



Cystic Fibrosis in a Case Report

Li Z*, Shen W and Fang X

Department of Pediatrics, The First People's Hospital of Jiashan, China

Case Presentation

A 13-year-old male teenager presented to our emergency department in May 2018. He complained of fainting once and had a mild cough for 3 days and low-grade fever (38.0C) prior to admission. He had history of frequent respiratory infections and no chronic diarrhea.

He was a second-born to non-consanguineous healthy parents, born full term after an unremarkable pregnancy. His birth weight was 2.8 kg, and meconium was passed on the first day of life. No history of CF or other chronic pulmonary disease was found in his family. There are no known intermarriages with Caucasians in either family.

On physical examination, he was undernourished with 30 kg body weight and 125 cm height. He had a BMI of 17.5, which was below the 3rd percentile for age. On auscultation, diffuse fine crackles were present throughout both lung fields. Heat and abdomen were normal. Clubbing of the fingers was present. Laboratory examination showed white blood cells $106 \times 10^9/L$, C reactive protein 11.33 mg/l, other tests including CBC, serum electrolytes, urine, and isolation of *P. aeruginosa* in sputum culture. Chest radiograph (Figure 1): Bi-pneumonia. A Computed Tomography (CT) scan (Figure 2) revealed right upper vesicles, bilateral bronchiectasis with obvious exudative lesions pulmonary fibrosis. A head CT (Figure 3) scan revealed Para sinusitis bilaterally.

Genetic testing revealed that the patient was a compound heterozygote of mutations: CFTR; NM_000492; c.3068T>G; p.Ile1023Arg, p.I1023R; EX19; CD19. Pathogenic was inherited from the mother. The mutation of c.3068T>G had been reported.

Outcome and Follow-Up

His symptoms improved after antibiotics and expectorant treatment. After being discharged from our hospital in May 2018. The patient was re-hospitalized nine times for pulmonary infections and hemoptysis (once) in the last their year. He frequents respiratory infections, with isolation of *P. aeruginosa* and oral azithromycin prophylaxis.

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*Correspondence:

Zhongjie Li, Department of Pediatrics,
The First People's Hospital of
Jiashan, No. 1218, Tayo South Road,
Jiashan, Zhejiang 314100, China, Tel:
13305838962;

E-mail: lizhongjie52@dingtalk.com

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Discussion

Cystic Fibrosis (CF), resulting from Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutations, is one of the most common autosomal recessive disorders in Caucasians



Figure 1: Chest radiograph: Pneumonia.

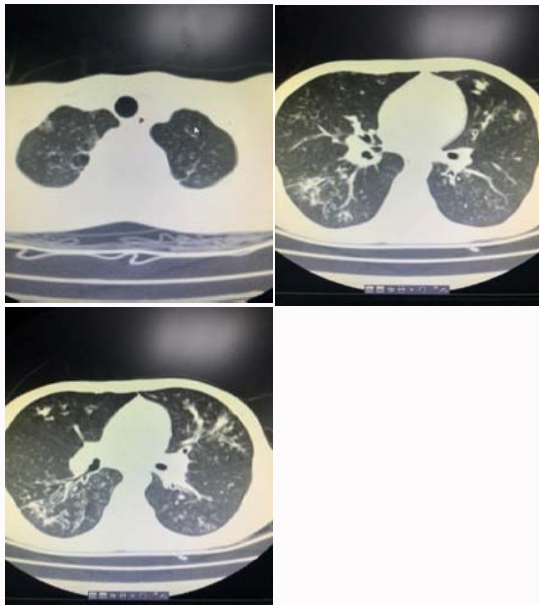


Figure 2: Revealed right upper vesicles, bilateral bronchiectasis with obvious exudative lesions pulmonary fibrosis.

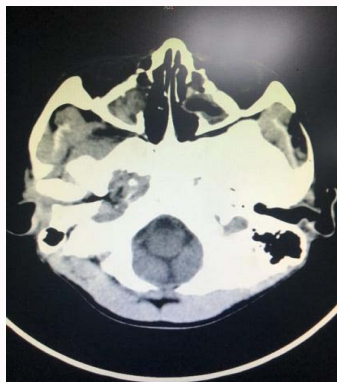


Figure 3: Revealed pansinusitis bilaterally.

[1]. CF has seldom been reported in Chinese individuals. The patient had history of frequent respiratory infections, each chest X-ray: Pneumonia. Clubbed finger was found in physical examination, further CT examination indicates bronchiectasis, Genetic testing

revealed CFTR; NM_000492; c.3068T>G; p.Ile1023Arg, p.I1023R geographic region. The p.I1023R is indeed a founder mutation in southern Han Chinese (Another Chinese-specific mutation, p.G970D, appears to be more commonly observed in northern Chinese with CF) [2]. A patient with the p.I1023R mutation exhibited had recurrent pneumonia and bronchiectasis. *Pseudomonas aeruginosa* was isolated from the patient. Chinese CF patients have a different CFTR mutation spectrum, that will result in a low diagnostic yield when applied to Chinese patients. Carry out sweat chloride test. It has been only implemented some hospital in Beijing or Shanghai. In addition, The Society of pediatrics, Chinese Medical Association, can formulate CF guidelines for Chinese pediatricians. with the development of gene detection technology, the cost of CFTR gene mutation testing will be cheaper. We are confident that diagnosis of CF will be more detected in China.

As the spectrum of clinical presentation is very variable, important for clinicians from multiple specialties to be vigilant and suspect this diagnosis in condition such as recurrent pulmonary infection, male infertility, pancreatitis, nasal polyposis, and malabsorption even in patients [3-5]. It is necessary to educate Chinese clinicians about the signs, symptoms, and diagnosis of cystic fibrosis.

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