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# Primary Ciliary Dyskinesia in a 10-Year-Old Girl: A Case Report of Recurrent Respiratory Infections and Bronchiectasis

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## Abstract

**Background:** *Primary Ciliary Dyskinesia (PCD) is a rare inherited disorder characterized by impaired ciliary function, leading to recurrent respiratory infections, bronchiectasis, and other complications. This case report details the clinical presentation, diagnostic workup, and management of a 10-year-old girl diagnosed with PCD.*

**Case Presentation:** *We report the case of a 10-year-old female from Nellur Channagiri, who presented with a history of recurrent cough, fever, and poor weight gain over the past two years. She was born at term with a history of respiratory distress requiring NICU admission. Over the years, she had multiple admissions for respiratory infections and was treated with intravenous antibiotics. On physical examination, she exhibited clubbing and coarse crepitations in both lungs. Her chest X-ray and CT thorax revealed features of bronchiectasis. A bronchoscopy confirmed adenoid hypertrophy, while a clinical exome report identified Primary Ciliary Dyskinesia. The patient is undergoing airway clearance therapy and regular follow-up to manage the disease.*

**Conclusion:** *This case highlights the importance of considering PCD in patients with recurrent respiratory infections and bronchiectasis. Early diagnosis and appropriate management can help mitigate disease progression and improve quality of life.*

## 1. Introduction

Primary Ciliary Dyskinesia (PCD) is a genetic disorder that affects the structure and function of motile cilia, leading to chronic respiratory tract infections, middle ear effusion, and bronchiectasis. The condition is often underdiagnosed due to its variable clinical presentation and overlap with other respiratory conditions. This case report describes the diagnostic journey of a 10-year-old girl presenting with recurrent respiratory infections, ultimately diagnosed with PCD.

## 2. Case Presentation

A 10-year-old female, Miss B., from Nellur Channagiri, presented to our hospital with complaints of a cough lasting two weeks and a fever of eight days. Her mother provided a reliable history, noting that Miss B. Had experienced recurrent episodes of cough and fever over the past two years, with no significant weight gain during this period. She had been treated with intravenous antibiotics on several occasions. The patient was born at term with a birth weight of 2.5 kg and had a postnatal history of respiratory distress requiring NICU admission for four days. She underwent cataract surgery with intraocular lens placement at the age of four. Her developmental milestones were normal, but she had been frequently absent from school due to illness, which affected her academic performance.

## 3. Clinical Findings:

On examination, the patient was conscious and cooperative, with no signs of pallor, cyanosis, or significant lymphadenopathy. However, she exhibited Grade 2 clubbing, and her anthropometric measurements indicated that both her weight (18 kg) and height (118 cm) were below the 3rd percentile. Auscultation revealed coarse crepitations bilaterally.

## 4. Investigations:

A complete blood count showed elevated white blood cells (TLC: 15,700), and her CRP levels were mildly elevated. Her serum electrolytes were within normal limits, but her chest X-ray and CT thorax showed signs of bronchiectasis. A bronchoscopy performed at an outside center

revealed Grade 3 adenoid hypertrophy with mucopurulent secretions.

Given the recurrent infections and presence of bronchiectasis, further diagnostic workup included a sweat chloride test, which yielded indeterminate results. Finally, a clinical exome sequencing confirmed the diagnosis of Primary Ciliary Dyskinesia.

## 5. Discussion

Primary Ciliary Dyskinesia results in defective motile cilia, which are responsible for clearing mucus from the respiratory tract. This leads to chronic sinopulmonary infections, as seen in this patient, who had multiple episodes of pneumonia and bronchiectasis. The diagnosis of PCD is often delayed due to its non-specific symptoms and overlap with conditions like cystic fibrosis. Management focuses on airway clearance techniques, including postural drainage and chest physiotherapy, along with aggressive treatment of infections. This patient was started on airway clearance therapy, including the use of positive expiratory pressure devices and regular chest physiotherapy sessions to enhance mucociliary clearance. Additionally, she undergoes regular spirometry and chest imaging to monitor pulmonary function and prevent further deterioration.

## 6. Conclusion

This case underscores the importance of considering PCD in patients with recurrent respiratory infections and bronchiectasis, particularly when there is a history of neonatal respiratory distress or situs inversus. Early recognition and management are crucial in preventing complications and improving long-term outcomes.

## 7. Follow-Up

The patient continues to receive regular follow-up, including spirometry to assess pulmonary function and imaging to monitor disease progression. Her management includes aggressive treatment of infections and ongoing physiotherapy to promote effective airway clearance.

## References

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