Kartagener's Syndrome in a Non-Compliant Patient

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INTRODUCTION

In 1933 Kartagener's syndrome (KS), a rare autosomal recessive disorder, was clinically recognized by Manes Kartagener, M.D [1]. KS makes up about half of all primary ciliary dyskinesias, with a prevalence of 1 in every 32,000 people [2]. The classic triad of signs and symptoms includes bronchiectasis, sinusitis and situs inversus. A mutation in the *DNAI1* and *DNAH5* genes causes ciliary dysfunction throughout the body, resulting in a variety of physiological impairments characteristic of KS including impaired respiratory ciliary clearing of pathogens (leading to recurrent sinupolmonary infections and sinusitis), impaired sperm motility (leading to infertility) and impaired embryogenesis (leading to dextrocardia and situs inversus) [3,4]. Here, we present a case of pulmonary complications in a non-compliant patient with Kartagener Syndrome. This case is important both because of its rarity and the clinical sequelae of non-compliance on pulmonary complications.

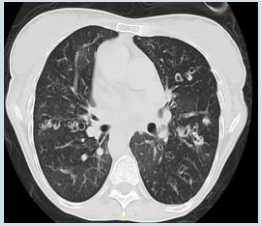
CASE

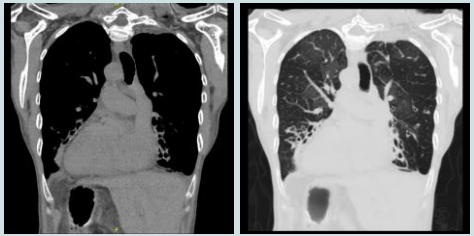
Patient Information: A 52-year-old female with a past medical history of Kartagener's syndrome with situs inversus, extensive bronchiectasis on home oxygen supplementation, pulmonary hypertension and Heart Failure with Reduced Ejection Fraction presented to the ED on 11/13/2018, with a chief complaint of shortness of breath. She reports her shortness of breath has significantly worsened over the last 3 days. It is associated with bilateral lower extremity edema and fatigue. She admits to chronic cough with whitish/yellowish sputum production which is unchanged. Home medications include inhaled tobramycin, acapella valve and has received courses of azithromycin in the past. The patient admits to limited functional capacity, only being able to walk short distances without experiencing shortness of breath. She admits to multiple hospitalizations, with recent bronchoscopy at Mount Sinai Hospital revealing colonization of *Pseudomonas*. The patient admits to non-compliance with home oxygen for the last 3 months. All other systems reviewed and negative.

Clinical Findings: The patient presented with an oxygen saturation of 60% on room air and respiratory rate of 22 with otherwise stable vital signs. Physical Examination was significant for mild respiratory distress, utilizing accessory muscles of respiration but is able to speak in full sentences without difficulty, cachectic appearance, heart sounds best appreciated in the right chest, rales in left upper and lower lobes and rales in the right lower lobe, 1+ bilateral lower extremity edema. Physical exam otherwise unremarkable.

TIMELINE

3/2018-2/2016	PCP / Pulmonary Clinic: Initial imaging showing Kartagener's, Bronchiectasis, obstructive PFTs, Sinusitis.
5/2/2018	First Hospitalization: Queens Hospital, Respiratory Failure, Discharged on prednisone, noncompliant.
5/16/16	Pulmonary Clinic: 51% Sat on RA, sent for Second Hospitalization at Mount Sinai, intubated, Sputum Cx growing *Psuedomonas.*
6/2018-8/2018	Pulmonary Clinic: Azithromycin / Tobramycin for colonization, Mucolytic / O2 supplementation poor compliance.
11/13/18 – 11/16/18	Third Hospitalization: At Jamaica Hospital Medical Center for Respiratory Failure, noncompliant with BiPAP therapy.





CT chest without contrast 11/14/2018

DIAGNOSTICS / THERAPEUTICS

Diagnostic Assessment: Initial testing was significant for chronic polycythemia, hyponatremia, hyperkalemia, transaminitis, elevated BNP and chronic respiratory acidosis with metabolic compensation and hypoxemia. Chest X Ray revealed mild bilateral pleural effusion with bronchiectatic changes, large lung volumes and patchy appearance throughout. Sputum cultures were significant for moderate GNR on stain and eventually grew *Pasteurella* species. CT Chest without contrast revealed extensive bilateral bronchiectasis and associated changes, with dextrocardia / situs inversus and left pleural effusion.

Therapeutic Interventions: The patient was initially started on venturi mask 50% flow, furosemide 40mg IV BID, Levofloxacin 500mg IV Q24hr x 48hr followed by 500mg PO Q24hr x 5 days, Ceftriaxone 1g IV Q12hr, Solumedrol 40mg IV Q8hr and Ipratropium Bromide/Albuterol Sulfate 3mL nebulization Q6hr PRN. The patient underwent diuresis with daily electrolyte monitoring and replacement and titration of her oxygen requirement with daily blood gases. Oxygen saturation and blood gases indicated BiPAP therapy, however the patient was non-compliant throughout the hospital course. Sputum Gram Stain and Culture allowed tailoring of antibiotics to Levofloxacin. Aggressive respiratory therapy for bronchiectatic mucous plugging continued throughout hospital course.

Follow-up and Outcomes: The patient improved clinically with diuresis and antibiotics, however refusal of BiPAP therapy resulted in suboptimal correction of respiratory acidosis. The patient was discharged on furosemide 40mg daily PO, azithromycin 500 mg PO 3 times a week, levofloxacin 500mg daily for 7 days and was to restart inhaled tobramycin. Pulmonary team assessment that patient will require future lung transplant. The patient was non-compliant with scheduled PCP and Pulmonary Clinic appointments thereafter.

DISCUSSION

Our case demonstrates an exacerbation of pulmonary complications in a patient with Kartagener's syndrome who is non-compliant with home therapy. Common complications of Kartagener's syndrome include chronic and recurrent sinusitis, pneumonia, bronchiectasis and otitis media [5]. Late diagnosis and noncompliance with medical treatment have been associated with worsening progression of lung disease [5].

While there is no cure for KS, therapeutic goals are to minimize complications through mucous clearance, chest physiotherapy and antibiotic courses [6]. Routine vaccination compliance and lung irritant avoidance / lung hygiene are the cornerstones of preventative management [7]. Severe bronchiectasis can be managed with lobectomy or lung transplantation.

While KS patients with significant treatment burden experience worse quality of life and worse treatment compliance, patient education has been shown to improve prognosis and treatment compliance [5]. Moreover, longitudinal studies designed to study lung function in patients with KS have reported no significant correlation between age of diagnosis and deterioration of lung function capacity over time in patients compliant with follow-up appointments and treatments including oxygen supplementation when needed [4]. In our case, the patient's initial non-compliance with oxygen therapy, which resulted in her acute exacerbation was unfortunately unchanged from her discharge non-compliance, which will likely result in future pulmonary deterioration.

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