A CASE OF KARTAGENER SYNDROME WITH PULMONARY EMBOLISM

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ABSTRACT

Material: *Kartagener syndrome is a rare autosomal recessive genetic related disease with extremely low incidence and frequent missed diagnosis and misdiagnosis in clinic. Kartagener syndrome complicated with pulmonary embolism is rarer in clinic.*

Method: *A female patient with cough, cough purulent sputum, haemoptysis, bronchiectasis and situs inversus viscerum features by iconography is reported in this study. Meanwhile, Spiral CT pulmonary angiography (CTPA) suggests filling defects in the trunk and branches of the right pulmonary artery, considering the diagnosis of Kartagener syndrome (incomplete type) with pulmonary artery thromboembolism.*

Results: *The patient had pulmonary embolism and haemoptysis due to the bronchiectasis of Kartagener syndrome. After proscribing anti-infection, expectorant, sequential haemostasis and anticoagulant treatment, the patient's symptoms improved and she was discharged.*

Conclusion: *Kartagener syndrome is a rare clinical disease. When imaging suggests the presence of bronchiectasis, situs inversus viscerum, with or without sinusitis, the diagnosis of Kartagener syndrome should be considered. Individualized haemostatic and anticoagulant regimens should be developed for patients with Kartagener syndrome complicated with pulmonary embolism.*

Keywords: *Kartagener syndrome, pulmonary embolism, haemoptysis.*

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Introduction

Kartagener syndrome is a rare autosomal recessive genetic related disease, and is a subtype of primary immotile cilia syndrome. The incidence of the disease is extremely low, approximately 1/20000 to 1/100000[1, 2]. Only sporadic cases are reported in the literature at home and abroad[3, 4], and missed diagnosis and misdiagnosis often occur in clinic. In recent years, with increased awareness among medical staff, the diagnosis of Kartagener syndrome has been much easier. Recently, a patient with Kartagener syndrome was admitted to our department and was found to be complicated with pulmonary embolism during hospitalisation, which is rarer in clinical practice. The case is reported as follows.

Case presentation

The 47-year-old female patient, a farmer, was admitted to our department on 25 October, 2018 due to "repeatedly coughing purulent sputum for more than 40 years, intermittent haemoptysis for more than 10 years and aggravation for 5 days". The patient began to cough and cough purulent sputum when she was young, more than 40 years ago. After being treated with anti-infection, expectorant and other treatment, the cough was alleviated and purulent sputum was reduced, but the cough and cough purulent sputum appeared repeatedly.

More than 10 years ago, the patient began to appear haemoptysis, and this was clinically diagnosed as "tuberculosis" in the local centres for disease con-

trol (CDC). The patient was given HREZ quadruple anti-tuberculosis treatment for approximately 1 year. Subsequently, the patient still coughed repeatedly, coughed purulent sputum and intermittent haemoptysis, all of which can be alleviated after anti-infection, haemostasis and other treatment.

During catabasis, the patient can tolerate daily activities and light physical exertion, only having breathing difficulties during strenuous activity. Five days ago, the patient appeared with cough aggravation again, purulent sputum increased to about 50 ml per day, no smell of pus, and accompanied by haemoptysis, with bright red blood, approximately 50 to 100 ml per day; in addition, the patient felt asthmatic and tired when she walked on a level road.

The patient was admitted to hospital with chest CT (Figure 1-2), suggesting that "the right lung was damaged, the left lower lobe had bronchiectasis, and total organ reverse position in the thoracoabdominal cavity". The patient was given haemostatic treatment such as haemocoagulase, but she continued to haemoptysis and was admitted to our hospital. The patient had no history of chronic sinusitis and is married with one son and one daughter.

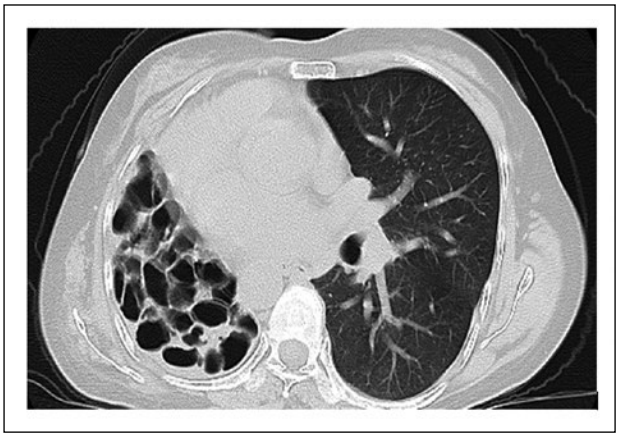

Figure 1: Chest CT - right lung lesion.

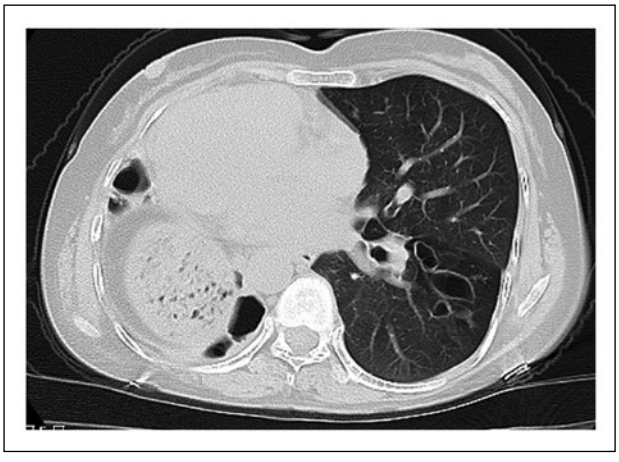

Figure 2: Chest CT - bronchiectasis in the lower left lung.

Body examination at admission

Body temperature was 36.6°C, pulse was 98 times/min, respiration was 26 times/min and blood pressure was 118/68 mmHg, with normal development, wasting appearance, clear mind, poor spirit, no congestion, oedema of bulbar conjunctiva, no cyanosis of lip, no irritation of jugular vein, negative sign of hepatic neck reflux, right deviation of trachea, right collapse of chest, no three concave signs, right respiratory movement and voice tremor weakened. Right lung percussion presented dullness, left lung percussion presented a hyper resonant note, on the right side could be heard a tubular respiratory sound, and in the right lung and left lower lung could be heard a bubble. The apical pulsation was located at 0.5 cm in the middle line of the right clavicle of the fifth intercostal area, with a heart rate of 98 beats/min, with regular heart rhythm. The patient had no concave oedema in both lower extremities.

According to the history and chest CT, the initial diagnosis of admission was:

- Bronchiectasis with haemoptysis;
- Right lung lesion;
- Total visceral translocation.

Auxiliary examination after admission:

- Blood gas analysis (FIO_2 21%): PH 7.50, pO_2 68 mmHg, pCO_2 36 mmHg, SaO_2 95%, HCO_3- 28.1 mmol/L.
- Blood routine: white blood cell count 8.55×109/L, N% 66.3%, Hb 100 g/L, PLT 244×109/L, CRP 13.88 mg/L.
- Coagulation function: prothrombin time 11.3 s, prothrombin percentage activity 118.4%, international standardised ratio 0.97, D-dimer 98 ug/ml, procalcitonin <0.01 ng/ml. The liver function, kidney function, electrolyte, fasting blood glucose, and myocardial enzyme spectrum were not significantly abnormal.
- Sputum culture: Citrobacter freundii, piperacillin, levofloxacin, amikacin, imipenem sensitive. The patient had significantly elevated D-dimer.
- Spiral CT pulmonary angiography (CTPA, Figure 3-4): right pulmonary artery trunk and branch filling defect, considering emboli; left pulmonary artery trunk and its main branch vessels were naturally shaped, no obvious stenosis and filling defect. The right lung had blade-like high-density shadow, with multiple cystic light and shadow inside, considering the right lung damage. The patient had total situs inversus viscerum.

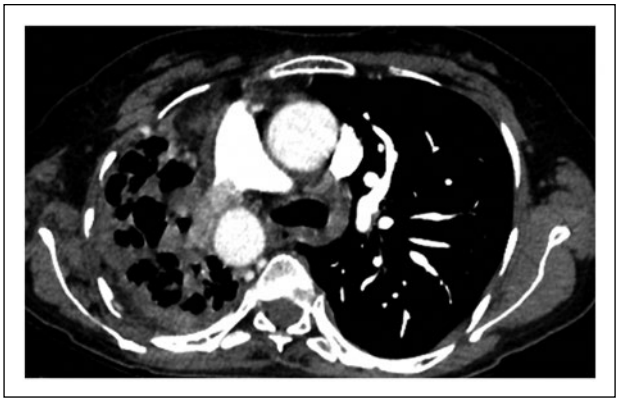

Figure 3: Filling defects in the main trunk and branches of the right pulmonary artery.

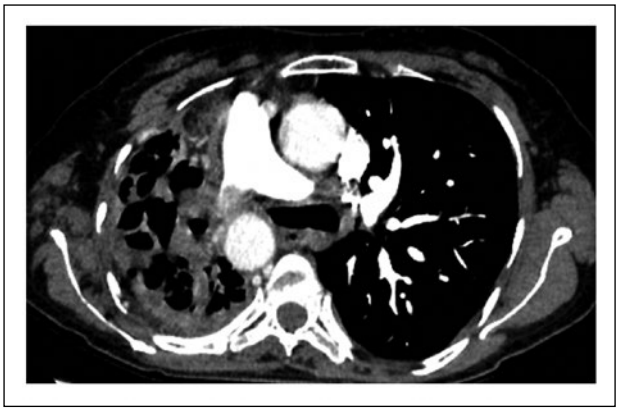

Figure 4: Filling defects in the main trunk and branches of the right pulmonary artery.

• Echocardiography (Figure 5): the heart was located in the right thoracic cavity. Left ventricular posterior wall beat amplitude decreased, aortic valve, mitral valve, tricuspid valve slightly closed incomplete, PASP 25 mmHg, pulmonary artery trunk diameter 19 mm.

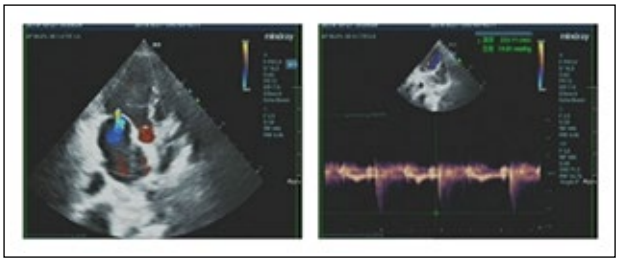

Figure 5: Colour Doppler echocardiography - the heart is located in the right chest cavity.

• Paranasal sinus CT (Figure 6-7): no obvious abnormality.

At this point, combined with the patient's history, chest and sinus CT and CTPA, the diagnosis was considered to be:

• Kartagener syndrome (incomplete type);

• Pulmonary artery thromboembolism.

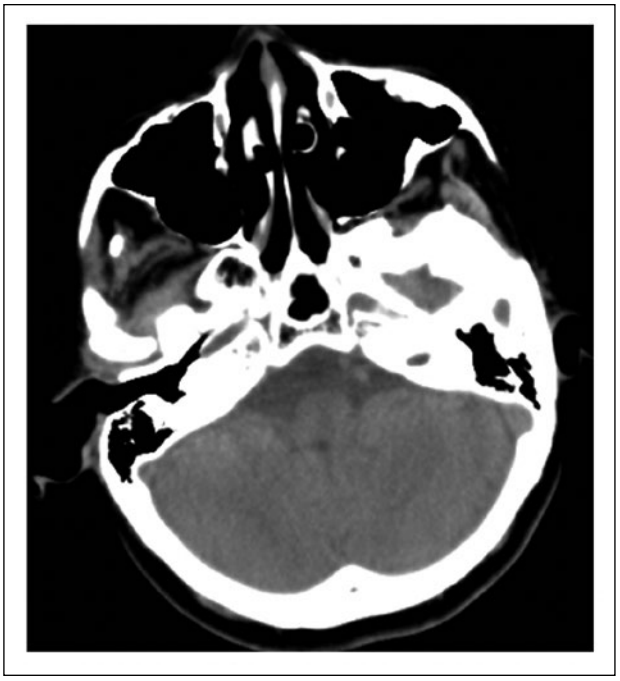

Figure 6: Paranasal sinus CT1- no obvious abnormality.

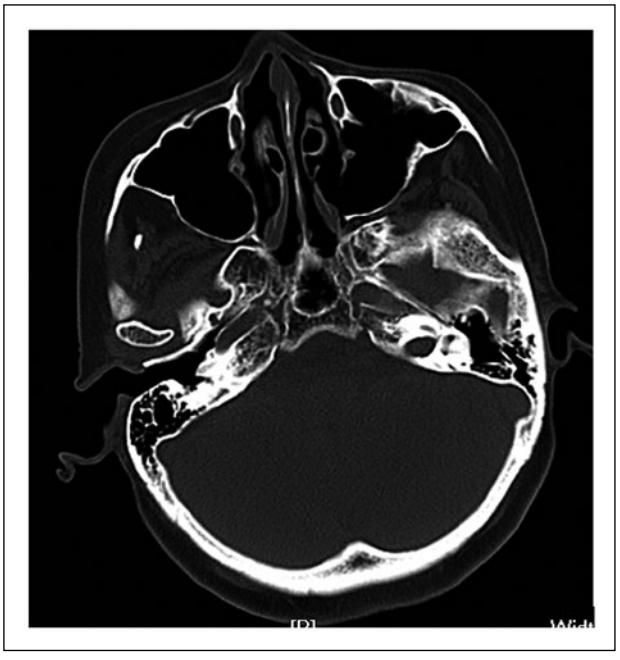

Figure 7: Paranasal sinus CT2- no obvious abnormality.

Treatment

The patient had increased cough and purulent sputum, haemoptysis and pulmonary embolism also existed. The patient was treated with piperacillin sulbactam for anti-infection, bromhexine for expectorant, pituitrin for haemostasis (day 1-4), subcutaneous injection of low molecular heparin for anticoagulation (day 5-14). Haemoptysis did not recur on the 4th day after admission, cough, dyspnoea gradually alleviated, purulent sputum decreased, and the patient was improved and discharged 2 weeks after ad-

mission. The patient was followed up until the press release and was discharged from hospital for more than 3 months. The patient had no cough and dyspnoea aggravated, and still had yellow sputum, 20-40 ml daily, and occasionally had a small amount of blood sputum which could stop on its own.

Discussion

Kartagener syndrome is a rare autosomal recessive genetic-related disease, a subtype of primary ciliated immobile syndrome with a very low incidence of approximately 1/20000 to 1/100000[1, 2]. Sporadic cases have been reported in the literature at home and abroad[3, 4].

Clinically, the main manifestations of Kartagener's syndrome are bronchiectasis, chronic sinusitis (nasal polyps) and situs inversus viscerum[5]. In this case, the patient had only bronchiectasis and situs inversus viscerum and no chronic sinusitis manifestations, known as incomplete Kartagener syndrome. Kartagener's syndrome may present with multiple systemic symptoms, but respiratory symptoms are the most common cause[6]. The patient in this study had recurrent cough, cough purulent sputum and intermittent haemoptysis, usually diagnosed as bronchiectasis, a common and frequent disease by primary hospitals. The patient is even treated according to pulmonary tuberculosis during the long-term diagnosis and treatment, which is a clinically common misdiagnosis and missed diagnosis, although very few cases of Kartagener syndrome may also be complicated with pulmonary tuberculosis[7]. Therefore, the possibility of Kartagener syndrome should be considered when the patient has clinical and imaging manifestations of bronchiectasis, accompanied by visceral contraposition.

Pulmonary embolism is a common disease relative to Kaltagener's syndrome[8]. The typical pulmonary embolism can manifest as chest pain, haemoptysis and dyspnoea triple syndrome[9]; however, patients with pulmonary embolism usually do not have the above three symptoms simultaneously, and none of the above symptoms are specific. Therefore, pulmonary embolism is also often missed and misdiagnosed[10]. In recent years, with the improvement of clinicians' consciousness and the application of CTPA, the diagnostic rate of pulmonary embolism has increased markedly, and the mortality of acute pulmonary embolism has also decreased significantly[11, 12]. In this case, the patient had haemoptysis, dyspnoea, D-2 polymer significantly increased by screening, the right pulmonary artery trunk and branch filling defect detected by CTPA; the diagnosis of pulmonary embolism is clear.

The diagnosis of Kartagener syndrome complicated with pulmonary embolism is not particularly difficult, but the treatment is contradictory and controversial. At present, there is no treatment to promote the recovery of ciliary motor function, and there is no radical cure for Kaltagener's syndrome. Although it is reported that lung transplantation is the only effective treatment[13], the clinical development is influenced by many factors, and the long-term prognosis requires further observation. In clinic, the main treatment is removal of the inducement, symptomatic treatment, improvement of symptoms, and delay of the treatment progress[14]. The patient's cough, cough purulent sputum and haemoptysis may be considered a result of Kartagener's syndrome and can be given anti-infection, expectorant and haemostasis treatment. However, elevated D-dimer and CTPA confirmed the presence of pulmonary artery thrombosis, which is located in the main trunk of the right pulmonary artery, so anticoagulation and even thrombolytic therapy seems to be imperative. The current guidelines for the diagnosis and treatment of pulmonary embolism are mainly directed towards the recommendations of diagnosis and treatment of acute pulmonary embolism[15]. There was damage to the right lung structure in this case, no high risk factors such as deep venous thrombosis in the lower extremity, no hemodynamic changes, severe hypoxia, right cardiac insufficiency and myocardial enzymatic changes, and the clinical manifestations do not seem to conform to the changes of acute pulmonary embolism. We speculated that the right pulmonary artery thrombosis was related to the destruction of the right lung structure, the endothelial injury of the pulmonary artery, and the stasis of the blood flow in this patient for a long time.

The current guidelines do not recommend how to determine if there is an acute pulmonary embolism, whereas there are no guidelines for the treatment of pulmonary embolism with changes in lung structure (or on the basis of changes in lung structure). The patient's right lung structure was completely damaged and there was no ventilation function in normal lung tissue. Does the right pulmonary artery embolisation require regular anticoagulant therapy, in the case of bronchiectasis with haemoptysis, and can the patient benefit from anticoagulant therapy? This is something we need to consider. In the course of the patient's treatment, we also developed

individualised treatment measures for the patient, adjusting the treatment plan to the changes in the patient's condition. The patient had haemoptysis on admission, so we chose pituitrin to avoid haemostatic drugs such as hemocoagulase, which may increase the risk of thrombosis. Later, haemoptysis of the patient was stopped, and subcutaneous injection of low molecular weight heparin was performed for low level anticoagulation, following which the patient's cough, dyspnoea and other symptoms were alleviated. There was no conclusive evidence that long-term oral anticoagulants were beneficial to the patient, and the patient's follow-up compliance was poor, so no oral anticoagulants were given.

In conclusion, Kartagener's syndrome is a rare clinical condition. When imaging suggests the presence of bronchiectasis, situs inversus viscerum, with or without nasosinusitis, the diagnosis of Kartagener syndrome should be considered. The particular feature of this case is that the patient has bronchiectasis and haemoptysis, while lung destruction is complicated with pulmonary embolism. There are contradictions in the treatment and no ready-made guidelines for reference; therefore the patient's later conditions and treatment require continued follow-up tracking.

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