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Case Report

Kartagener's Syndrome: A Case Report

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Abstract

Background: Kartagener's syndrome is a disease characterized by the co-existence of primary ciliary dyskinesia and situs inversus. Although it is transmitted by an autosomal-recessive pattern of inheritance. Most parents of affected children are normal and have no evidence of impaired ciliary structure or function. The basic problem lies in the defective movement of cilia which affecting the action of cilia lining the respiratory and reproductive systems leading to recurrent respiratory tract infections, and infertility.

Case Presentation: A 36-year-old woman from Cairo, Egypt presented with recurrent episodes of nasal congestion, paranasal discomfort, and productive cough since childhood. Clinical and imaging findings showing features of bronchiectasis and situs inversus totalis. Patient was under follow up for three years from 2015 to 2018 where she had recurrent exacerbation during the period of 2016; which, was improved significantly since 2017 after emphasis of psychosocial support and encourage the patient for regular physiotherapy. She was treated with low dose azithromycin (250 mg OD), mucolytic, social support and chest physiotherapy. She was clinically better with the above management, with significant reduction in the number of respiratory exacerbation.

Conclusions: Patients with Kartagener's syndrome characterized by defective movement of cilia which affecting the action of cilia lining the upper and lower respiratory tract, sinuses leading to recurrent upper and lower respiratory tract infection. Early diagnosis and management of the disease is crucial to prevent the associated complications of lung damage and respiratory failure. The medical treatment only could be not enough. The association of medical treatment with regular physiotherapy, social and psychological support improve prognosis and reduce number of exacerbations. Genetic counseling and fertility investigations should be assessed with all diagnosed cases.

Keywords: *Bronchiectasis; Kartagener's Syndrome; Sinusitis; Situs Inversus; Social Support; Psychological; PCD Primary Ciliary Dyskinesia*

Introduction

Kartagener's syndrome (KS) is a coexistence of PCD and situs inversus; which occurs in 50% of PCD cases. It is a genetic condition with an autosomal recessive inheritance [1,2] characterized by recurrent upper and lower respiratory tract infections since childhood associated with fertility problems in adulthood [1,2].

The first one who describe kartagener syndrome was Siewart in 1904. In 1933, Kartagener recognized the relationship between recurrent respiratory tract infection and situs inversus; where he reported four cases [2]. The estimated prevalence of PCD is about 1 in 30,000 [3] though it may range from 1 in 12,500 to 1 in 50,000 [1]. Early diagnosis and treatment reduces the damage of the respiratory tract and its associated complications [1].

Normal ciliary function is crucial for respiratory host defense through a process of mucociliary clearance. The respiratory ciliary function responsible for carrying the mucous blanket of the upper respiratory tract to the gastrointestinal tract, and to protect the upper airway from exposure to pathogens, allergens, and toxins. Also, cilia improve the motility of sperm, and visceral orientation during embryonic life. Kartagener's syndrome associated with mutations in genes, mainly affecting DNAI1 on 17 chromosome, DNAH5 on 14 chromosome and DNAH11 on 18 chromosome which results in numerous defects including structural abnormalities of the dynein arms, radial spokes, and microtubules of the cilia [5,6,9].

Case Presentations

A 36-year-old woman presented first time to our medical out-patient clinic in June 2015. She presented with progressive cough and expectoration of moderate amount yellow sputum associated with fever (temperature 38.5°C). The patient was tired and toxic. Chest X ray done (Figure 1) showing features of basal lung infiltrations and dextrocardia. Sputum culture was grown for *Klebsiella* which was sensitive to quinolones and penicillin group. Patient admitted in the hospital for three days; CT chest was done (Figure 2 and 3) then discharged after clinical improvement and received a course of antibiotic on Moxifloxacillin 400 mg OD for two weeks. Patient since this time under regular follow up. During 2015 - 2016 she has about seven exacerbations with recurrent pneumonia and sinusitis; where, she was advised for long term oral antibiotic azithromycin guided by culture sensitivity results. During this period patient advised for follow up in dietician clinic and planned for regular course of physiotherapy. She was not compliant for her management plan due to social problems. In 2017, patient referred for psychiatry clinic and started full management plan included in socio- psychological support where her case discussed with the husband and her mother after patient approval where they sharing her plan and expectation. Patient started to visit physiotherapy regularly under family support and encourage her for daily exercise. During 2017 - 2018 patient had only one attack of exacerbation gaining weight and significant improvement of quality of life.

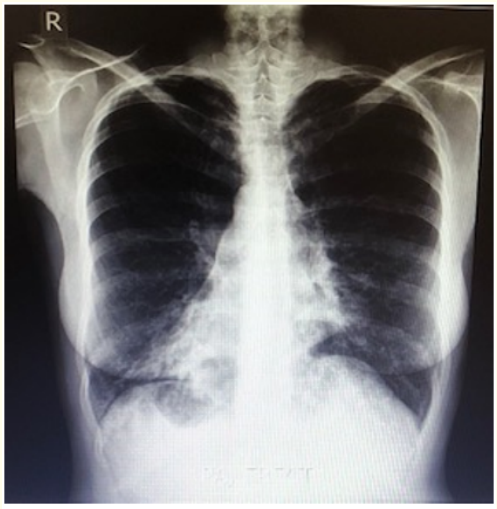

Figure 1

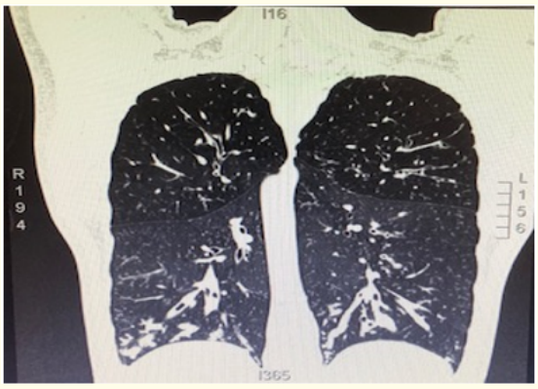

Figure 2

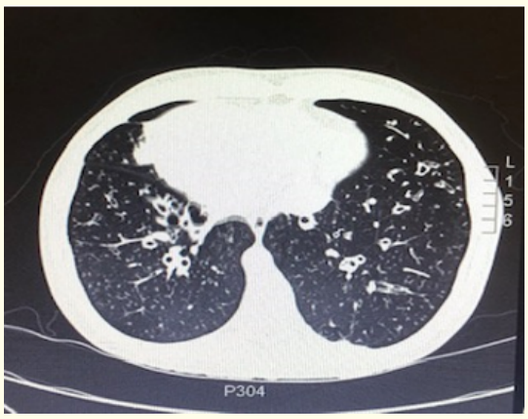

Figure 3

Discussion

Mucociliary transport in the respiratory tract is important for the normal respiratory function by eliminating the respiratory secretion including toxins and different pathogens. Kartagener's Syndrome is a variant of Primary Ciliary Dyskinesia (PCD); characterized by an ultra-structural defect in the formation of cilia leading to poor mucociliary clearance; resulting in recurrent respiratory tract infection and sinusitis since childhood and fertility problems in adulthood [1,2].

Defect in the tiny hair like structure, the cilia lining the respiratory tract (upper and lower), sinuses, eustachian tubes, middle ear and fallopian tubes which are recognized by structural and functional abnormalities of ciliary ultrastructures, encoded by the mutated genes DNAI1 and DNAH5. The genetic mutations affecting the ciliary function through developmental ciliary defect in its structure and movement [5,6].

Abnormal ciliary motility in the respiratory tract decrease mucociliary clearance of the respiratory secretions; which predisposes for recurrent infection and colonization of the bacteria induces chronic inflammation and lung destruction [5-7].

The diagnostic criteria recommended for this syndrome including recurrent respiratory tract infection since childhood associated with one or more of the following: (a) situs inversus or dextrocardia in a patient or a sibling, (b) Immotile spermatozoa, (c) Absent or impaired tracheobronchial clearance, and (d) cilia ultrastructural defect on electron microscopy [7,8].

Screening tests for diagnosis of Kartagener's Syndrome include:

- Reduction of exhaled nasal nitric oxide level (~10% of normal).
- Prolonged saccharin clearance time (> 1 hour) assessing nasal epithelial mucociliary function.
- High-speed video microscopy for assessing ciliary beat frequency and pattern.
- Electron microscopic for detecting ultrastructural ciliary defect.
- Genetic testing for DNAI1 and DNAH5 mutations are confirmatory laboratory tests [6,7].

Our patient presented clinically with recurrent attacks of lower respiratory tract infection, and sinusitis since childhood most of attacks in the last three years associated with *Klebsiella* infection. Chest X ray and HRCT chest showing bronchiectatic changes more in the lower lobes of the lungs, cardiac apex and gastric fundus in the right side. Other specific laboratory investigations not done [7,9,10].

The medical, physical and psychological treatment for patients with KS including chest physiotherapy, mucolytics, and antibiotics including long-term low-dose prophylactic antibiotic could required in those with recurrent chest infection. Encourage patient for immunization for influenza, pneumococcal, and *Haemophilus influenza* vaccines are crucial [7,9,10].

Conclusion

Kartagener syndrome is a disease affecting the function of cilia in the upper and lower respiratory tract leading to recurrent sinusitis, otitis media and pneumonia. In addition, the patients have fertility problems due to improper ciliary function in fallopian tube for female patients and sperms tail in men. The patients need not only long term plan for medical treatment; but also social, family and psychological support which declared to improve overall prognosis and quality of life.

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