

Meconium Ileus (MI), Presentation of Cystic Fibrosis needs more Research?



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Editorial

Gastrointestinal (GI) tract gets affected earlier in the development stage of cystic fibrosis due to CFTR mutations [1]. CFTR gene is present all over the intestinal epithelial cells [2]. It controls secretion of chloride, bicarbonate and fluids. CFTR mutations result in abnormal electrolyte composition leading to abnormality in fluid secretions which alters the epithelial surface. This creates a dry luminal environment and bicarbonate deficiency in the proximal small intestine [3]. This can lead to terminal ileum obstruction, which when left untreated can result in rupture and sepsis known as meconium ileus (MI) in cystic fibrosis neonates. This can also present itself as distal intestinal obstructive syndrome (DIOS). The material which is mucofeculent, adheres to mucosal surface, giving appearance as bubbly-granular mass in the right lower quadrant on radiographs of the abdomen. It is found that less than 50% of patients that develop DIOS had MI as infants [4].

The studies done in twins provide evidence of the role of environmental and modifier genes in cystic fibrosis [5]. MI occurs in 20% of cystic fibrosis patients with severe mutation [6]. Meconium ileus (MI), distal intestinal obstruction syndrome (DIOS), and cystic fibrosis liver diseases are seen in 5 -20% of cystic fibrosis patients carrying severe class I -III mutations on both alleles. But no phenotype correlation could be established [7]. The role of modifier genes in MI and liver diseases due to cystic fibrosis is very important [8]. There are reports that some newborn infants with MI may not suffer from cystic fibrosis but these cases are very rare [9]. In these cases we must confirm diagnosis by other methods like sweat chloride tests, nasal potential difference sequencing before parental counseling.

Our small study of 29 infants who were referred as suspected cystic fibrosis with various Gastrointestinal (GI) tract manifestations. GI tract manifestations of cystic fibrosis include meconium ileus, intestinal obstruction, and abdominal distension. We had 5 (7.9%) cases of meconium ileus, out of them, 1 case presented with abdominal wall edema and erythema. We had 3 infants, with

complicated meconium ileus accompanied by abdominal wall erythema and edema, which had intestinal obstruction at birth requiring surgery. Intestinal obstruction in these 3 cases required surgery and out of which 1 case found to have atresia and 2 cases had perforations and meconium peritonitis as per operative findings. Ultrasono graphic findings include abdominal wall calcification in 3 cases and echogenic fetal bowel wall in 3 cases in antenatal period. Rizzo LC et al 2015 study reported meconium ileus (MI) in 8.62% to 16% cases (196). MI occurs in 7-10% of patients with cystic fibrosis. Infants with simple meconium ileus may present at birth with abdominal distension and they may not pass meconium. Meconium plug syndrome was reported in 1 case in our study. Some can present with bilious vomiting. We had 1 case reported with bilious vomiting and 5 cases with abdominal distension none of them showed F508del mutation. Probably they may be having large deletion or may not be the cases of CF. We need to do large studies with advanced techniques like sequencing or Multiplex ligation-dependent probe amplification (MLPA) for accurate diagnosis.

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