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Case Report

Kartagener syndrome with renal amyloidosis: a case report

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ABSTRACT

Kartagener syndrome is a rare disorder caused by defective ciliary function. It is described as a triad of cystic bronchiectasis, chronic sinusitis, and situs invertus. Renal involvement, although uncommon, is reported. The available evidence in literature consists of case reports and series, which describe various patterns of renal involvement. We hereby describe a case of a young female who presented with advanced renal failure and hypertension requiring urgent hemodialysis. On evaluation, she was found to have the classic triad of bronchiectasis, chronic sinusitis, and situs invertus leading to the diagnosis of Kartagener syndrome. On workup for the cause of renal failure, we found membranoproliferative glomerulonephritis due to amyloidosis secondary to bronchiectasis. This is a rare association and only 1 case has been reported in the available literature. There are no consensus guidelines regarding treatment of this condition, due to paucity of available data and because of rarity of the condition. This disease should be reported to augment the available literature.

Keywords: Kartagener syndrome, Renal amyloidosis, Chronic kidney disease, Autoimmune disorder, Primary ciliary dyskinesia

INTRODUCTION

Kartagener syndrome is a rare disease, also termed as primary ciliary dyskinesia. It is described as a triad of cystic bronchiectasis, chronic sinusitis and situs. Renal involvement although uncommon, is not rare. The available evidence in literature consists of case reports and series, which describe various different patterns of renal involvement. We hereby describe a case of a young female with Kartagener with advanced renal failure.

CASE REPORT

A 41-year-old female was brought to the emergency department with complaints of productive cough, nausea, vomiting and shortness of breath. She had a history of cough with expectoration since childhood and primary infertility which was never investigated and a sinus surgery for chronic sinusitis. On examination, blood pressure in the right arm, supine position was 178/112 mmHg.

On appearance she looked pale and emaciated. On auscultation, the chest was full of coarse crepitations and the apex beat was not localised. She underwent routine investigations, which revealed renal dysfunction and anaemia.

Patient report is mentioned in the table (Table 1).

She was admitted and started on anti-hypertensive medications. But she was becoming nauseous, and her blood pressure failed to lower down. So, she was planned for urgent initiation of renal replacement therapy in the form of haemodialysis. A double lumen non tunnelled haemodialysis catheter was inserted under ultrasound guidance and a post procedure chest X-ray was done (Figure 1).

Table 1: Initial blood reports.

Parameters	Values
Creatinine	15 mg/dl
Urea	257 mg/dl
Uric acid	7.6 mg/dl
Sodium	132 mmol/l
Hb	7.4 g/dl
WBC	7610/mm^3
Platelet count	450000/mm^3
Urine 24 hours protein/creatinine ratio	11.24
Calcium	8.56 mg/dl
Magnesium	2.46 mg/dl
Phosphorus	4.14 mg/dl
Potassium	4.4 mmol/l
Chloride	110 mmol/l
Urine microscopic examination	15-20 pus cells/hpf, 3-4 RBC/hpf, no cast/crystal

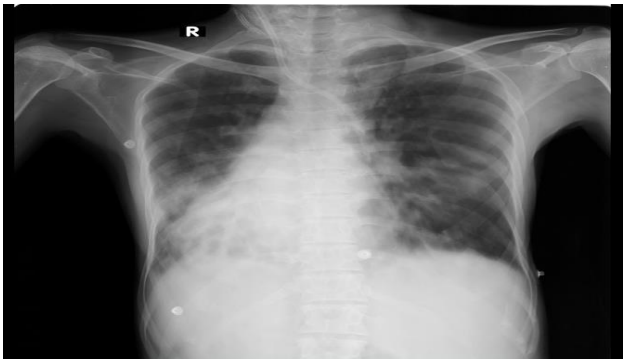

Figure 1: Chest X-ray showing dextrocardia and the position of the tip of the dialysis catheter in the innominate vein.

Chest X-ray confirmed dextrocardia and the position of the tip of the dialysis catheter in the innominate vein. After stabilisation, she underwent a CT scan (Figure 2a and b) of the chest which revealed changes of cystic bronchiectasis involving diffuse lung parenchyma across all lung lobes.

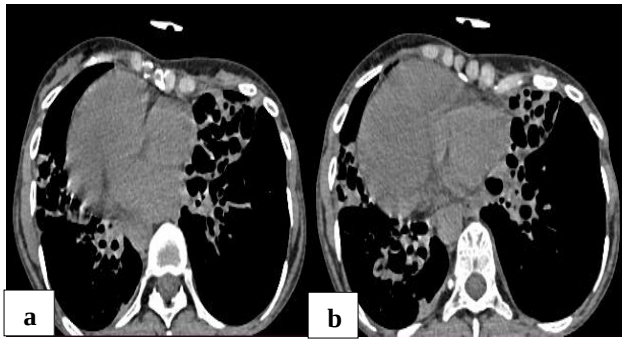

Figure 2: (a) and (b) CT thorax cystic bronchiectasis involving diffuse lung parenchyma across all lung lobes.

Ultrasound imaging of the abdomen confirmed reverse lateralisation of abdominal viscera i.e. situs inversus totalis. Both kidneys were normal in size and shape and highly echogenic, with partial loss of corticomedullary differentiation with no evidence of obstruction.

Clinical diagnosis of PCD- Kartagener's syndrome was made. No molecular, genetic or ultrastructural testing was done due to unavailability. X-ray of paranasal sinuses (Figure 4) revealed changes of chronic rhinosinusitis.

Patient was managed with broad spectrum antibiotics, antihypertensives, mucolytics and antiemetics. Multiple sessions of haemodialysis were done with anaemia correction.

Patient suffered an episode of unprovoked seizure during dialysis course which managed supportively on the lines of dialysis disequilibrium and patient recovered soon. An MR imaging of the brain was performed which revealed no parenchymal abnormality. Sputum culture showed growth of pseudomonas aeruginosa, so antibiotic was changed accordingly.

After stabilization, ultrasound guided kidney biopsy was performed which revealed: mesangioproliferative pattern with congophilia (Figure 3). On light microscopy, there was diffuse irregular mesangial matrix expansion. The mesangial material looked pale, eosinophilic with H&E stain. It did not stain with PAS and silver methenamine stains. Greenish birefringence was noted in Congo red stained sections viewed under polarized light. Out of 44 glomeruli observed, 18 (35.5%) were globally sclerosed/ solidified. Direct immunofluorescence was negative for immunoglobulins, complement factors and kappa/lambda chains. Immunohistochemistry on the extracellular deposited material showed intense positivity for serum amyloid associated protein (SAAP). The histopathological examination suggested amyloid deposition, secondary to chronic inflammation, which in this case could be bronchiectasis.

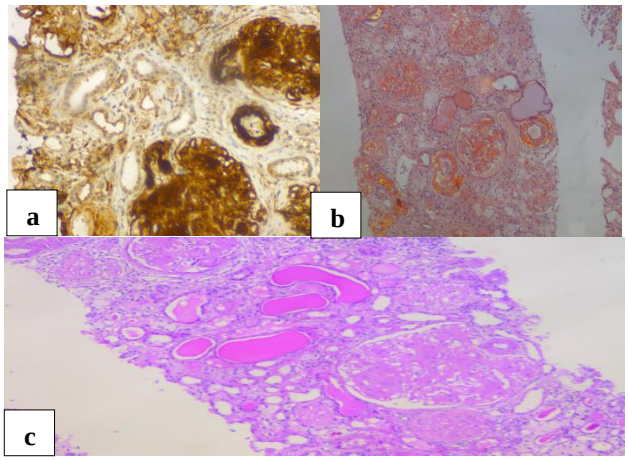

Figure 3: (a), (b), and (c) Renal biopsy showing mesangioproliferative pattern with congophilia.

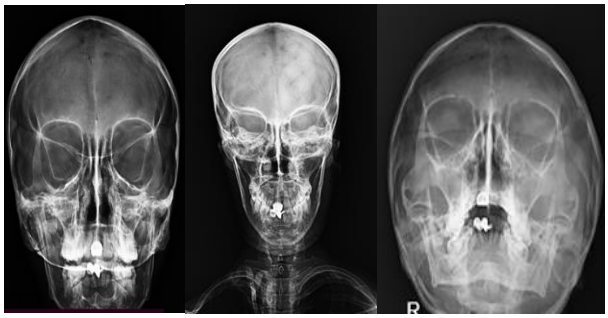

Figure 4: X-ray PNS and X-ray skull showing chronic rhinosinusitis.

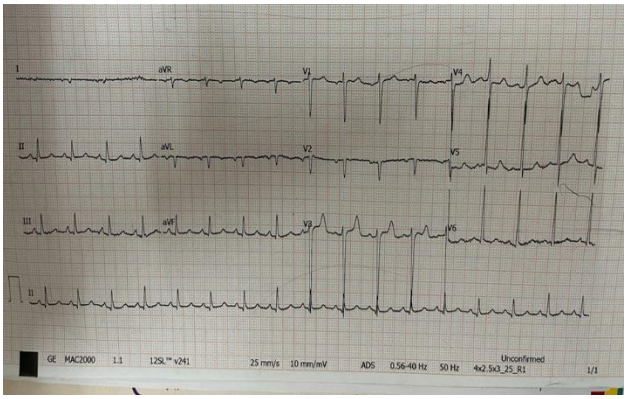

Figure 5: ECG showing dextrocardia.

The patient was managed with hemodialysis, antibiotics, supportive pulmonary care including oxygen inhalation, mucolytics, bronchodilators and chest physiotherapy. The sputum culture confirmed Pseudomonas, and the antibiotics were changed accordingly. After improvement, she was vaccinated against pneumococcus and influenza. She is currently on thrice weekly in-centre hemodialysis with regular monitoring for any change in her renal function.

DISCUSSION

Primary ciliary dyskinesias are genetically heterogeneous disorders of ciliary motility, also called immotile cilia syndrome, characterized by ineffective ciliary clearance. These are rare disorders and occur at an estimated frequency of 1 out of 32000 births. Kartagener Syndrome and Young's syndrome are the two important variants.[1]

It was first described by Siewert in 1904 and subsequently the mechanism underlying this clinical constellation was stated to be ciliary immobility; however, the syndrome including clinical triad of chronic sinusitis, bronchiectasis, and situs inversus was first recognized by Kartagener in 1933.[2,3]

Most common mode of inheritance is autosomal recessive and multiple mutations affecting dynein gene present in heavy or intermediate chain are described. Common mutations involve the heavy (dynein axonemal heavy chain 5) or intermediate (dynein axonemal intermediate chain 1) (DNAI1 and DNAH5) chain of dynein genes in ciliary outer dynein arms. Ultrastructural analysis commonly reveals defective dynein arms, and other axonemal components are rarely involved.[4,5]

The dynein arms are a set of radial protuberances which originate from the longitudinally situated nine pairs of microtubules. They are organized in a circumferential manner looping around the cilia. When the peripheral microtubules slip over the inner microtubules the cilia undergo bending. The ATPase dynein arms are the primary source of energy in this. The cilia work in tandem with each other to impel the mucous layer towards the cephalic direction. This is achieved because of the arrangement of the ultrastructure, which is symmetrical in the entire plane.[6,7]

To make sure all the viscera are oriented anatomically and host defense of the respiratory unit and for the sperm motility, ciliary function is vital.[8]

Right from the neonatal age, child can exert symptoms such as respiratory distress. Recurrent infections including otitis, pneumonitis, and rhinitis can be the reason for prolonged symptoms with advancing age. Such patients demonstrate a characteristic purulent expectoration which can be because of obscured airflow and ineffective clearance of secretions leading to long standing respiratory infection. The end outcome of this pathology is a vicious cycle of bronchial dilatation, ineffective clearance of secretions, recurrent infections and additional bronchial damage. The ultimate manifestation of the disease is a classic troika of symptoms: recurrent infection + chronic cough + excessive purulent sputum production.

The embryological nodal cilia are dysfunctional leading to laterality defects (such as situs invertus totalis and infrequently heterotaxy and congenital heart disease) in almost half of the cases. Infertile males are also common because of the defunct sperm tails axonemes. Similarly, female infertility is also reported.

The ciliary defect is not exclusive to Kartagener syndrome. There is a significant clinical overlap seen with polycystic liver and kidney disease, central nervous system problems including retinopathy and hydrocephalus, and biliary atresia. Some ciliopathies involving sensory cilia, including autosomal dominant or recessive polycystic kidney disease, Bardet-Biedl syndrome, and Alstrom syndrome. These pathologies can also superimpose to chronic respiratory symptoms, also bronchiectasis.[9]

Histopathological diagnosis of PCD is a dilemma. The uncommon procedure named saccharin method in which the cilia is visualized under electron microscopy and tested for mucocilliary function by nasal scrape or brush biopsy is also not gold standard, as around about 3-30% patients of PCD with clinical manifestations can show a normal ciliary structure. Hence diagnosis largely remains clinical.

Renal involvement is unusual in this entity with only a few cases reported worldwide. In a case the pattern of renal involvement with ciliary dyskinesia was polycystic kidney disease, which authors believed was a broader spectrum of ciliary dysfunction. Also, one case each have reported IgA nephropathy, focal segmental glomerulosclerosis and amyloidosis. One case has reported renal cell carcinoma in a patient with Kartagener syndrome. One case had chronic renal failure without any histopathological examination performed.[10-14]

Differential diagnosis of this syndrome are Young's syndrome, cystic fibrosis, alpha-1 antitrypsin deficiency, immunodeficiency states associated with recurrent infections like IgG or IgA deficiencies etc.

Cessation of disease progression and pulmonary improvement should be the management goal. Aggressive antimicrobial killing, improving airway clearance and preventing the repeated infections which could potentially enflame the bronchiectasis is a strong foundation. Influenza and Pneumococcal vaccines prophylactically along with timely pulmonary toilet has favorable outcome. Patients refractory to antibiotics or those who develop recurrent pneumonitis/hemoptysis can be a candidate for segmental lung resection or lobectomy.

For chronic rhinosinusitis, endoscopic sinus surgery may be required and for infertility, assisted reproduction techniques are helpful. For secondary amyloidosis, no specific therapy has proven beneficial.

CONCLUSION

We hereby report a rare case of Kartagener syndrome having chronic rhinosinusitis, cystic bronchiectasis and situs invertus along with renal involvement in the form of secondary amyloidosis leading to advanced renal failure.

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