Journal of
Medical Case Reports

CASE REPORT Open Access

Kartagener syndrome with minimal change disease: a case report

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Abstract

Background Kartagener syndrome is characterized by chronic sinusitis, bronchiectasis, and total visceral transposition. While there are few reports of Kartagener syndrome combined with kidney disease, there are none that specifically report Kartagener syndrome in conjunction with minimal change disease. This is the first report of a rare case of Kartagener syndrome with minimal change disease, which presented with the typical triad and was clinically diagnosed.

Case presentation A 24-year-old Chinese woman was admitted to the hospital with 2 weeks of foamy urine and edema of the eyelid and lower limbs. After admission, the examination indicated nephrotic syndrome and total visceral transposition. Computed tomography imaging revealed sinusitis bronchiectasis, and she was diagnosed with minimal change disease with Kartagener syndrome. A renal biopsy revealed minimal changes. After symptomatic antiinflammatory therapy, the patient was given telmisartan 50 mg orally once daily to reduce urinary protein levels. A total of 1 month after discharge, her 24-h urine protein content was < 1 g, with normal liver function and improved kidney disease.

Conclusion We describe a rare case of Kartagener syndrome accompanied by glomerular disease and minimal change disease. The patient was treated symptomatically with antiinflammatory agents and will be monitored long term. We believe our findings will provide valuable guidance and reference for the treatment of such cases in the future.

Keywords Case report, Kartagener syndrome, Minimal change disease, Nephrotic syndrome

Introduction

Background

Kartagener syndrome is a primary ciliary dyskinesia (PCD) disease, accounting for 50% of PCD diseases [1]. PCD is an inherited clinical syndrome, typically following an autosomal recessive pattern, with few cases of X-linked inheritance [2]. It is a rare genetic condition characterized by abnormal or absent ciliary movement, with a prevalence of 1:10,000–20,000 live births [3]. Impaired ciliary movement inhibits mucociliary clearance, and the bacteria-filled mucus stays in the airways of the ears, nose, sinuses, and lungs, causing inflammation and repeated infections. In addition to affecting mucociliary clearance, PCD also results in abnormal development or placement of internal organs, resulting in a complete reversal of the chest and abdominal organs, appearing as a mirror image of normal placement. The absence of cilia and sperm motility in these patients can also lead to male infertility. Cilia in the fallopian tubes are damaged, leading to an increased chance of infertility or ectopic pregnancy in women. The exact prevalence of subfertility in PCD is unclear but appears to be higher in men (up to 83% affected) than in women (up to 61% affected) [4]. PCD is diagnosed on the basis of the electron microscopic structure of the

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respiratory cilia obtained by nasal scraping or bronchial brush biopsy. Kartagener syndrome is characterized by chronic sinusitis, bronchiectasis, and total visceral transposition. While there are few reports of Kartagener syndrome combined with kidney disease, there are none that specifically report Kartagener syndrome in conjunction with minimal change disease. Here, we describe a case of Kartagener syndrome combined with minimal change disease, which presented with the typical triad and was clinically diagnosed. The patient's condition improved after symptomatic treatment. In addition to investigating the pathogenesis of Kartagener syndrome combined with renal disease, we believe our case report provides valuable information for medical professionals treating such patients.

Case presentation

The patient was a 24-year-old Chinese woman who was admitted to a local hospital with foamy urine for 2 weeks and edema of the eyelids and lower limbs for 1 week. Routine urine tests showed urinary protein > +3. Her liver (albumin 16.2 g/L) and renal functions were normal, and nephrotic syndrome was diagnosed. The patient was admitted to our hospital for a renal puncture biopsy.

Laboratory data

Blood cell analysis showed a white blood cell count of 13.4 × 10^9/L (normal range: 3.5–9.7 × 10^9/L); neutrophilic granulocytes at 80.1% (normal range: 42.3–71.5%); and blood platelets of 446 × 10^9/L (normal range: 135–350 × 10^9/L). Urine analysis revealed that urine protein was 4+; liver function tests revealed that albumin was 19.1 g/L (normal range: 35–53 g/L); triglycerides were 2.03 mmol/L (normal range: 0.4–1.69 mmol/L); and cholesterol was 11.15 mmol/L (normal range: 3.36–5.69 mmol/L). She had normal renal function, with creatinine levels at 39.3 umol/L (normal range: 45–84 umol/L).

Symptoms

The patient felt hot, and she had a body temperature of 37.4 ℃, cough with yellow sputum, and shortness of breath. There was no chest tightness when lying flat at night, no dizziness or headache, no nausea or vomiting, no body rash, no joint pain, no photoallergy, and no alopecia.

Personal history

The patient had a history of recurrent respiratory infections due to bronchiectasis. Total visceral translocation was detected by electrocardiogram during childhood. The patient followed a good diet, had good sleep and normal stools, and had recently gained more than 10 kg in weight. She was sterile and had no children. The patient had no history of exposure to renal toxins and no application of nonsteroidal antiinflammatory drugs (NSAIDs).

Family history

Her mother had diabetes, and her sister had dextrocardia.

Physical examination

Her body temperature was 37.4 ℃. Her blood pressure (BP) was 110/80 mmHg. Her respiratory rate (R) was 16 bpm, and her pulse (P) was 80 bpm. The patient exhibited eyelid edema and a coarse breathing sound with moist crackles. The heart apex beat was 0.5 cm inside the fifth intercostal clavicular line at the right margin of the sternum. A heart sound auscultation was located on the right side, showing a regular heart rhythm with no pathological murmurs. In addition, she had mild edema in both lower limbs.

Urine analysis

The urine protein level was 2+ to 3+ at admission, and it was 2.41, 2.88, and 5.02 g/24 h later, with normal renal function.

Liver function

Total protein 39.2 g/L, and albumin was 16.5 g/L.

Other tests

No abnormalities in complement 3 (C3), complement 4 (C4), or antineutrophil cytoplasmic antibodies (ANCA); tests were negative for antinuclear antibody series. She had normal thyroid function; tests were negative for hepatitis B surface antigen and hepatitis C antibody, human immunodeficiency virus (HIV), and treponema pallidum particle agglutination (TPPA). C-reactive protein (CRP) was normal. Electrocardiography (ECG) revealed dextrocardiography and sinus rhythm. Cardiac Doppler ultrasound results showed no abnormalities in the dextrocardia or intracardiac structures, and the overall systolic function of the left ventricle was normal at rest.

Computed tomography (CT) findings

CT imaging showed visceral transposition of the thoracic organs, bronchi of both lungs (Fig. 1A), and abdominal organs (Fig. 1B), which appeared as a mirror image of normal placement. Lesions were detected in the middle and lower lobes of the right lung with pleural effusion. Bronchiectasis was observed in both lungs (Fig. 1A). Sinus CT showed bilateral maxillary sinuses, ethmoid sinuses, sphenoid sinusitis, and thickened left inferior turbinate mucosa (Fig. 1C).

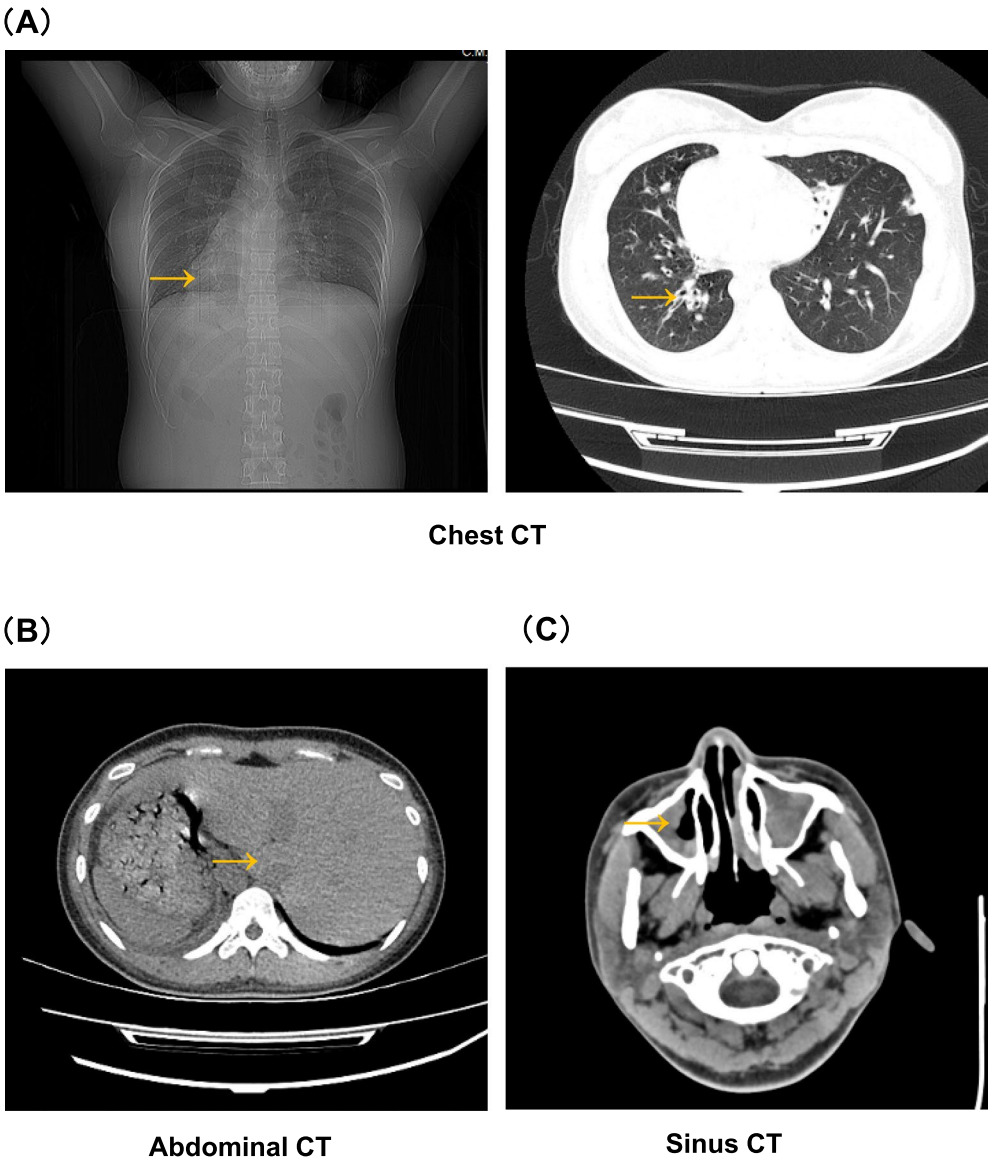

Fig. 1 Computed tomography findings. **A** Chest computed tomography. **B** Abdominal computed tomography. **C** Sinus computed tomography. Chest computed tomography showing the transposition of the thoracic organs, bronchi of both lungs, as a mirror image of the normal placement. Dextrocardia as shown by arrow in **A** and bronchiectasis of both lungs as shown by arrow in **A**. Abdominal computed tomography indicates abdominal organ transposition, as shown by arrow in **B**. Sinuses computed tomography showed bilateral maxillary sinuses, ethmoid sinuses, sphenoidal sinusitis, and thickened left inferior turbinate mucosa, as shown by arrow in **C**

Kidney biopsy

Pathological findings showed 12 glomeruli, of which one had global sclerosis, while the others had minor changes. Segmental mesangial hyperplasia was observed in some glomeruli, with visceral epithelial swelling. In addition, the patient was diagnosed with turbidimetric degeneration of renal tubular epithelial cells, focal tubular atrophy, focal interstitial edema, and focal inflammatory cell infiltration, including monocytes and lymphocytes (Fig. 2). Immunofluorescence staining indicated immunoglobulin (Ig)M was deposited in the mesangial region nonspecifically. Owing to limited conditions, electron microscopy was not performed. The possibility of minimal change disease was considered.

Diagnosis of minimal change disease

On the basis of the patient's history of infertility, bronchiectasis, repeated lung infections, dextrocardia, and total visceral transposition, she was diagnosed with Kartagener syndrome. She was recommended to

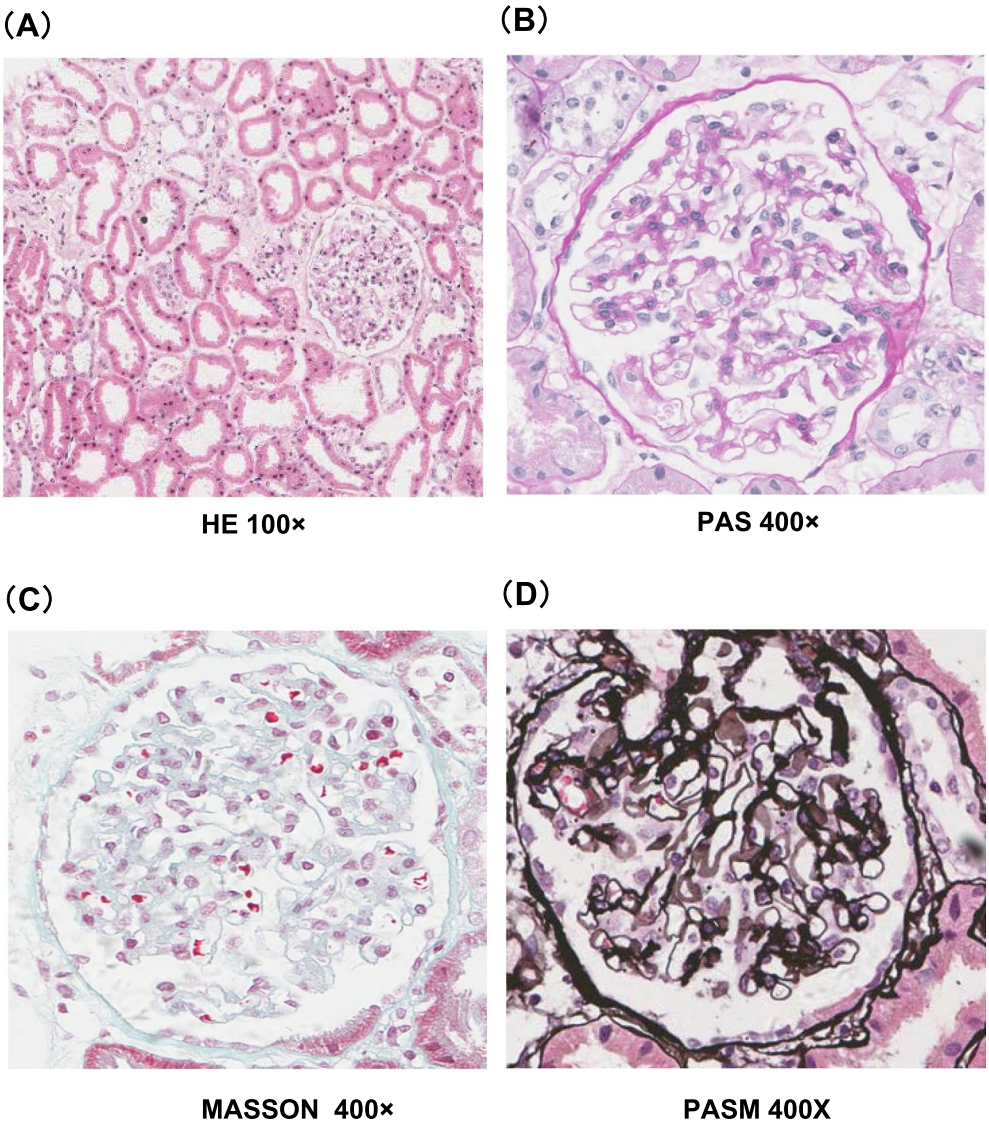

Fig. 2 Kidney biopsy findings. Pathological findings with **A** hematoxylin and eosin, original magnification: 100×. **B** Periodic acid–Schiff, **C** Masson's trichome, and **D** periodic Schiff-methenamine staining. Original magnification: 400×. Pathological findings showed 12 glomeruli, of which one had global sclerosis while the others had minor changes. Segmental mesangial hyperplasia was observed in some glomeruli, with visceral epithelial swelling

undergo nasal mucosal electron microscopy, but the patient refused. The patient had abundant proteinuria, hypoproteinemia, and dyslipidemia. Nephrotic syndrome was diagnosed, secondary nephropathy was ruled out by laboratory examination, and minimal change disease was confirmed by renal biopsy.

Treatment course and clinical follow-up

There is currently no curative therapy for Kartagener syndrome. Thus, the focus is on symptomatic measures such as regular airway cleaning and treatment of recurrent respiratory infections [5]. Because the patient had a respiratory infection on admission, she was treated with respiratory quinolone (levofloxacin). The infection improved after antiinflammatory treatment. To clarify pathology and guide follow-up treatment, renal puncture biopsy was performed. The renal biopsy indicated minimal change disease. Four 24-h urinary protein measurements were performed; the results were 5.02, 2.88, 2.41, and 2.23 g (Fig. 3). Albumin was measured two times, with results of 16.5 and 23.2 g/L (Fig. 4). The patient had a large amount of proteinuria and hypoproteinemia; therefore, steroid hormones should be used. However, considering that the patient

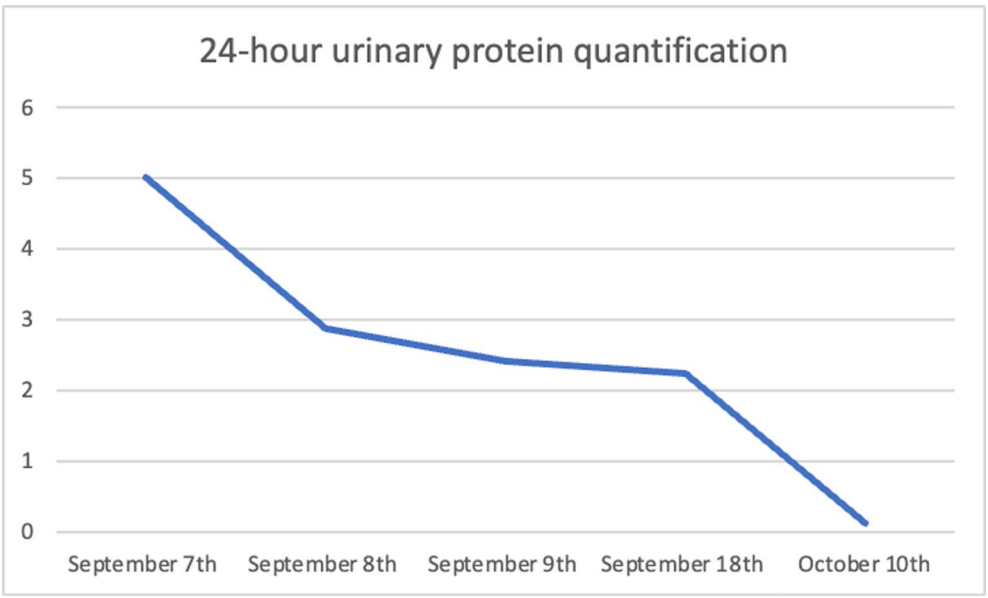

Fig. 3 Quantitative changes of urinary protein. 24 h urinary protein quantification was measured four times during hospitalization; the results were 5.02, 2.88, 2.41, and 2.23 g. Urinary protein quantification during outpatient follow-up was 0.12 g/24 h

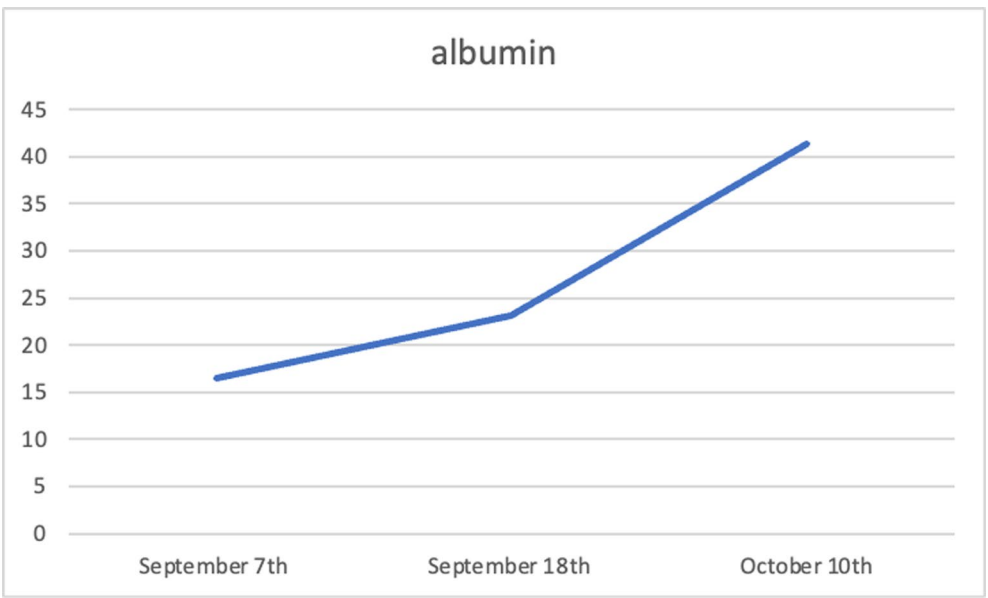

Fig. 4 Albumin change. Albumin levels were measured twice during the hospital stay, with results showing levels of 16.5 and 23.2 g/L. The albumin level at outpatient follow-up was 41.1 g/L

had an infection, the urinary protein quantity decreased after anti-infection treatment, and the serum albumin level increased. This did not exclude the possibility of spontaneous remission; therefore, hormone therapy was not used. The patient was administered telmisartan (50 mg orally once daily) to reduce urinary protein levels. A total of 1 month after discharge, the outpatient follow-up showed that the edema had subsided, albumin was 41.1 g/L, and urinary protein quantity was 0.12 g at 24 h, which was in line with complete remission. The patient was followed up for 1 year, and no drugs were used. There was no recurrence of nephrotic syndrome.

Discussion and conclusion

Human cilia line the upper respiratory tract, lower respiratory tract, mastoid cavity, tympanic sinus, tympanic cavity, eustachian tube, fallopian tube, sperm flagella, and ventricles. The motility of the cilia in these organs promotes fluid flow and has important physiological roles. Defects in the genes encoding ciliary proteins or assembling these proteins result in autosomal recessive genetic diseases such as PCD, characterized by ciliary dysfunction and impaired mucociliary clearance [6]. The clinical manifestations include sinusitis, otitis media, bronchiectasis, and repeated lung infections [7]. The loss of the oscillating ability of the sperm flagella and the failure of fallopian tubes with abnormal cilia to ensure the normal delivery of eggs result in male and female infertility [4]. After 10–15 days of gestation, the abnormal cilia in the embryo are unable to guide the viscera in the right spiral rotation, resulting in random translocation of the viscera. Therefore, it is estimated that approximately 50% of patients with PCD can have visceral translocation [1, 7]. Patients with congenital heart disease, especially those with lateral defects, may also present with PCD [8]. To date, pathogenic variants in more than 40 genes are known to cause PCD [9]. Genetic causes have been identified in approximately 70% of patients with PCD, and mutations in different genes lead to variable phenotypes [9]. Studies have indicated an association between PCD and alterations in *DNAH5* and *DNAI1* genes [7]. The PCD diagnosis depends on the electron microscopic structure of the respiratory cilia obtained by nasal scraping or bronchial brush biopsy. The current diagnostic criteria for the ciliary ultrastructure include the deletion, structural modification, or duplication of the inner dynein arms (IDAs) or outer dynein arms (ODAs). Emerging diagnostic tests include genetic testing, nasal nitric oxide (NO) measurements, immunofluorescence analysis, and high-speed video microscopy to identify subtle ciliary dyskinesia [5, 7]. In recent years, nasal NO measurement has emerged as a noninvasive screening test for PCD. Nasal NO production is significantly reduced (5–20% of normal) in patients with PCD [10].

Kartagener syndrome complicated by kidney disease is rare, with only a few reports. We have reported a rare case of Kartagener syndrome which had the typical triad (chronic sinusitis, bronchiectasis, and total visceral transposition) and was accompanied by minimal disease progression. Kartagener syndrome has been reported to be accompanied by IgA nephropathy [11], focal segmental glomerulosclerosis [12], renal amyloidosis [13], mesangioproliferative glomerulonephritis [14], and chronic renal failure [15]. The involvement of transforming growth factor (TGF-β), Smad protein signaling, and T cell-secreted vascular permeability factor (VPF) may cause Kartagener syndrome with nephrotic syndrome [16, 17].

This is the first report of a rare case of Kartagener syndrome accompanied by minimal change disease progression, which was treated symptomatically. No hormones or immunosuppressants were used. Antiinflammatory agents and angiotensin 2 receptor blocker drugs were effective. The examination of the electron microscopic structure of the respiratory cilia obtained by nasal scrape or bronchial brush biopsy is recommended to further confirm the diagnosis. We believe our findings will provide valuable guidance and reference for the treatment of such cases in the future.

Abbreviations

PCD	Primary ciliary dyskinesia
NSAIDs	Nonsteroidal antiinflammatory drugs
BP	Blood pressure
R	Respiratory rate
P	Pulse
C3	Complement 3
C4	Complement 4
ANCA	Antineutrophil cytoplasmic antibodies
HIV	Human immunodeficiency virus
TPPA	Treponema pallidum particle agglutination
CRP	C-reactive protein
CT	Computed tomography
IgM	Immunoglobulin M
IgA	Immunoglobulin A
IDAs	Inner dynein arms
ODAs	Outer dynein arms
NO	Nitric oxide
TGF-β	Transforming growth factor β
VPF	Vascular permeability factor

Acknowledgements

The authors thanks Yuting Wang for illustration assistance.

Author contributions

QH contributed to data collection and manuscript writing. JL and HZ supervised the data and finalized the manuscript. All authors read and approved the manuscript.

Funding

This research was supported by the Chinese Nature Science Foundation 82170740 (HZ), 82100743 (JL); Applied Basic Research Program of Liaoning Province, 2022JH2/101300048 (HZ); Liao Ning Revitalization Talents Program, XLYC2002081 (HZ); Pandeng Scholar of Liaoning Province, 2013222 (HZ); and Outstanding Scientific Fund of Shengjing Hospital, 202206 (HZ). The funders conceptualized this case report and carried out analysis of patient's clinical course, outcomes, and interpretation of findings, provided critical review comments, and submitted the manuscript.

Availability of data and materials

The datasets used and/or analyzed during the current study are available from the corresponding author on reasonable request.

Declarations

Ethics approval and consent to participate

A human subject research protocol was approved in advance by the Institutional Review Boards of the Affiliated Hospital of China Medical

University. The patient provided written informed consent prior to research participation.

Consent for publication
Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests
The authors declare no conflict of interest.

Received: 14 November 2024 Accepted: 11 March 2025
Published online: 07 April 2025

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