CLINICAL QUIZ (p125) ANSWER

According to the patient's medical history, symptoms, signs, radiology examination, and test results, he was diagnosed with Kartagener syndrome (KS) and treated with antibiotics, an expectorant, and inhalers for asthma. When his symptoms improved significantly, we encouraged him to improve his fitness, improve his nutrition, and avoid infection. We have followed him for 6 months, and no recurrence of KS has been observed.

## What is the Kartagener syndrome?

In 1933, Kartagener first reported a 'sinusitis-bronchiectasis-situs inversus syndrome', now commonly known as one of the subtypes of the primary ciliary dyskinesia (PCD). PCD is an autosomal recessive disease caused by structural defects of the cilia and may also be associated with the retinitis pigmentosa GTPase regulator (RPGR) gene.[1] When situs inversus is associated with PCD, it is referred to as KS. Recent studies have indicated that KS-associated genes are located on chromosomes 16p12.1-12.2 and 15q13.1-15.1.[2]

These mutations lead to defects in the ciliary dynein arms that in turn lead to the absence of ciliary motility. When the cilia become dyskinetic, their coordinated, propulsive action is diminished and bacterial clearance is impaired. This lack of ciliary function is the basis of the KS phenotype and manifests as recurrent upper and lower respiratory tract infections, including sinusitis, otitis media, and bronchiectasis, as in the patient described. Furthermore, because of immotile spermatozoa, male patients with this syndrome are almost invariably infertile. Due to disorders of early embryonic cilial movement, the internal organs cannot complete translocation and situs inversus results.

## What is the early diagnosis way of the Kartagener syndrome?

The lung function of some KS patients is significantly impaired in childhood,[3] which makes it vital for clinicians to have a high index of suspicion of KS for children with repeated upper and lower respiratory tract infections during childhood and adolescence. The early diagnosis of KS is enhanced if appropriate examination and pulmonary function tests are performed. The concentration of exhaled NO can be an objective index for KS screening.[4]

Previously, confirmation of the diagnosis of KS was achieved by electron microscopic examination (EM) of cells from bronchial mucosal biopsy or assessment of ciliary function under a light microscope.[5] Jorissen et al reported that up to 28% of patients who have clinical manifestations of KS and cilial gene mutations have normal cilial ultrastructure on EM.[6] Thus, structural defects inside the cilia on EM can support a diagnosis of KS, but a normal result does not exclude KS.

## How to cure Kartagener syndrome?

There is no effective specific treatment for KS. Therapies for KS include antibiotics, relieving cough, eliminating phlegm, treating asthma, and enhancing immunity. Usually the prognosis is good when KS is diagnosed and treated early. That long-term,low-dose clarithromyein has been shown to have a good response to primary ciliary dyskinesia.[7] Influenza and pneumococcal vaccination should be utilised to prevent respiratory tract infections in children. For bronchiectasis associated with KS, symptomatic treatment is a mainstay, and lobectomy is usually not required.[5] Functional endonasal sinus surgery can relieve the pulmonary infection and sinusitis.[8] Hearing loss typically decreases over time, so does otitis media.[9] With the discovery of the pathogenic genes and mutation sites of KS, there is now hope for future gene therapy for KS. As a congenital autosomal recessive disease, the treatment of KS through in situ remediation of a genetic defect or insertion of functional genes still needs to be studied.

## Revelation

In summary, this case serves to remind clinicians that a high index of suspicion should be maintained for the diagnosis of KS in patients who present with recurrent upper and lower respiratory tract infections, sinusitis, or bronchiectasis. Early diagnosis and treatment of the disease can effectively delay structural changes in the lungs of KS patients, avoid the occurrence of pulmonary heart disease and pulmonary abscess, and greatly improve the prognosis of patients.

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