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Case Report

Kartagener Syndrome: A Case Report

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ABSTRACT

Kartagener syndrome is a rare, autosomal recessive genetic disorder that causes defects in the action of ciliary movement, characterized by clinical triad of situs inversus, chronic sinusitis, and bronchiectasis. We present the case of an 18-year-old male with repeated respiratory infections. He was diagnosed with Kartagener syndrome based on his clinical presentation and imaging features. In patients presenting with recurrent upper and lower respiratory tract infections, Kartagener syndrome should always be kept in mind. The correct diagnosis of this disorder in early life is very important to prevent complications and improve a patient's quality of life.

Keywords: Kartagener Syndrome (KS), Primary Ciliary Dyskinesia (PCD), Situs Inversus, Bronchiectasis, Dextrocardia.

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INTRODUCTION:

Kartagener syndrome (KS) is a subset of a larger group of ciliary motility disorders called primary ciliary dyskinesias (PCDs)[(1)]. KS is an inherited autosomal recessive disorder that causes defects in the action of ciliary movement, comprising triad of sinusitis, situs inversus, and bronchiectasis[(2)].Siewert[(3)] first explained the combination of situs inversus, chronic sinusitis, and bronchiectasis in 1904. Manes Kartagener[(4)]first recognized this clinical triad as a congenital syndrome in 1933 and described this syndrome bearing his name. Its estimated incidence is approximately 1 in 30,000 live births[(5)] . Normal ciliary function is critical for respiratory host defense and motility of sperm and ensures proper visceral orientation during embryogenesis. In KS, the gene mutation at *DNAI1* and *DNAH5* leads to impaired ciliary motility, which predisposes to recurrent sinopulmonary infections, infertility, and errors with left–right body orientation[(5)]. Males and females are affected equally[(6)] .

This syndrome should be kept in mind in differential diagnosis of chronic respiratory infections and must be diagnosed as early as possible to improve a patient's quality of life. Appropriate and early treatment and care of these patients can improve prognosis and prevent the progress of respiratory problems such as bronchiectasis and pneumonia. This study aims to contribute toward a greater understanding of KS.

ETHICS APPROVAL

The patient's mother consented to the publication of this deidentified case report. Institutional review board approval is not required for de-identified single case reports.

CASE REPORT

A 18 years old male,non smoker ,non alcoholic was presented in medicine opd on 7 august 2023 . Patient was referred from ENT opd in view of ECG reporting.He went to ENT opd with chief complaints of recurrent episodes of nasal congestion,2 days history of productive cough,cold and running nose. There was no history of fever, hemoptysis or travel outside the city in the recent past. There was no history of allergy, sleep apnea, pulmonary tuberculosis and weight loss.He had no exposure to birds or contact with sick person.

He had a history of frequent cold and repeated respiratory infections. In his family there was no history of asthma, tuberculosis, and atopy. His parents marriage was non consanguineous. His all siblings were healthy and alive without any comorbidities.

At the time of presentation the patient was vitally stable, maintaining saturation on room air. On cardiovascular examination, an apex beat was felt on the right fifth intercostal space along the midclavicular line. Heart sounds were best audible on the right side of his chest. On chest auscultation, there was diminished air movement in both lungs, bilateral coarse crackles at the bases of both lungs were audible. Audiogram was done on 28 july 2023 suggestive of bilateral profound hearing loss. Semen analysis was done and suggestive of azoospermia.

Other physical exam findings were unremarkable. Chest X-ray revealed cardiac apex and aortic arch on right side, suggesting dextrocardia, and right-sided stomach air suggesting a case of situs inversus, with normal lungs field (Figure 1).

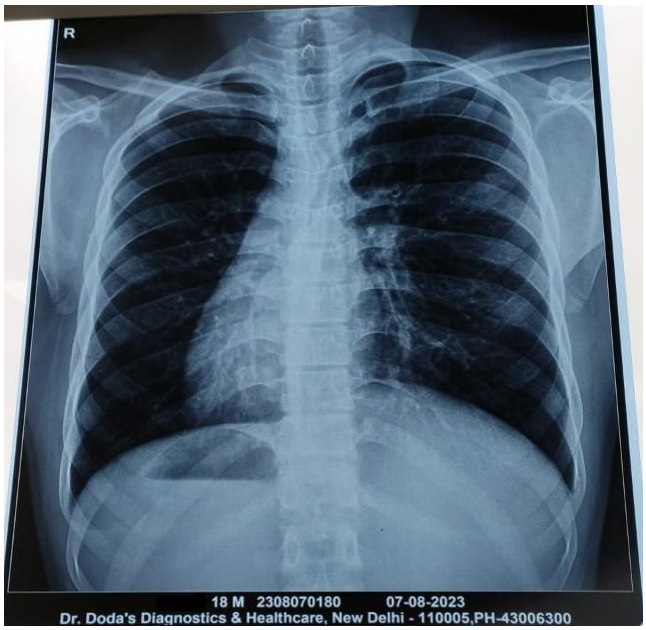


Figure 1: Chest x-ray showing dextrocardia

Electrocardiogram showed signs of dextrocardia, inverted P and T waves in lead I and aVL (global negativity),positive P and T waves and positive QRS complex in lead aVR (global positivity) , right axis deviation and QRS complexes get progressively smaller in leads V 1–V 6.(Figure 2)

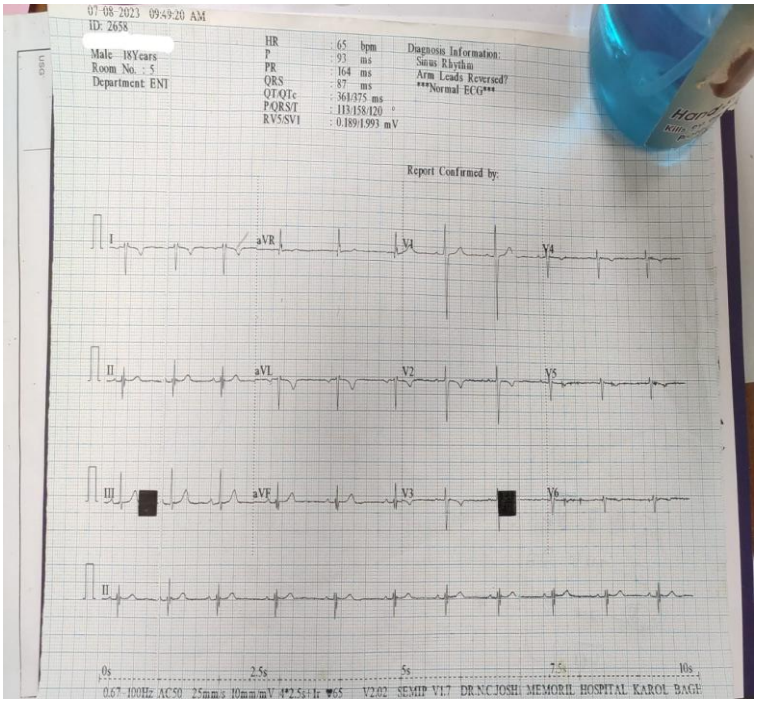
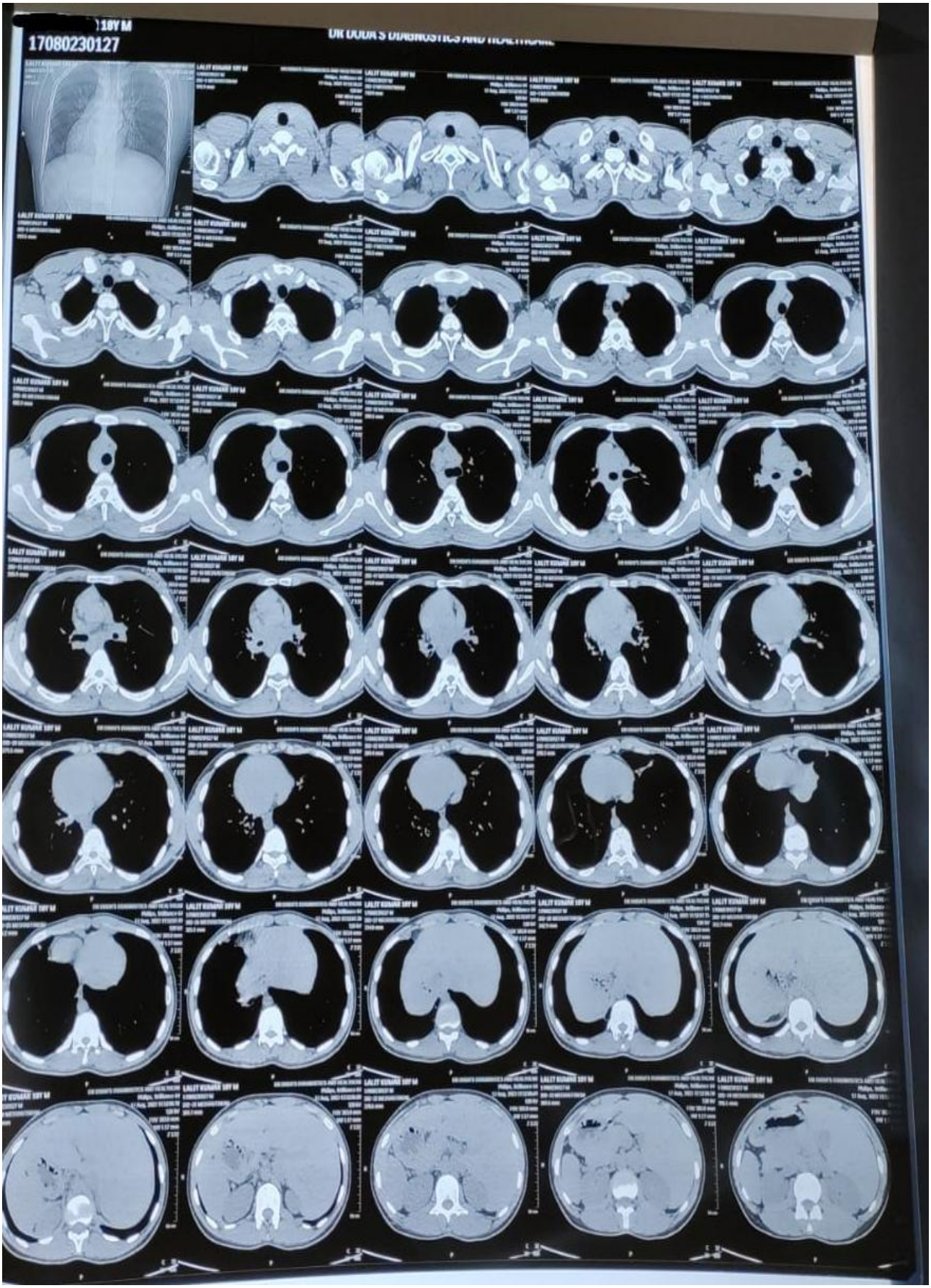


Figure 2: ECG Showing dextrocardia with poor wave progression

A chest computed tomography (CT) scan showed dextrocardia with radiographic evidence of bronchiectasis (Figure 3).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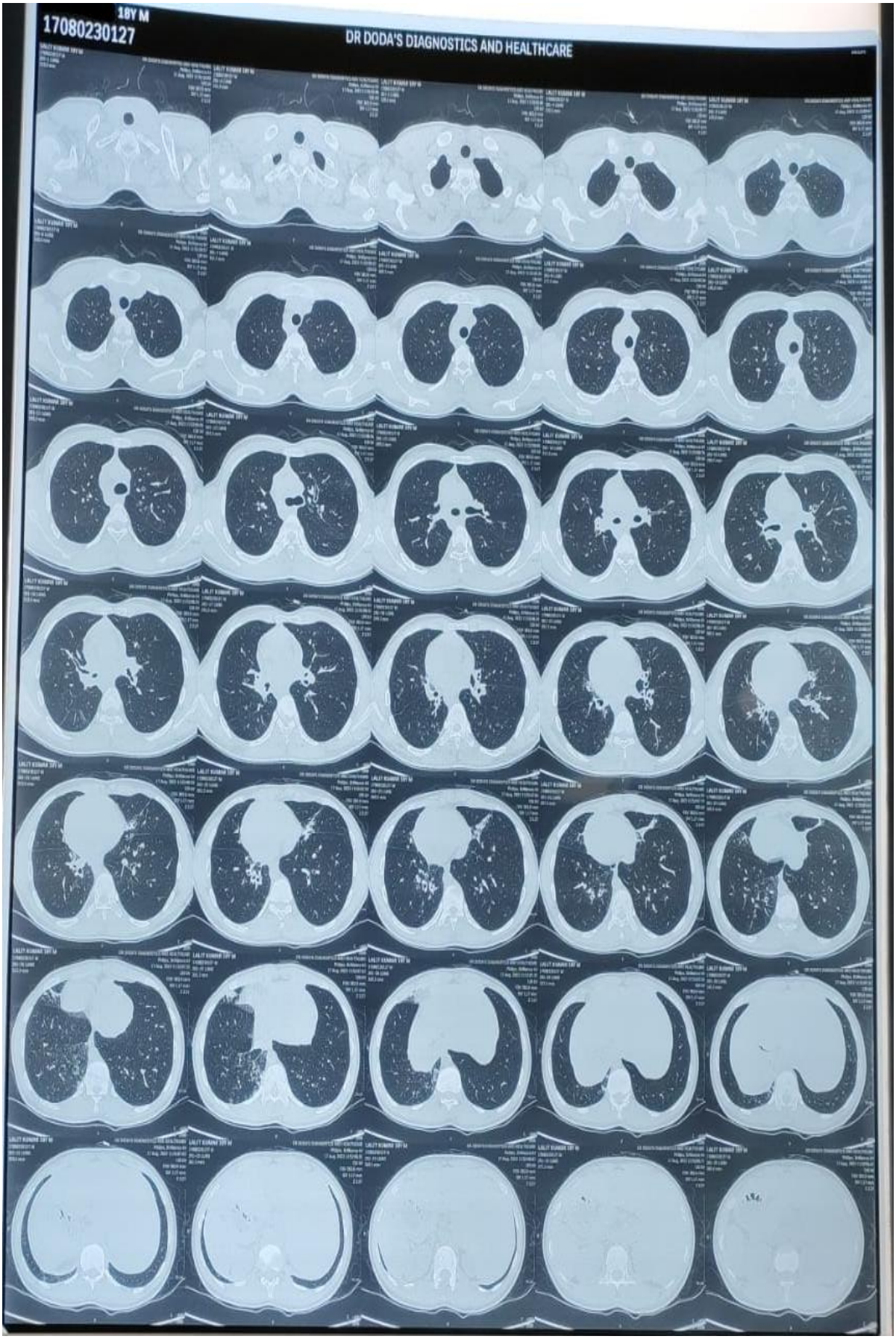
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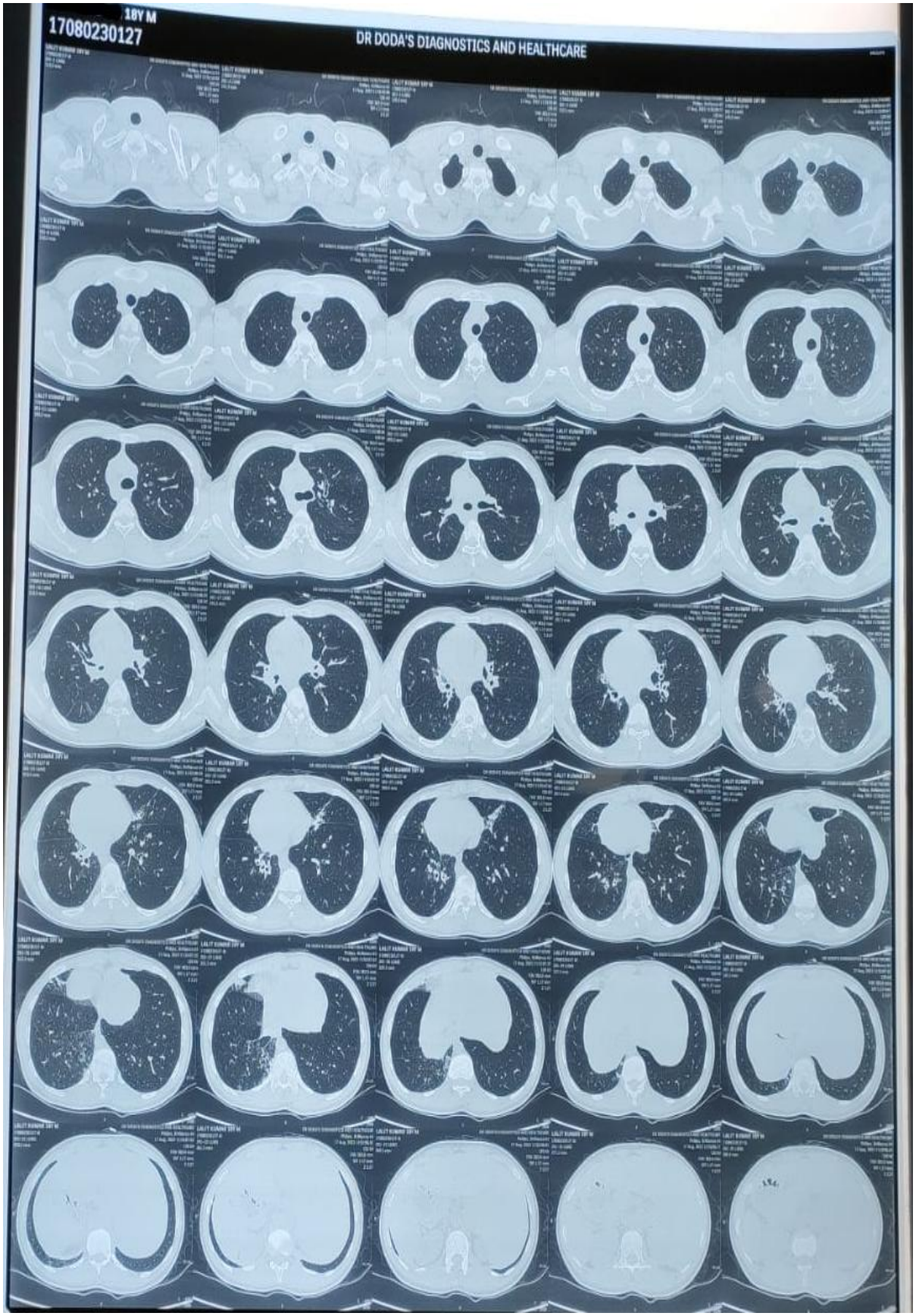
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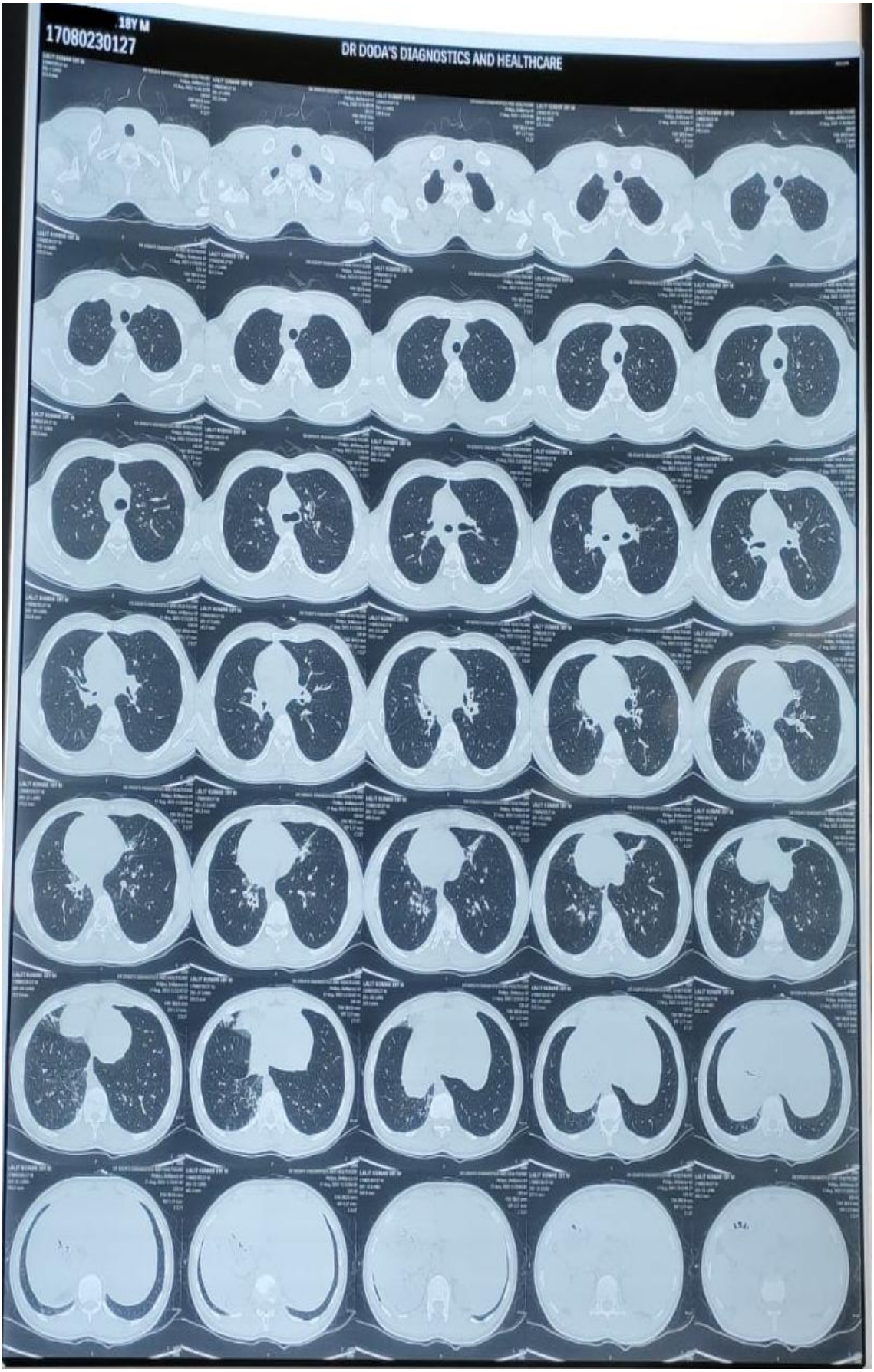
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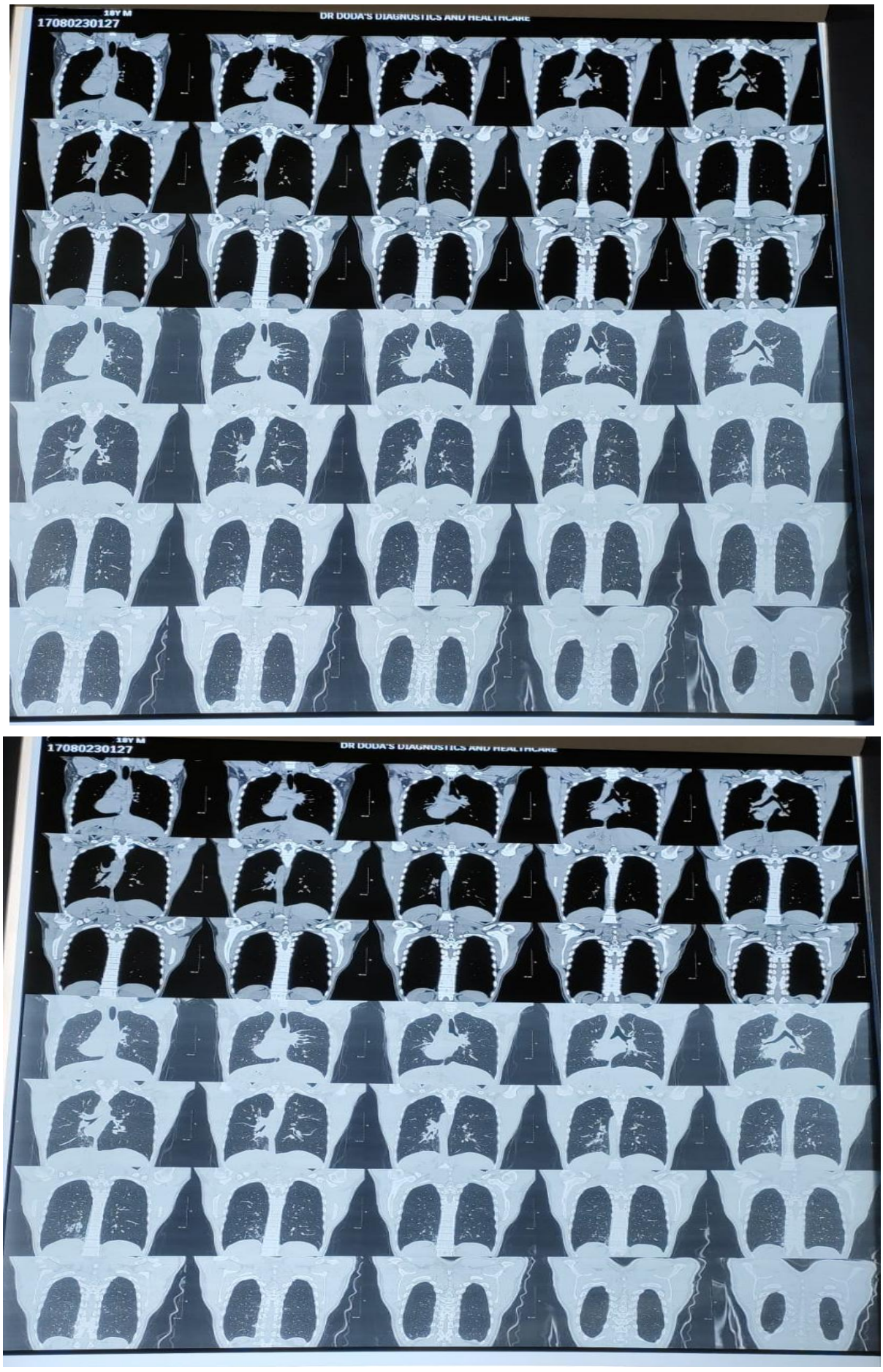

Figure 3: HRCT chest showing bronchiectatic changes in the both lungs fields and bilateral ground glass opacities

Clinical and imaging findings revealed chronic sinusitis, bronchiectasis, dextrocardia, and situs inversus. He was treated with orally administered antibiotics, mucolytic, and chest physiotherapy. He was symptomatically better with the above therapy, and started on a long-term low-dose prophylactic antibiotic.

FINAL DIAGNOSIS

A diagnosis of KS was made based on clinical presentation and imaging features. The case represented a classical KS.

DISCUSSION

Disorders of ciliary motility may be congenital or acquired. Congenital disorders are labeled as PCDs. Nearly 50% of PCD patients have situs inversus. Such cases of PCD with situs inversus are known as Kartagener's syndrome [(1)]. clinical symptoms in PCD varies,some may begin with neonatal respiratory distress ,or later develop chronic productive cough due to bronchiectasis, asthma, chronic rhinosinusitis.Other complications are ectopic pregnancy and infertility both in males and in females. [(7)] . Clinical and radiographic evidence of bronchiectasis develops as the disease progresses; bronchiectasis and obstructive impairment may be apparent in preschool children[(8)] .

Normal ciliary beating is also necessary for visceral rotation and orientation during embryonic development. Patients with KS may have either situs solitus where there is dextrocardia only or situs inversus totalis, where all the visceral structures are on the opposite side [(2)]. In this case, it was a situs inversus totalis because the cardiac position, as well as the abdominal viscera, was a mirror image of the normal anatomy.

Most of the disease-causing mutations are said to involve two genes coding for the dynein axonemal heavy chain 5 (DNA H5) and dynein axonemal intermediate chain 1 (DNA I1)[(9)] . The complete syndrome has high familial evidence, appearing only in one generation and multiple siblings. These features and the high incidence of consanguinity among the apparently normal parents of affected children support the contention that the genetic abnormality is carried as an autosomal recessive gene[(6)].

The diagnostic criteria recommended for this syndrome include history of chronic bronchial infection and rhinitis from early childhood, combined with one or more of following features: *(i)* situs inversus or dextrocardia in a patient or a sibling, *(ii)* alive but immotile spermatozoa, *(iii)* absent or impaired tracheobronchial clearance, and *(iv)* cilia showing characteristic ultrastructural defect on electron microscopy[(5)] . According to this diagnostic criteria, diagnosis of KS was made in our patient.

Diagnosis can be made by tests to prove impaired cilia function, biopsy, and genetic studies. Semen analysis of postpubertal males may reveal either abnormal sperm motility or aspermia [(10)].

The Saccharin test is also used for diagnosis. It is a screening test to detect abnormal mucociliary clearance. This test measures the time taken for a pellet of saccharin placed on the inferior turbinate to be tasted, 30 min is the cutoff point that discriminates normal people from patients with impaired nasal mucociliary clearance[(11)]. Measuring exhaled nasal nitric oxide involves measurement of the expired NO from one nostril. There are no agreed cut-off values but nasal nitric oxide levels in patients with PCD are consistently only 10%–20% of the average normal values[(12)].Mucociliary transport, which is reduced in these patients, can be measured in situ by administering an inhalation aerosol of colloid albumin tagged with 99Tc. This test uses aerosol particles tagged with 99mTc and external measurement of the radioactivity[(13)] . Electron microscopy of a nasal or bronchial biopsy can reveal defected cilia structure. Genetic testing for mutations in the genes *DNAI1* and *DNAH5* is available through specialized laboratories[(14)] . In the present case, these tests were not performed because of the technical condition in our hospital. However, these procedures are invasive and available only at specialized centers; therefore, the diagnosis of KS in this case was clinical, supported by imaging studies.

As a genetic disease, KS has no definite treatment. Treatment of patients is symptomatic and includes intermittent or constant oral or intravenous administration of antibiotics to treat respiratory infections. Bronchiectasis and pneumonia should be treated with inhaled bronchodilators, mucolytics, oral corticosteroids, and chest physiotherapy. Administration of influenza and pneumococcal vaccines is also necessary to prevent frequent infections[(15)] . Although there is no specific treatment for this rare syndrome, failure to diagnose this may subject the patient to unnecessary repeated admissions to hospitals, investigations, and inappropriate treatment. Genetic counseling and fertility issues should be addressed once Kartagener's syndrome is diagnosed. For end-stage KS, double lung transplantation may be useful. Wang et al.[(16)] reported successful pulmonary surgery in a 49-year-old woman, which involved double lung transplantation.

Early diagnosis and clinical follow-up at regular intervals are essential in these patients to prevent complications. A long-term low-dose prophylactic antibiotic is required in those with frequent exacerbation of bronchiectasis (≥3 times/year). Influenza and pneumococcal vaccination should be routinely given

CONCLUSIONS

A high index of suspicion is needed to make an early diagnosis so that timely treatment options may be offered to prevent problems associated with it. The correct diagnosis of KS in early life is essential in the overall prognosis of the syndrome, as many of the long-term complications can be prevented if timely management is instituted, as was done in this case.

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