

# Meconium thorax observed in a Chinese neonate with cystic fibrosis

Yanzhen Wan<sup>1</sup>, Xiangmao Bu<sup>1</sup>, and Peng Sun<sup>1</sup>

<sup>1</sup>Qingdao Women and Children's Hospital

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## Abstract

CF is considered a rare disease among the non-Caucasian population. The clinical phenotypes and genotypic spectrum of Chinese CF patients are reported to be different from that observed in Caucasians[(1)](#ref-0001). Our patient carried compound heterozygous mutations which are not included in the Caucasian CFTR common mutation-screening panel and have not been reported yet. CFTR dysfunction causes a spectrum of diseases, with a range in the number of organs involved and varying disease severity. Typical phenotypic features of CF include respiratory diseases (bronchiectasis with persistent airway-based infection and inflammation), gastrointestinal diseases (meconium ileus (MI)), hepatobiliary manifestations (pancreatic insufficiency), and male infertility[(2)](#ref-0002). Our patient presented severe manifestations, namely colonic perforation, meconium thorax, severe pneumonia, diaphragmatic defects-caused marked diaphragm elevation and respiratory failure. It's difficult to ascertain whether the diaphragmatic defects in our patient is related to CF. MI is often the first manifestation of CF and occurs in approximately 20% of CF patients. CF should therefore be high in the differential diagnosis of any infant presenting with MI[(3)](#ref-0003).

## Meconium thorax observed in a Chinese neonate with cystic fibrosis

To the Editor,

A full-term male neonate was delivered by emergency cesarean section after the discovery of bilateral pleural effusion at 40 weeks of gestation. Clinical examination revealed general edema, abdominal distension and liver palpated 4cm under the rib. Radiographic and ultrasonographic results showed bilateral pleural effusion and ascites without calcification. Thoracentesis was performed on day 1 of life. Meconium-like turbid and sticky fluid (Figure 1D) was drained from the pleural cavity. Cytologic examination showed a large amount of white blood cells. In addition, squamous epithelial cells (Figure 1A, 1C), heme crystal (Figure 1B) and old red blood cells were also observed, indicating the possible existence of perforation of the gut and diaphragmatic defect. The baby underwent an exploratory laparotomy on day 2. Perforation of the sigmoid colon was identified and defunctioning ileostomy was performed. However, owing to the liver enlargement, no obvious diaphragmatic defects were observed. Postlaparotomy period was basically uneventful. He was discharged in a good general condition 23 days after birth. Nevertheless, on day 33 of life, the baby was re-admitted presenting with difficulty in breathing and poor response. Radiographs revealed severe pneumonia and marked elevation of bilateral diaphragm, so left and right diaphragmatic exploration by thoracoscope was performed successively on day 49 and day 55. A posterolateral hernial sac (defect area: 3×3 cm) was identified in the left diaphragm and most of the right diaphragm was missing (defect area: 5×4 cm). So, the adhesions were divided; pyothorax was cleared; the herniated organs were repositioned into the abdomen and the diaphragm defects were repaired. After the operation, the baby became clinically stable and was discharged home on day 73 of life. Whole exome sequencing revealed that the baby carried two heterozygous mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. One is c.1210-11T[?]G located in intron 9 (paternal, pathogenic) and the other is c.283 A[?]G (p.L95E) located in exon 4 (maternal, possibly pathogenic). Considering his manifestations, the baby was finally diagnosed with cystic fibrosis (CF).

CF is considered a rare disease among the non-Caucasian population. The clinical phenotypes and genotypic spectrum of Chinese CF patients are reported to be different from that observed in Caucasians(1). Our patient carried compound heterozygous mutations which are not included in the Caucasian CFTR common mutation-screening panel and have not been reported yet. CFTR dysfunction causes a spectrum of diseases, with a range in the number of organs involved and varying disease severity. Typical phenotypic features of CF include respiratory diseases (bronchiectasis with persistent airway-based infection and inflammation), gastrointestinal diseases (meconium ileus (MI)), hepatobiliary manifestations (pancreatic insufficiency), and male infertility(2). Our patient presented severe manifestations, namely colonic perforation, meconium thorax, severe pneumonia, diaphragmatic defects-caused marked diaphragm elevation and respiratory failure. It's difficult to ascertain whether the diaphragmatic defects in our patient is related to CF. MI is often the first manifestation of CF and occurs in approximately 20% of CF patients. CF should therefore be high in the differential diagnosis of any infant presenting with MI(3).

### Figure legend

Figure 1: Pleural fluid drained from the left pleural cavity and its cytologic examination (x100 objective; Wright–Giemsa stain). A and C, Squamous epithelial cells. B, Heme crystal. D, the drained pleural fluid.

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### Author names, affiliations:

Xiangmao Bu, Peng Sun, Yanzhen Wan #

Clinical laboratory, Qingdao Women and Children's Hospital, Qingdao Women and Children's Hospital Affiliated to Qingdao University, Qingdao 266034, China

# Correspondence to: Yanzhen Wan, Clinical laboratory, Qingdao Women and Children's Hospital, Qingdao Women and Children's Hospital Affiliated to Qingdao University, Qingdao 266034, China. E-mail: [wanyanzhen1979@163.com](mailto:wanyanzhen1979@163.com)

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