# Pseudo-Bartter Syndrome in a Chinese Infant with Cystic Fibrosis Caused by c.532G>A Mutation in *CFTR*

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To the Editor: We present a Chinese case of cystic fibrosis (CF) with Pseudo-Bartter syndrome (PBS) in a 6-month-old female infant. She started to have recurrent cough and wheezing after "a common cold" at 3-month-old and had been hospitalized several times for "pneumonia". She received oral antibiotics and nebulized bronchodilators which resulted in no improvement. In addition, she developed hypokalemia (2.5 mmol/L) and metabolic alkalosis. She was transferred to our hospital for ongoing care. She had an uncomplicated birth history. She was the fourth child born to consanguineous parents. Her elder brother died of severe pneumonia and bronchiectasis at 13 years of age. Her elder sister was diagnosed as asthma at 6 years old which is well controlled. Physical examination showed moderate dehydration, failure to thrive (3rd percentile), and diffuse rhonchi on auscultation. A computed tomography (CT) scans illustrated scattered infiltrates and mild consolidation in the right lung [Figure 1a]. Six months later, a repeat CT scan demonstrated mild bilateral bronchiectasis [Figure 1b]. Genetic analysis revealed she had homozygous mutation c.532G>A (exon 5) in *CFTR*. His mutation was also found in both of her parents. There were no evidences of congenital pulmonary/heart disorders, tuberculosis, gastroesophageal reflux, primary immunodeficiency diseases, primary ciliary dyskinesia, and Bartter syndrome. Finally, she was diagnosed as CF with PBS. With antibiotics, fluid therapy, respiratory supportive treatment, and nutrition supplement, she improved significantly. CF is the most frequent monogenic disease in Caucasians but is rare in Asian. Few patients with CF can have hyponatremia, hypokalemia, and metabolic alkalosis that mimic Bartter syndrome, which called PBS.[1,2] PBS as a rare phenotype of CF with the prevalence of 16.8%. Indeed, it can be an initial presentation of CF, especially in infants. PBS is well recognized and has been reported in the literature in patents of Caucasian origin. It is frequently missed as simple dehydration or Bartter syndrome. It is reported that about 30% of patients present initially in the nephrology clinics and are diagnosed as Bartter syndrome.[3] In China mainland, just 53 cases with CF have been reported from 1974 till now.[4,5] Moreover, the main phenotypic characteristics of these cases were bronchiectasis, sinusitis, and recurrent pneumonia. The phenotypic features differed that described in Caucasian patients.[4,5] The final diagnosis is often delayed in China because of lack of newborn screening, atypical phenotype, and low frequency in the population. In addition, PBS has rarely been reported in Chinese origin. Therefore, the patients with atypical features of CF are likely to be greatly underdiagnosed in China. This case expands the phenotypic spectrum of CF patients in Chinese origin. Pediatricians should keep CF with PBS in the differential diagnosis of infants presenting with recurrent pulmonary infection, failure to thrive and electrolytes disturbance, especially in those with dehydration, hypokalemia, and metabolic alkalosis who are usually suspected to have Bartter syndrome. With the development of molecular diagnostic technique, CF is not as infrequent as once thought in the Chinese population.

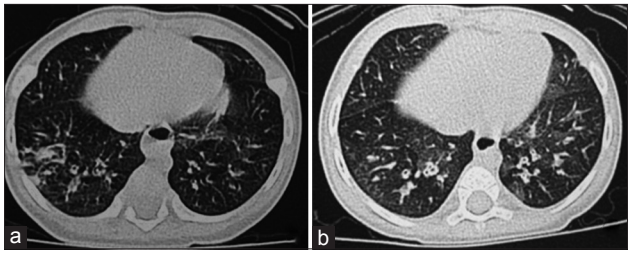


**Figure 1:** Manifestations of chest computed tomography scans. (a) Computed tomography scans on admission showing the scattered patchy lesion and mild consolidation in the right lung; (b) computed tomography scans 6 months later after discharge showing bilateral bronchial wall thickening, mild multiple bronchiectasis.

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## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient's parents have given consent for images and other clinical information to be reported in the journal. The patient's parents understand that name and initial will not be published and due efforts will be made to conceal identity; however, anonymity cannot be guaranteed.

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## Conflicts of interest

There are no conflicts of interest.

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