DOI: http://doi.org/10.4038/jccp.v53i2.7964 **Case report**

A young woman with cystic bronchiectasis: mystery revealed

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Abstract

Bronchiectasis is a disabling respiratory disease which results in permanent distortion and dilatation of the affected bronchi. It is a chronic obstructive lung disease with clinical manifestations of chronic cough, sputum production, and recurrent chest infections. It has several aetiologies which include infections, primary immunodeficiency, recurrent aspiration, inhaled foreign body, cystic fibrosis and primary ciliary dyskinesia. Yellow nail syndrome (YNS) is a very rare disease with a clinical triad of nail changes, respiratory manifestations and lymphoedema. Diagnosis can be made in the presence of two of these symptoms. We present a case of a young woman with cystic bronchiectasis and maxillary sinusitis, later diagnosed with yellow nail syndrome. A 22-year-old woman diagnosed with cystic bronchiectasis for 3 years presented with an infective exacerbation. She was noted to have characteristic nail changes involving hands and feet which raised suspicion of YNS. It should be considered in a patient with bronchiectasis and characteristic nail changes, even in the absence of lymphoedema. Therefore, a high degree of clinical suspicion and exclusion of other underlying causes are necessary to arrive at a timely diagnosis.

Key words: bronchiectasis, yellow nail syndrome, sinusitis, nail changes

Introduction

Bronchiectasis is a heterogenous disease condition characterized by irreversible airway dilatation with chronic bronchial inflammation.[1] It is a leading cause of respiratory morbidity. There are 3 morphological types of bronchiectasis; cylindrical (or tubular), varicoid and cystic (or saccular). Out of these, cystic bronchiectasis is less commonly encountered in clinical practice.[2] Bronchiectasis has several etiologies. Most patients have idiopathic bronchiectasis, but investigating for an underlying etiology which is amenable to medical therapy, is of utmost importance.[1]

Yellow nail syndrome (YNS) is an extremely rare disorder of unknown etiology characterized by the clinical triad of yellow and thickened nail changes, respiratory involvement and lymphedema.[3] Respiratory manifestations include recurrent pleural effusions, chronic bronchitis, chronic sinusitis and bronchiectasis. Bronchiectasis is present in nearly 44% of cases of YNS.[3]

We report a case of a young women, diagnosed with idiopathic cystic bronchiectasis 3 years ago with characteristic nail changes which was overlooked, ultimately fitting into the clinical diagnosis of YNS.

Case history

A 22-year-old woman presented to us with fever, worsening cough, shortness of breath and copious amount of greenish sputum for one-week duration. She had been diagnosed with cystic bronchiectasis 3 years ago, at the age of 19, when she had presented with cough and mucopurulent sputum production. Since then, the clinical course has been complicated by frequent infective exacerbations, some of which were due to *pseudomonas aeruginosa*. She is a child of non-consanguineous parents and there is no family history of a similar disease. There was no past history of infections such as pertussis, measles or tuberculosis and neither does she have a history of recurrent chest infections, diarrheal illnesses to suggest immune

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deficiency state. No history of occupational exposure, active or passive smoking. She doesn't have features of any connective tissue disorder.

On examination, she was thin built with a body mass index (BMI) of 18 kg/m^2. Her nails were thickened with yellowish brown discoloration and onycholysis. There was grade III clubbing with excessive curvature of the nails. All the toenails and fingernails were affected (Figure 1). Peripheral lymphedema was not detected. On auscultation, coarse crepitations were heard over bilateral lung fields more in the lung bases. Cardiovascular system was normal with no evidence of pulmonary hypertension. Abdominal and neurological examinations were unremarkable.

Chest X ray showed bilateral extensive ring shadows with some showing fluid levels (Figure 2).

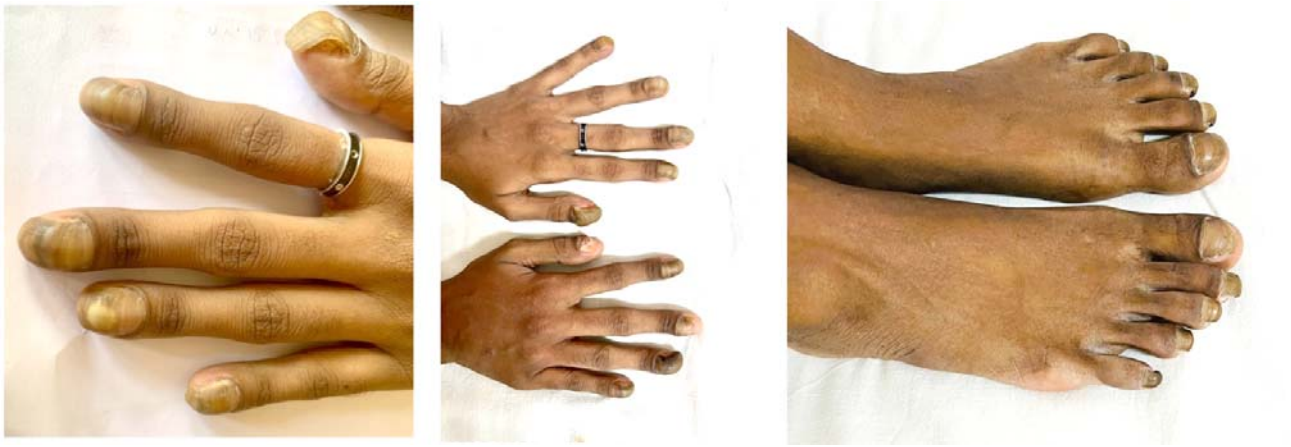

Figure 1. Hands and feet showing nail changes.

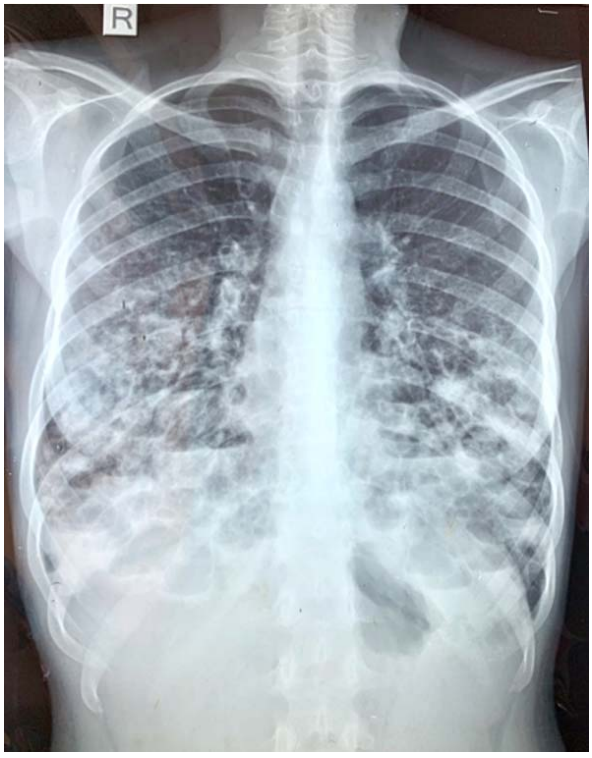

Figure 2. Chest Xray PA showing bilateral extensive ring shadows with some showing fluid levels.

High-resolution CT (HRCT) of the chest revealed marked cystic bronchiectasis with fluid levels involving bilateral lungs predominantly in the basal segments, right middle lobe and anterior segments of upper lobe (Figure 3). Xray sinus view and contrast-enhanced computed tomography (CECT) of paranasal sinuses showed bilateral maxillary sinusitis. Transthoracic echocardiogram was normal with no evidence of pulmonary hypertension.

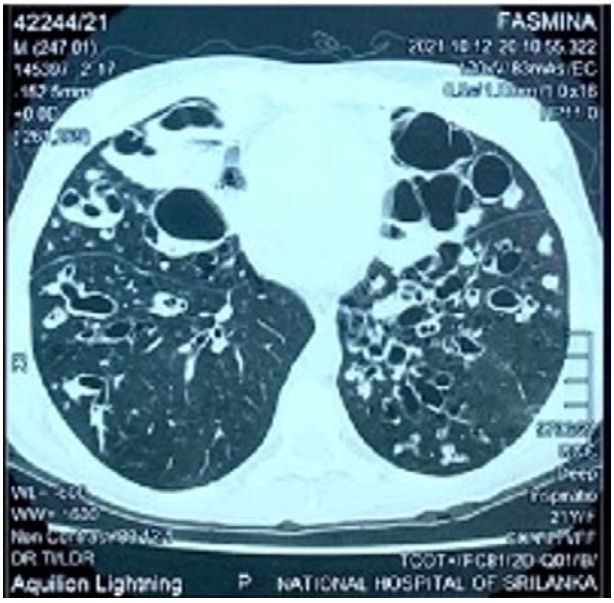

Figure 3. HRCT chest showing marked cystic bronchiectasis with fluid levels affecting bilateral lungs with predominance in the bilateral basal segments, right middle lobe and anterior segments of upper lobe.

She had been extensively investigated previously for a primary cause for the young onset cystic bronchiectasis. She was screened for cystic fibrosis where her sweat chloride test was normal and cystic fibrosis transmembrane conductance regulator (CFTR) gene mutations were negative. The bronchoscopy reveled dilated bronchial tree with increased thick secretions with no evidence of intraluminal obstructive pathologies. Bronchoalveolar lavage (BAL) for mycobacterial tuberculosis, *Aspergillus galac-tomannan* antigen and fungal cultures were reported as negative. Skin prick tests for *Aspergillus fumigatous* and retroviral screening was negative. Her serum immunoglobulin levels and complement levels were within the normal range making humoral immuno-deficiency a less plausible etiology. Serum Alpha 1 antitrypsin level was within normal limits.

The nail changes had appeared when she was 16-years-old, involving all the nails of hands and feet simultaneously. The nails were slow growing and curved which were treated as fungal infections with no response. Her first respiratory symptoms of bronchiectasis were noted three years after the onset of nail changes. Specialized opinion was sought from the consultant dermatologist and clinical diagnosis of YNS was made.

Discussion

YNS is a rare disease with an estimated prevalence of <1/1,000,000 worldwide. The first case was described in 1964 and less than 400 cases have been reported in the literature so far. YNS occurs mainly in adults after 50 years of age, but juvenile and familial forms have been described. The etiology is unknown, but some association with underlying impairment of lymphatic drainage has been postulated. YNS is frequently an isolated disorder, but rarely may have an association with autoimmune diseases or malignancies.

Diagnosis of YNS is mainly clinical and investigations are carried out to exclude other causes. The classic triad of YNS is thickened yellow nails, primary lymphoedema and respiratory manifestations. Presence of nail changes along with one other along with one other clinical feature is adequate to diagnose the disease, as the complete triad is present in only up to 27%-60% of patients. Though the yellow nails are the main clinical sign leading to the diagnosis, the time line of the development of yellow nails and other systemic manifestations vary, thereby delaying the diagnosis of YNS.

In the majority, respiratory manifestations precede the onset of nail changes. Chronic cough, rhinosinusitis, pleural effusions mainly due to chylothorax and chronic bronchiectasis predominate in descending order of prevalence.[4] Our patient had cystic bronchiectasis with no evidence of peripheral lymphoedema, which occurs in 22% of patients with YNS.[4]

Diagnosis of YNS was delayed by many years in our patient, despite having the classic nail changes at the time of onset of respiratory symptoms. The nail changes in YNS are quite distinctive which include, yellowish discoloration, marked thickening, slow growth, excessive curvature and onycholysis.[3] Even though, the name implies as “yellow nail”, the nail

discoloration varies from pale yellow to dark greenish color. In South Asian setting, the nail color may further vary to a darker brownish color which can easily misdiagnosed as fungal infection as in our patient.[5]

Treatment options for YNS are limited with no promising benefit. Oral vitamin E (1000-1200 IU / day) and zinc supplements have shown partial response in some patients.[3,6] It is hypothesized that antioxidant properties of vitamin E helps to prevent free radical mediated oxidative damage and production of the lipofuscin pigment in nails. Although YNS is not caused by fungal infections, some use triazole antifungals with minimal benefits.[3] We treated our patient with oral vitamin E and zinc supplements. Infective exacerbation of bronchiectasis was managed with pseudomonas eradication therapy, chest physiotherapy with airway clearance techniques. She was started on antibiotic prophylaxis to prevent further infective exacerbations. At three months follow up, she was free of new respiratory infections with minimal chest symptoms while maintaining a good state of health.

Conclusion

YNS is a rare clinical entity which doesn't always present as the classic clinical triad. It should be considered in a patient with chronic respiratory manifestations such as bronchiectasis or chronic sinusitis with characteristic nail changes even in the absence of lymphoedema. High degree of clinical suspicion and exclusion of other possible causes are mandatory to arrive at a definitive diagnosis.

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Authors' contributions

AG collected information, followed up the patient, did literature review and drafted the manuscript. MDUE, JA and RK were involved in the diagnosis and management of the patient and preparing the manuscript. All the authors read and approved the final manuscript.

Consent for publication

Informed written consent for publishing the patient's details including the photos, in the journal was obtained from the patient by the corresponding author.

Competing interests

The authors declare that they have no competing interests.

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