose in children with Down syndrome due to overlapping clinical features and communication difficulties. Bronchiectasis, characterized by irreversible bronchial dilatation, is often associated with recurrent infections and may be particularly severe in children with underlying genetic conditions.

Case Presentation: We report a 12-year-old girl with Down syndrome and prior congenital diaphragmatic hernia repair who presented with a long-standing history of productive cough and recurrent lower respiratory tract infections since infancy. Physical examination revealed underweight status, digital clubbing, and inspiratory crackles predominantly in the left lower lung zone. Imaging demonstrated extensive cystic bronchiectasis with left lung volume reduction, and the patient underwent left pneumonectomy. Histopathology confirmed severe acute and chronic bronchiectasis with patchy fibrosis and superimposed acute bronchopneumonia.

Conclusion: This case highlights the rare occurrence of severe unilateral cystic bronchiectasis in children with Down syndrome and prior congenital diaphragmatic hernia repair. Early recognition and surgical intervention may be necessary in cases with extensive lung destruction

I) INTRODUCTION:

Bronchiectasis has been defined as a chronic, localized bronchial dilatation, usually associated with inflammation and is irreversible [1]. As defined by bronchography, bronchiectasis has been described as cylindrical, varicose or cystic, which correlates with increasing degree of severity [2]. Bronchiectasis is increasingly recognized as a major cause of respiratory morbidity around the world, with early diagnosis and appropriate management being advocated as cohort data in children have consistently shown that these reduce further lung deterioration [3].

Children with Down syndrome are prone to respiratory complications, including recurrent infections and structural lung anomalies, which may predispose them to bronchiectasis. Additionally, children with a history of congenital diaphragmatic hernia repair may experience asymmetrical lung

development. Previous surgical interventions for CDH can impact long-term lung development and contribute to chronic respiratory morbidity.

The diagnosis of bronchiectasis in children with Down syndrome presents unique challenges due to overlapping clinical features and potential communication difficulties that may delay recognition. Severe unilateral cystic bronchiectasis requiring pneumonectomy in the pediatric population is exceptionally rare and warrants detailed documentation. High-resolution CT (HRCT) with 1.5-3 mm collimation has high sensitivity and specificity compared with bronchography and is now the investigation of choice in suspected bronchiectasis [2].

II) CASE PRESENTATION

A Patient Information and Clinical History

A 12-year-old girl with Down syndrome presented to our center with a long-standing history of productive cough and recurrent lower respiratory tract infections since infancy. The patient had a significant past medical history including surgical repair of congenital diaphragmatic hernia in the neonatal period. Her respiratory symptoms had been progressive, with increasing frequency of infections requiring multiple courses of antibiotics and several hospital admissions.

B Clinical Findings

On physical examination, the patient appeared underweight for her age. Digital clubbing was noted on inspection of the fingers, a hallmark of chronic suppurative lung disease. Auscultation revealed inspiratory crackles predominantly in the left lower lung zone. The presence of coarse crepitations predominantly in the left lower lung zone provided clinical localization. No wheeze was appreciated on examination. The patient demonstrated reduced exercise tolerance and chronic fatigue related to her respiratory condition..

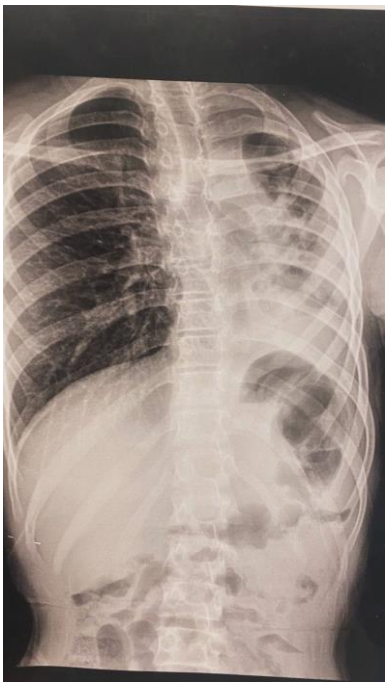
C Diagnostic Assessment

Laboratory investigations at the time of admission, including complete blood count, renal function, and electrolytes, were unremarkable. Chest radiograph demonstrated multiple cystic lesions confined to the left lung. The chest radiograph showed multiple cystic lesions in the left lung.

Contrast-enhanced computed tomography scan of the chest revealed total left lung volume reduction with clusters of cystic spaces of variable size covering the entirety of the left lung . These cystic spaces demonstrated air-fluid levels, and the radiological findings were consistent with severe cystic bronchiectasis. The CECT scan revealed total left lung volume reduction with clusters of cystic spaces of variable size covering the whole left lung, showing air-fluid levels . The main features of bronchiectasis in CT scans include increased broncho-arterial ratio (the signet-ring appearance), bronchial wall thickening, lack of bronchial tapering, presence of bronchial structures in the lung periphery, mucus plugging, and mosaic perfusion reflecting air-trapping [4].

D Therapeutic Intervention

Given the extensive destruction of the left lung with no viable salvageable tissue and the chronic symptomatic nature of the disease, the patient underwent left thoracotomy and pneumonectomy.
Operative Findings: Upon exploration, the left lung was found collapsed with extensive adhesions. Pulmonary architecture was obliterated, with no discernible separation between the upper and lower lobes. The lung parenchyma contained multiple cavities and cystic areas filled with purulent material. Perfusion was markedly poor throughout the lung tissue, indicating non-viable lung.



Surgical Management: Given the absence of healthy lung tissue, a left pneumonectomy was performed.

The surgical procedure was performed without immediate complications. Postoperatively, she was managed in the intensive care unit with intravenous antibiotics and chest physiotherapy. The postoperative recovery was closely monitored with attention to respiratory function, fluid balance, and pain management.

E Pathological Findings

Gross examination of the surgical specimen revealed a markedly distorted left lung architecture with multiple cystic spaces filled with mucopurulent material . Microscopic examination confirmed the diagnosis of severe acute and chronic bronchiectasis with patchy fibrosis . Histological features included extensive bronchial wall destruction, chronic inflammatory cell infiltration, squamous metaplasia of the bronchial epithelium, and superimposed acute bronchopneumonia . The findings were consistent with bronchiectasis, with superimposed acute bronchopneumonia and negative for malignancy.

F Follow-up and Outcomes

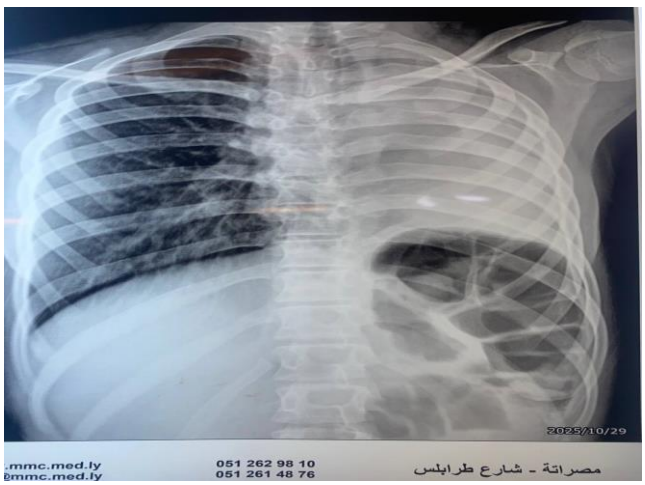
The patient recovered uneventfully from the surgical procedure and was discharged with instructions for follow-up and continued pulmonary care . At subsequent follow-up visits, she demonstrated improved exercise tolerance and resolution of chronic cough . The patient's quality of life improved significantly following surgical intervention, with reduction in the frequency of respiratory infections and improved nutritional status.

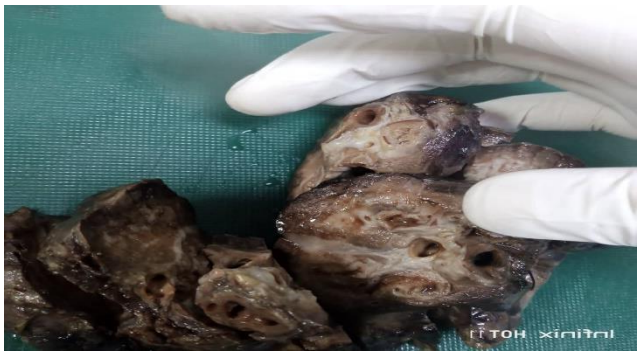
III) DISCUSSION

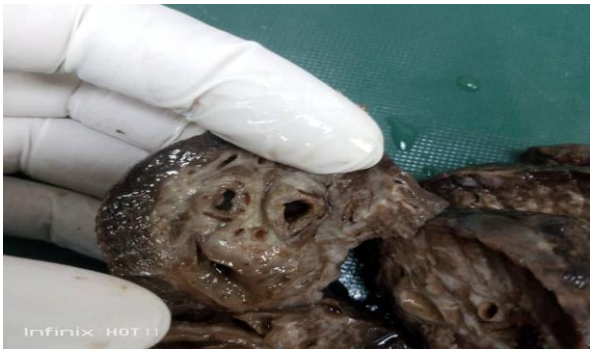
A Epidemiology and Clinical Context

HRCT defined non-CF bronchiectasis is not an uncommon problem in tertiary referral populations, with studies reporting that bronchiectasis cases can constitute 9.6% of all new referrals to tertiary respiratory pediatric referral centers [5]. In indigenous populations and certain high-risk groups, the prevalence is even higher, ranging from 63 to 1600 per 100,000 children in countries such as Australia, New Zealand, and the USA [6].

B Pathophysiology in Down Syndrome







Children with Down syndrome are particularly vulnerable to developing bronchiectasis through multiple mechanisms. The vicious cycle of infection-inflammation begins with predisposing factors including genetics, epigenetics, low birthweight or prematurity, and in-utero tobacco smoke exposure [7]. Early persistent infection leads to inflammation with or without atelectasis, resulting in epithelial and endobronchial injury and mucociliary disruption [7]. This progresses through protracted bacterial bronchitis and chronic suppurative lung disease to radiographic bronchiectasis, characterized by airway remodeling and ongoing disease progression with chronic infection, persistent biofilms, inflammation, and immune dysregulation [7].

C Clinical Presentation and Diagnostic Features

Unilateral cystic bronchiectasis is rare and often presents with recurrent infections, chronic cough, and digital clubbing. The chronicity of symptoms since infancy in our patient, combined with progressive lung destruction, necessitated aggressive intervention. Imaging, particularly contrast-enhanced computed tomography, is critical for diagnosis and surgical planning [2].

A radiological diagnosis of bronchiectasis may be more common than previously thought and the changes that suggest this diagnosis are often quite subtle, especially with cylindrical bronchiectasis [2]. The term bronchiectasis is often loosely applied in reporting HRCT of the lungs where bronchial dilatation is present, which may lead to diagnostic confusion [2]. With increasing availability of ultrafast HRCT, it is likely that the number of CT scans done in children to investigate diseases of the chest will continue to increase [2].

D Reversibility Considerations

In the pediatric population, bronchial dilatation can resolve with medical treatment. Studies have shown that overall bronchial dilatation resolved completely in six children with medical treatment after a median period of 24 months [2]. Of 34 lobes affected, there was resolution of bronchial dilatation in 16 lobes (47%), with a further four lobes showing improvement [2]. Of 16 lobes where the diagnosis was bronchiectasis in collapsed, consolidated lung, there was complete resolution in nine (56%) [2]. However, in our case, the severity and extent of disease precluded conservative management.

Complete radiological resolution of bronchiectasis on HRCT scan has been reported in cases of post-pneumonic and idiopathic bronchiectasis following appropriate treatment [5]. Early diagnosis and treatment may improve the long-term prognosis as it is known that aggressive treatment with antibiotics and physiotherapy slows disease progression of bronchiectasis [5].

E Microbiology and Infection

The respiratory pathogens most associated with pediatric bronchiectasis provide important context for management. Non-typeable Haemophilus influenzae and Streptococcus pneumoniae are frequently identified pathogens in children with chronic suppurative lung disease [8]. H. influenzae is the most common bacterial species infecting the lower airways of children with endobronchial suppuration, including protracted bacterial bronchitis, recurrent or non-responsive community-acquired pneumonia, and bronchiectasis [9]. Although H. influenzae infection may be an important risk factor for bronchiectasis, there are likely to be other contributory factors [9].

F Risk Factors and Disease Progression

Pulmonary exacerbations, especially those needing admission to hospital, are the only known independent risk factor for long-term pulmonary decline in children with bronchiectasis [6]. Understanding factors associated with recurrent and severe acute respiratory exacerbations over time may help determine which children might benefit most from close monitoring and aggressive treatment strategies [10]. Children with severe pulmonary infections and those with recurrent lower respiratory tract infections early in life are more likely to have severe and recurrent exacerbations for at least 2 years [10].

Findings from longitudinal studies support the hypothesis that protracted bacterial bronchitis and bronchiectasis represent a clinical spectrum [9]. Children with protracted bacterial bronchitis have endobronchial bacterial infection and neutrophilic airway inflammation, factors known to be injurious to the airways [9]. In a subset of children receiving close follow-up by pediatric pulmonologists, recurrent episodes (>3 per year) of protracted bacterial bronchitis precede a diagnosis of bronchiectasis [9].

G Diagnostic Criteria in Children

The radiologic definition of airway dilatation and bronchiectasis in children has substantial implications for clinical practice [3]. Currently, the gold standard for diagnosing bronchiectasis is through data obtained from high-resolution CT scan, in which one of the key markers is increased bronchoarterial ratio [3]. In the context of a chronic wet cough, bronchiectasis was diagnosed when the broncho-arterial ratio was larger than 0.8 with or without peribronchial thickening or lack of bronchial tapering [11].

We recommend that childhood bronchiectasis is defined as a clinical syndrome (persistent or recurrent episodes of chronic wet or productive cough, sometimes with coarse crackles and digital clubbing), confirmed radiographically using pediatric BAR data (abnormal when >0.80) [4]. Use of pediatric specific rather than the adult derived definitions (ie, irreversibility and a single higher BAR value) is important in the context of stimulating efforts to prevent disease progression or even reverse the changes with early and intensive treatment in children [4].

H Management Approaches

Treatment of bronchiectasis aims to resolve acute infection and control infection and inflammation, improving symptoms (persistent or recurrent wet cough and breathlessness), reducing frequency of acute pulmonary exacerbations, preserving lung function, and improving quality of life [6]. The management of treatable traits includes addressing modifiable factors such as vaccinations, malnutrition, hygiene practices, health education, socioeconomic factors, environmental tobacco smoke or pollutants, and housing conditions [7].

For medical management, long-term azithromycin therapy has shown efficacy in reducing exacerbations in indigenous children with non-cystic fibrosis bronchiectasis or chronic suppurative lung disease [6]. Pulmonary exacerbations can be reduced through aggressive antibiotic therapy and chest physiotherapy, particularly in children with recurrent episodes [6].

Generic modifiable factors include adherence to treatment regimens (with reminders such as SMS and other technologies), nutrition and malnutrition management (including vitamin D supplementation and growth monitoring), prevention of tobacco and vaping exposure, reduction of indoor and outdoor pollution, allergen and fungal exposure reduction, exercise promotion, and infection prevention strategies such as breastfeeding [7]. In adults with bronchiectasis, low adherence is associated with poorer outcomes, though no randomized controlled trials have been conducted [7].

I Surgical Considerations

In cases of severe localized disease with extensive lung destruction, surgical intervention may be necessary. Clarification of the diagnosis of bronchiectasis has major practical implications for the management of patients, allowing rational treatment with antibiotics and physiotherapy to be instituted [5]. Studies in adults with bronchiectasis have shown that onset of sputum production commenced before the age of 10 years in 40% of cases, suggesting that the onset of disease is often in childhood [5]. Aim of surgery resection of affected part of lung usually.

J Inflammatory Mechanisms

Understanding the inflammatory pathways in bronchiectasis provides insight into disease progression. Patients experience declining pulmonary function related to chronic airway inflammation, which results from epithelial and immune cell secretion of proinflammatory mediators that promote neutrophil influx into the airways [12]. This inflammatory response may be disproportionate to the inciting infectious stimulus, resulting in an overly exuberant influx of neutrophils [12]. The neutrophils release proteases, including neutrophil elastase, that eventually overwhelm the antiprotease capacity of the lung and cleave structural proteins, leading to bronchiectasis [12].

The airway epithelium is an important source of interleukin-8, a CXC chemokine which is the principal neutrophil chemoattractant [12]. Airway epithelial cells also modulate the production of IL-1, tumor necrosis factor-α, and other mediators by recruiting immune cells, including macrophages and neutrophils [12]. High pulmonary levels of IL-6 and IL-1β in children with chronic suppurative lung disease are associated with impaired systemic immune responses [8].

K Clinical Implications

This case emphasizes the importance of early diagnosis, comprehensive imaging evaluation, and timely surgical management in extensive bronchiectasis affecting pediatric patients with Down syndrome. Severe unilateral cystic bronchiectasis can occur in children with Down syndrome, particularly those with additional risk factors such as a history of congenital diaphragmatic hernia repair. The clinical relevance and importance of awareness in similar cases are paramount for improving outcomes.

Clinicians caring for children with Down syndrome should maintain a high index of suspicion for chronic pulmonary complications and pursue aggressive diagnostic evaluation when indicated. Multidisciplinary management, including surgical intervention when appropriate, may be required for optimal outcomes. This case contributes to the limited literature on severe unilateral bronchiectasis in children with Down syndrome. Early recognition and intervention are essential to prevent progression and complications.

The CT scans of children should be reviewed in the context of the clinical course, to avoid reporting errors, which may label the child for life [2]. The medical and surgical treatment of bronchiectasis requires careful consideration of the individual patient's clinical presentation and disease severity [2].

IV) Conclusions

This case report documents a rare presentation of severe unilateral cystic bronchiectasis in a child with Down syndrome and prior congenital diaphragmatic hernia repair. The patient's clinical course demonstrates the importance of early recognition of chronic respiratory symptoms in high-risk populations. Despite the potential for reversibility of bronchiectatic changes with aggressive medical management in some cases, extensive unilateral disease may necessitate surgical resection to improve quality of life and prevent recurrent life-threatening and hospitalization infections.

The expanding literature, including recent references from 2018-2025, continues to expand our understanding of pediatric bronchiectasis management. Collaborative international case registries could help identify optimal management approaches for these rare presentations. Clinicians should be aware that children with Down syndrome and additional risk factors such as congenital diaphragmatic hernia repair require vigilant monitoring for the development of bronchiectasis, with prompt initiation of appropriate therapy when clinical suspicion arises.

V) PATIENT CONSENT AND ETHICAL APPROVAL

Informed written consent was obtained from the patient's parents for publication of this case report and any accompanying images . Consent was obtained from the patient's guardian . All ethical guidelines for case report publication have been followed, and patient confidentiality has been maintained throughout .

VI) CONFLICT OF INTEREST

The authors declare no conflicts of interest related to this case report .

VII) FUNDING

This case report received no specific grant from any funding agency .

VIII) ACKNOWLEDGMENTS:

The authors thank the patient and family for their cooperation and consent for this publication.

IX) References:

(1) E. A. Gaillard, H. Carty, D. Heaf, and R. L. Smyth, "Reversible bronchial dilatation in children: Comparison of serial high-resolution computer tomography scans of the lungs," European Journal of Radiology, 2003.
(2) N. Kapur, J. P. Masel, D. Watson, I. B. Masters, and A. B. Chang, "Bronchoarterial ratio on high-resolution CT scan of the chest in children without pulmonary pathology," Chest, 2011.
(3) A. B. Chang, A. Bush, and K. Grimwood, "Bronchiectasis in children: Diagnosis and treatment," The Lancet, 2018.
(4) K. M. Eastham, "The need to redefine non-cystic fibrosis bronchiectasis in childhood," Thorax, 2004.
(5) P. C. Valery et al., "Long-term azithromycin for indigenous children with non-cystic-fibrosis bronchiectasis or chronic suppurative lung disease (bronchiectasis intervention study): A multicentre, double-blind, randomised controlled trial," The Lancet Respiratory Medicine, 2013.
(6) A. B. Chang, A. Bush, and K. Grimwood, "Bronchiectasis in children: Diagnosis and treatment," The Lancet, 2018.
(7) S. J. Pizzutto, J. W. Upham, S. T. Yerkovich, and A. B. Chang, "High pulmonary levels of IL-6 and IL-1 in children with chronic suppurative lung disease are associated with low systemic IFN- production in response to non-typeable haemophilus influenzae," PLOS ONE, 2015.
(8) D. F. Wurzel et al., "Protracted bacterial bronchitis in children," Chest, 2016.
(9) G. J. Redding et al., "Respiratory exacerbations in indigenous children from two countries with non-cystic fibrosis chronic suppurative lung disease/bronchiectasis," Chest, 2014.
(10) V. Goyal, K. Grimwood, J. Marchant, I. B. Masters, and A. B. Chang, "Does failed chronic wet cough response to antibiotics predict bronchiectasis?" Archives of Disease in Childhood, 2014.
(11) A. Elizur, C. L. Cannon, and T. W. Ferkol, "Airway inflammation in cystic fibrosis," Chest, 2008.