Prenatal bowel dilatation: congenital chloride diarrhoea

A healthy primigravida had moderate polyhydramnion (amniotic fluid index (AFI) = 230 mm) at 32 gestational weeks. The fetus had dilated, fluid filled, and frequently contracting small bowels filling the whole abdomen (figs 1 and 2), most likely representing distal small bowel atresia or stenosis. At 33 weeks, increasing polyhydramnion (AFI = 430 mm) led to the aspiration of 1180 ml of clear amniotic fluid not stained with meconium. At 36 weeks, the patient had an uneventful spontaneous delivery of a female infant with a slightly distended abdomen. As plain abdominal and contrast colon radiographs were normal and the baby was stable, oral feeding was introduced. No passage of meconium was observed. There was no vomiting, but at the age of 2 days she had loose watery stools and had developed hyperbilirubinaemia. She did not gain weight during the following 12 days and became increasingly tired. Her abdomen was distended with visible intestinal contractions (fig 3).

She had metabolic alkalosis and abnormal laboratory variables: serum Na⁺, 104 mmol/l; serum K⁺, 2.7 mmol/l; serum Cl⁻, 57 mmol/l. Further investigation showed a faecal concentration of Na⁺ (25 mmol/l), K⁺ (48 mmol/l), and Cl⁻ (46 mmol/l) changing to 103, 33, and 160 mmol/l respectively. Hypovolaemia and hyponatraemia led to increased renin production and hyperaldosteronism.

Normal radiographic findings, no vomiting, and the absence of meconium and normal stools led to the suspicion of congenital watery diarrhoea. The picture of preterm birth, polyhydramnion, no passage of meconium, abdominal distension, hyperbilirubinaemia, hyponatraemia, hypokalaemia, and hypochloraeic metabolic alkalosis made the diagnosis of congenital chloride diarrhoea highly probable, and this was confirmed by the high faecal chloride concentration. The high renin and aldosterone levels corresponded well to other documented observations.¹

There is no treatment to cure the diarrhoea, but children with this condition become toilet trained at a normal age, their social adjustment is not impaired, and they usually live a perfectly normal life.² Congenital chloride diarrhoea should be considered as a possible diagnosis in cases in which prenatal ultrasound examinations show distended bowels in combination with polyhydramnios.

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Arch Dis Child Fetal Neonatal Ed 2001 85: F65
doi: 10.1136/fn.85.1.F65

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