AUTHOR Parikka Vilhelmiina, Fihlman Mari, Raitio Arimatias

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Extremely viscous stool in a newborn leading to an early diagnosis of lifelong disease

A female infant, born at 37+3 gestational weeks after an uncomplicated pregnancy, was referred to level III NICU at the age of 24 hours due to a suspected bowel obstruction, with a history of not having passed meconium and some non-bilious emesis. Physical examination showed considerable abdominal distension with visible bowel loops. Abdominal X-ray identified dilated bowel loops, with no evidence of perforation (Figure 1).

Explorative laparotomy revealed significantly dilated loops of the ileum, with otherwise normal anatomy. The distal ileum was packed with extremely thick and sticky meconium, which could barely be removed through an enterotomy (Figure 2, Video). The bowel was emptied by milking and irrigation with saline, after which an ileostomy was performed. Meconium ileus and viscous meconium were suggestive of cystic fibrosis. Laboratory tests revealed a low faecal elastase level (<15 microg/l) and a high chloride level (133 mmol/l) in a sweat test. Two heterozygous mutations, F508del and CFTRdele2,3(21kb), were identified in genetic testing confirming the diagnosis of cystic fibrosis at the age of 19 days. Early diagnosis enabled an early introduction of pancreatic enzymes, as well as enhanced nutritional support. The bowel recovered well, and the stoma was closed after 7 weeks.

Meconium ileus is manifested in one fifth of the patients diagnosed with cystic fibrosis and is often the first manifestation of the disease.[1] Without screening, the median age at diagnosis can be more than 12 months.[2] Early diagnosis is associated with a better prognosis.[3] It is imperative that all infants with meconium ileus are tested for cystic fibrosis.

LEARNING POINTS/TAKE HOME MESSAGES (2-3 bullet points)

- Infants with cystic fibrosis have exceptionally thick and sticky meconium, which can lead to bowel obstruction called meconium ileus.
- Most of the infants with meconium ileus have cystic fibrosis, making it imperative to test all infants with meconium ileus for cystic fibrosis.
- Early diagnosis of cystic fibrosis allows introduction of multidisciplinary care and instigation of lifelong management and follow up, and is associated with a better prognosis.

1. Sathe M, Houwen R. Meconium ileus in Cystic Fibrosis. J Cyst Fibros. 2017;16 Suppl 2:S32-S39.
2. Pedersen MG, Højte C, Olesen HV, Pressler T, Skov M. Late diagnosis and poor nutrition in cystic fibrosis diagnosed before implementation of newborn screening. Acta Paediatr. 2019;(12):2241-2245.
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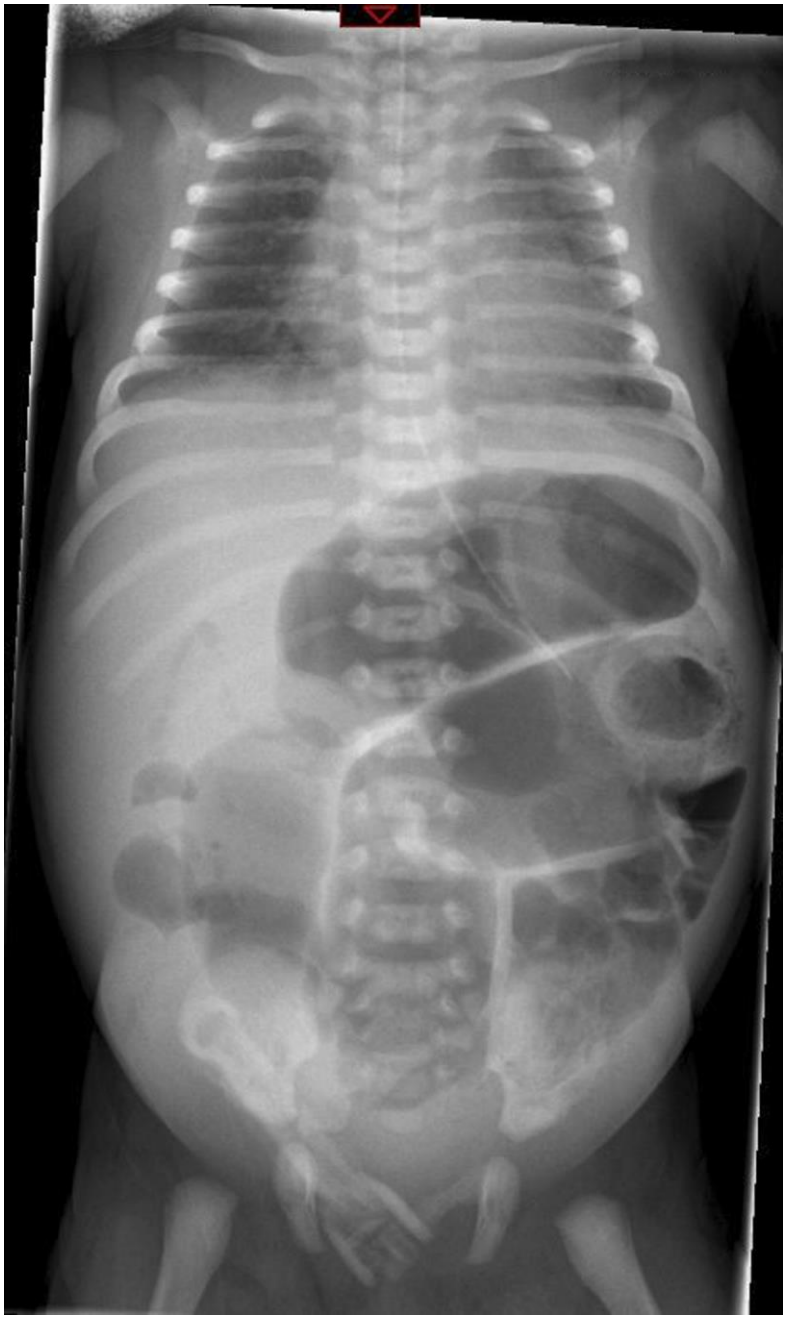

Figure 1. Long thorax x-ray showing distended bowel loops.

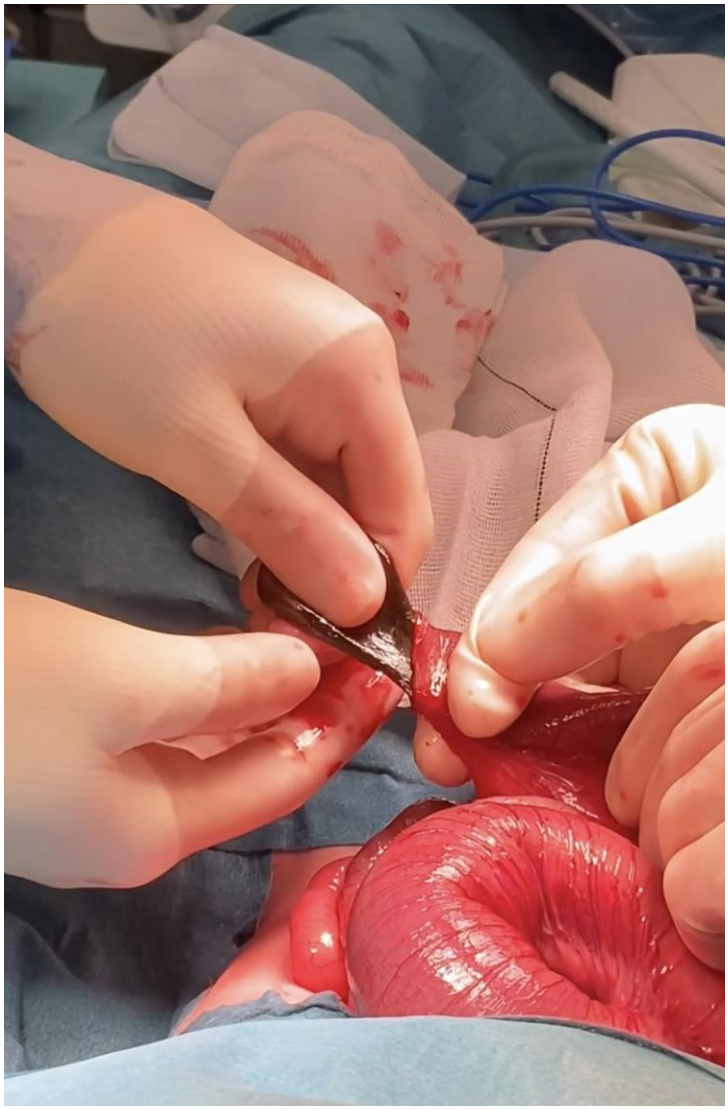

Figure 2. The distal ileum was packed with extremely thick meconium, which could barely be milked out through an enterotomy (see Supplemental video).

Supplemental video. Thick meconium was difficult to milk out through an enterotomy.