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Diagnosis of cystic fibrosis in an adult Japanese male

Yoshitaka Tomoda MD, PhD[1] | Sayuri Arai MD[2] | Kentaro Kawaguchi MD[2] | Shinji Nabeshima MD[2] | Takeshi Orihashi MD[2] | Yasuyuki Kihara MD, PhD[2] | Ryoji Kouzuma MD, PhD[2] | Kazutoyo Tanaka MD[1]

[1]Department of General Medicine, Saiseikai Fukuoka General Hospital, Fukuoka, Japan

[2]Department of General Internal Medicine, Kitakyushu General Hospital, Fukuoka, Japan

Correspondence
Yoshitaka Tomoda, Department of General Medicine, Saiseikai Fukuoka General Hospital, Fukuoka, Japan.
Email: yoshisoph@gmail.com

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Case: A 23-year-old Japanese male with a history of congenital bronchiectasis and chronic respiratory failure was transferred to our hospital following elevation in fever and an exacerbation of respiratory failure. The patient had been frequently hospitalized due to pneumonia and ileus since he was 7 months old. At the age of 10 years, cystic fibrosis (CF) was suspected, but could not be confirmed. He is the first child of non-consanguineous parents and his parents had no respiratory symptoms.

His chest *X*-ray showed consolidation in the right lower lung field and an annular shadow in bilateral lung fields (Figure 1). A chest computed tomography scan revealed right-sided middle and lower lobe consolidation, bilateral hyperinflation, and multiple cystic bronchiectasis with fluid collection (Figure 2). Sputum culture grew *Streptococcus pneumoniae*, and although antibiotics were administered, his symptoms persisted. As the specific medical history and imaging findings strongly suggested CF, we analyzed sweat chloride concentration and found it to be elevated at 88 mEq/L (normal range <50 mEq/L). Thus, a definitive diagnosis of CF was established. However, no mutations were observed in the cystic fibrosis transmembrane conductance regulator (*CFTR*) gene in the patient or his parents. He is currently on a waiting list for lung transplantation.

Cystic fibrosis is the most common autosomal recessive genetic disorder among Caucasians, but it is quite rare in Southeast Asia, including Japan, where the estimated incidence is 1 in 350 000.[1] Thus, physicians in Japan are unfamiliar with the clinical characteristics of CF, which may have led to the 23-year delay in establishing a CF diagnosis in this case.

Cystic fibrosis is caused by mutations in the *CFTR* gene that lead to clinical manifestations such as pulmonary disease, meconium ileum, pancreatic insufficiency, and elevated chloride concentrations in sweat.[2] In this case, mutations in the *CFTR* gene were not detected. Reportedly, the profiles of *CFTR* gene mutations in the Japanese population are different from those in the Caucasian population, and a majority of *CFTR* mutations are of rare types.[3] Hence, in a Japanese CF case, *CFTR* mutations may be undetected by conventional analysis.

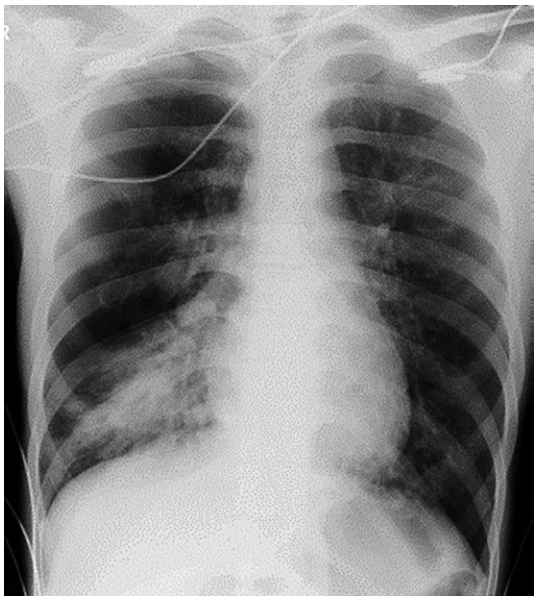

FIGURE 1 Chest *X*-ray image of a 23-year-old man. Image shows consolidation in the right lower lung field and an annular shadow in bilateral lung fields

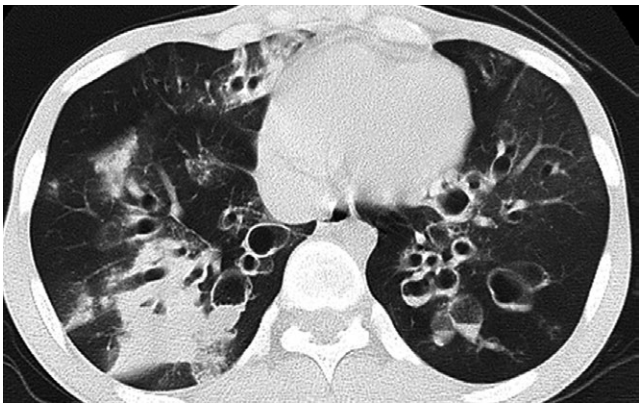

FIGURE 2 Chest computed tomography image of a 23-year-old man. Image shows right-sided middle and lower lobe consolidation and multiple cystic bronchiectasis

A typical form of CF can be diagnosed if a patient exhibits specific symptoms, including meconium ileus, respiratory symptoms, failure to thrive and pancreatic insufficiency, and elevated levels of chloride in sweat.[4] Our case fulfills the clinical criterion for CF.

Reportedly, primary ciliary dyskinesia (PCD), an inherited disorder of the function of motile cilia, shares several features with CF, including progressive bronchiectasis and decline in lung function.[5] Our case can be distinguished from PCD by clinical manifestation, lack of otitis and situs inversus, a history of ileus, and elevated levels of chloride in sweat.

In case of bronchiectasis, various differential diagnoses including PCD, bronchial asthma, allergic bronchopulmonary aspergillosis, diffuse panbronchiolitis, mycobacterium infection, viral infection, and sarcoidosis should be considered, but a detailed and specific medical history could provide information that leads to a definitive diagnosis.

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CONFLICT OF INTERESTS

The authors have stated explicitly that there are no conflicts of interest in connection with this article.

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