

# Intussusception in cystic fibrosis

M Holmes, V Murphy, M Taylor, B Denham

## Abstract

**Two cases of acute intussusception in older children with cystic fibrosis are reported. Both cases presented with symptoms and signs consistent with meconium ileus equivalent, which delayed the final diagnosis. Both cases required abdominal surgery but made full and uneventful recoveries.**

Intussusception occurring after the age of 2 years is relatively rare, but occurs in at least 1% of the population with cystic fibrosis.<sup>1</sup> We report two cases of intussusception occurring in patients with cystic fibrosis. A specific aetiology could not be found in either case.

## Case reports

### CASE 1

A boy was diagnosed as having cystic fibrosis at the age of 18 months by three positive sweat tests (his sweat sodium concentrations were 100, 100, and 113 mmol/l). He had been on pancreatic supplements since then with daily physiotherapy and regular courses of intravenous antibiotics. He had occasional meconium ileus equivalent, which had always settled promptly on treatment with 50 ml of oral Gastrografin (Schering, a mixture of sodium and meglumine diatrizoate). He was admitted to hospital age 10 years complaining of sudden onset of colicky abdominal pains and loose stools. Abdominal examination was normal. A plain radiograph of his abdomen showed appreciable retention of faeces consistent with meconium ileus equivalent. Gastrografin (50 ml) was given orally. The stools became more loose and more frequent over the subsequent three or four days but no relief was obtained. An abdominal ultrasound was performed which showed considerable constipation. A further 50 ml of oral Gastrografin was given. The next day the patient passed bright red blood rectally. A repeat plain radiograph of his abdomen (with the small bowel outlined by the Gastrografin) showed an obstruction at a point on the right side of the abdomen. A laparotomy was performed and an extensive ileocolic intussusception was found extending into the descending colon. The intussusception was reduced without resection. No underlying cause was found. Postoperative recovery was uneventful and the patient was discharged home two weeks later on normal diet and pancreatic supplements. There was no exacerbation of his chest condition during the illness.

### CASE 2

A boy was diagnosed as suffering from cystic

fibrosis at the age of 6 months by three positive sweat tests (his sweat sodium concentrations were 100, 105, and 108 mmol/l). He had been on pancreatic supplements with daily physiotherapy since then and regular courses of oral antibiotics for intercurrent infections. He had a history of intermittent bouts of abdominal pain with constipation. He was admitted to hospital at age 4 years complaining of colicky abdominal pains and constipation. Abdominal examination was normal. A clinical diagnosis of meconium ileus equivalent was made. Acetylcysteine was commenced (5 ml three times a day). The patient developed loose stools of increasing frequency over the subsequent three days, but otherwise showed no symptomatic improvement. A vague mass was identified in the left upper quadrant which was thought to be faeces. This theory was supported by a plain radiograph of his abdomen. Daily bowel washouts were commenced and the patient's condition improved sufficiently over the subsequent five days for him to be discharged from hospital. The next day he was readmitted with an occurrence of the original symptoms. Examination showed a distended abdomen but nothing else. A plain radiograph of his abdomen showed a complete absence of gas in the splenic flexure and descending colon but no dilated loops of bowel or fluid levels. Abdominal ultrasound showed echogenic masses throughout the colon that were thought to be due to a faecally loaded colon. Subsequent plain radiographs of his abdomen showed dilated loops of bowel and fluid levels. A laparotomy was performed because of the acute obstruction. An ileocaecal intussusception was found with rupture of the apex of the intussusception through the wall of the sigmoid colon. A resection of bowel was performed with ileocolic anastomosis. As the sigmoid colon at the site of rupture was of doubtful viability a colostomy was fashioned with a view to later repair and closure. This was done one month later. His recovery was uneventful. He was discharged on normal diet and pancreatic enzyme supplements and the problem has not recurred.

## Discussion

Acute intussusception is a common surgical problem in the first two years of life but is rarely observed in the older child.<sup>2</sup> It is a recognised complication in the older children who suffer from cystic fibrosis and its incidence has been put at 1% with an average age of onset between 9 and 12 years.<sup>1</sup> Despite this fact, it is a complication that is difficult to diagnose and is often overlooked.<sup>3</sup> It may present with identical symptoms to meconium ileus equivalent. Intussusception may not cause complete obstruction

The National Children's Hospital, Dublin

M Holmes  
V Murphy  
M Taylor  
B Denham

Correspondence to:  
Dr M Holmes,  
7 Pembroke Street,  
Dublin 4,  
Republic of Ireland.

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and although common presenting symptoms are colicky abdominal pains, vomiting and disturbance of bowel habit, absolute constipation, abdominal distention, and rectal bleeding are less commonly described.<sup>1</sup> Abdominal examination may be essentially normal apart from a 'faecal mass'. Bowel sound may be normal. Plain radiographs of the abdomen and abdominal ultrasound may not always be helpful in distinguishing intussusception from meconium ileus equivalent and a barium enema may be necessary. Some cases may not be diagnosed until laparotomy as in the two cases described here. Unlike intussusception in small children, hydrostatic reduction in older children is less successful and surgical reduction with or without resection is often necessary.<sup>1 2</sup> Apart from thick putty like material in the caecum (probably due to a combination of pancreatic enzyme deficiency and thick sticky mucus) no underlying cause is usually found.<sup>4</sup> It is thought that an inability of the intestine to propel the thick

putty like inspissated faeces along acts as the precipitating factor.

#### CONCLUSION

Although intussusception is a recognised complication of cystic fibrosis it is rarely reported. It can present in a similar way to meconium ileus equivalent. This may delay diagnosis. Where a presumptive case of meconium ileus equivalent fails to respond to Gastrografin or another appropriate osmotic agent, a diagnosis of intussusception should be considered.

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## Fatty infiltration in the liver in medium chain acyl CoA dehydrogenase deficiency

H C Losty, P Lee, M Alfaham, O P Gray, J V Leonard

#### Abstract

**Fatty infiltration of the liver at postmortem examination has been recommended as a criterion for selection of infants who have died suddenly and unexpectedly for further biochemical investigation for disorders of fatty acid oxidation. We describe a boy with medium chain acyl CoA dehydrogenase deficiency who died four months after diagnosis and in whom only minimal hepatic fatty infiltration was found.**

Medium chain acyl CoA dehydrogenase (MCAD) deficiency is an inherited disorder of fatty acid metabolism in which the ability to oxidise fat is impaired.<sup>1</sup> Patients often present within the first two years of life with hypoglycaemia, acute encephalopathy similar to Reye's syndrome and sudden, unexpected death. MCAD deficiency and the other rarer defects of fatty acid oxidation may be responsible for up to 5% of sudden, unexpected deaths in infancy (SIDS),<sup>2</sup> but it is not feasible to investigate all SIDS for fatty acid oxidation defects. As affected infants have had pronounced fatty infiltration of liver and muscle at postmortem examination, it has been recommended that this be used as a selection criteria.<sup>3</sup> We now report a patient with MCAD deficiency who died suddenly and unexpectedly four months after diagnosis. At postmortem examination only minimal fatty change in the liver was present.

#### Case report

This boy was the first child of unrelated parents, and he had been well until the age of 14 months when he presented with convulsions. For the previous 24 hours he had been febrile and had vomited several times. Several members of his family had diarrhoea and both *Giardia lamblia* and rotavirus were subsequently isolated from the patient's and the grandfather's stools. On admission he was unconscious with left sided focal seizures. He was hypoglycaemic (plasma glucose concentration 0.5 mmol/l) with a mild metabolic acidosis (pH 7.31, base deficit 10 mmol/l). Initially there was no hepatomegaly but during the next three days the liver became palpable 4 cm below the costal margin. His plasma ammonia concentration was 60 µmol/l. Although the aspartate transaminase activity was 204 IU/l (normal range up to 45 IU/l) and the alkaline phosphatase 1766 IU/l (normal range up to 340 IU/l), his alanine transaminase, γ-glutamyltransferase, bilirubin, and prothrombin time were normal. Electroencephalography demonstrated severe widespread abnormality with right sided focal features. He was treated with intravenous dextrose and diazepam but despite correction of the hypoglycaemia he had further convulsions over the next three days. He made a gradual recovery associated with a transient left sided weakness.

Analysis of organic acids in the urine collected on admission showed a hypoketotic

University Hospital  
of Wales,  
Heath Park,  
Cardiff CF4 4XW,  
Department of  
Medical Biochemistry  
H C Losty

Department of  
Child Health  
P Lee  
M Alfaham  
O P Gray

Institute of Child Health,  
London  
J V Leonard

Correspondence to:  
Dr Losty.

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