

## Cystic Fibrosis with a Unusual Presentation in Monozygotic Twins

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### ABSTRACT

Cystic fibrosis is an inheritant autosomal recessive disease. It is associated with mutations in Cystic Fibrosis Trans Regulator gene (CFTR) and has different presentations. We report two 2 month old female patients, products of a twin delivery presented with anemia, edema, hypoalbuminemia and pneumonia. After some work ups, diagnosis of cystic fibrosis was confirmed. This is an uncommon and interesting presentation of cystic fibrosis, which occurred simultaneously in one twin.

**Keywords:** Cystic fibrosis, Monozygotic Twins, Autosomal recessive

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### INTRODUCTION

Cystic fibrosis (CF) is an autosomal recessive disease. The responsible gene is located on chromosome 7. There are multiple mutations; the most common one is delta F 508. CF has variable presentations including chronic lung disease, exocrine pancreatic insufficiency, nasal polyposis, pansinusitis, rectal prolapse, pancreatitis, cholelithiasis and insulin dependent diabetes mellitus. It also causes salt depletion. Neonates may present with cholestasis, meconium ileus and meconium plug syndrome and later in life with cirrhosis or other hepatic dysfunction. The other presentation mostly in infant is CF syndrome with

triad of hypoalbuminemia, edema, and anemia. This syndrome rarely occurs simultaneously in monozygotic twins. We report one twins admitted in pediatric GI ward with CF syndrome, lung disease and mild liver involvement at the same time.

### CASE REPORT

These are two monozygotic full term female twins, who were fed with breast feeding and formula. They were referred with chief complaint of cough and respiratory distress. They had good appetite and correct feeding, but no significant weight gain after two months. Birth weight, height and head circumferences for twin 1 were 3100 g, 48 cm, and 33.2 cm, respectively, and for twin 2 were 3000 g, 49cm and 33.5 cm, respectively. On physical examination, both infants were severely pale and had respiratory distress. Weight, height and head circumference on admission were 3600 g, 52 cm and 37 cm for twin 1, respectively,

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