A newborn with watery diarrhoea

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A 35-year-old woman was referred to hospital at 32 weeks gestation because her uterus was large for date. Ultrasound examination done at that time is shown in the figure. At 34 weeks gestation, she gave birth to a baby boy weighing 2.2 kg, who had significant abdominal distension at birth. There was no vomiting. He did not pass meconium but after per rectum examination a large amount of light yellow watery stool was passed, followed by some relief of his abdominal distension. He continued to pass this type of stool. Blood laboratory tests at the age of 2 weeks showed a serum sodium of 131 mmol/l, potassium 3.1 mmol/l, chloride 91 mmol/l and bicarbonate 31 mmol/l. The family had an elder son who had had chronic diarrhoea since birth.

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Figure

Questions

- 1 What does the sonogram show?
- 2 What is the most likely diagnosis?
- 3 How can the diagnosis be confirmed?

Answers

QUESTION 1

Antenatal ultrasound at 32 weeks gestation showing marked dilatation of foetal intestinal loops and polyhydramnios.

QUESTION 2

The most likely diagnosis is congenital chloride diarrhoea (CCD). The diagnosis was suspected on the basis of maternal polyhydramnios, dilated foetal intestinal loops, premature delivery, absence of meconium, passage of large amount of watery stool since birth, blood tests showing hypochloraemic alkalosis, and the presence of a sibling with chronic diarrhoea since birth. The diagnosis was confirmed in this sibling to be CCD.

QUESTION 3

Confirmation of the diagnosis requires the measurement of both stool and urine electrolytes. In CCD there is a significant increased chloride concentration in stool (exceeding the sum of sodium and potassium) with no chloride in the urine.

Discussion

CCD is a rare autosomal recessive disorder with a worldwide occurrence, although most cases have been reported from Finland and the Middle East.¹⁻³ In certain areas in the Arabian Peninsula there is an incidence of 1:5500.³ Recently, the CCD gene has been accurately located on chromosome 7.⁴

In CCD, there is absence or impairment of active chloride/bicarbonate exchange in both the ileum and colon. The defective chloride absorption leads to osmotic diarrhoea. This malabsorption also explains the metabolic abnormalities seen in these patients.

The clinical presentation varies with the age of the child. During foetal life, significant maternal polyhydramnios occurs as a result of the intrauterine foetal diarrhoea. Antenatal ultrasound demonstrates significant dilatation of the foetal intestinal loops (figure).

Almost all patients with CCD are born prematurely, possibly as a result of the intrauterine diarrhoea. Postnatally, these children exhibit significant abdominal distension due to fluid accumulating in their intestine. There is no passage of meconium at birth and stools are profuse and watery. The diarrhoea may be passed unnoticed because the fluid is thought to be urine and this can cause delay in diagnosis. Some infants survive for several months without diagnosis. These children fail to grow and their developmental milestones are delayed. Any intercurrent infection, particularly gastroenteritis, can disturb further the delicate water and electrolyte balance and can lead to their demise. In the early neonatal period, the blood biochemistry is within normal limits with occasional acidosis, but after a few weeks, they show the characteristic laboratory finding of

Summary points

- CCD is rare inherited disorder mostly seen in communities with a high percentage of consanguineous marriages
- antenatal ultrasound by an experienced operator can suggest the diagnosis
- postnatally, differentiation from intestinal atresia is important to avoid unnecessary surgery
- the characteristic metabolic abnormality of hypochloraemic alkalosis is not present from birth but takes several weeks to develop
- life-long supplementation with sodium and potassium chloride solution is required to correct the metabolic abnormality, although this therapy will not change the character of the stool
- with early and adequate therapy these children can achieve normal growth and development

hypochloraemic hypokalaemic metabolic alkalosis. Other medical conditions can show similar metabolic abnormalities; especially important are cystic fibrosis and Bartter syndrome. The high faecal chloride (>100 mmol/l), exceeding the sum of faecal sodium and potassium and a chloride-free urine will confirm the diagnosis. The presence of other siblings with CCD, as in the case presented, will allow early identification of similar cases in the family.

Because of the antenatal ultrasonographic findings, the postnatal abdominal distension and the absence of passage of meconium, other surgical conditions may be considered before the correct diagnosis is made. These conditions include intestinal atresia, meconium ileus and aganglionosis. Some newborn infants with CCD are subjected to unnecessary laparotomies because of misdiagnosis as one of the above acute surgical conditions.⁵ The diagnosis of CCD cannot be verified by amniocentesis since the electrolyte concentration in amniotic fluid is normal.

Since the intestinal defect cannot be corrected, life-long treatment with both sodium and potassium chloride solutions is required. The rationale behind this therapy is the normal jejunal absorption of these electrolytes. Such treatment will not correct the diarrhoea but will prevent its secondary effects. Chronic hypovolaemia and hypokalaemia will result in renal tubular dysfunction in some of these children. Children with CCD are at increased risk of developing volvulus postnatally, a complication that may occur at any age from 2 days to 15 years.⁶ If the diagnosis is made early and treatment is given adequately, children with CCD can achieve normal physical growth and mental development.

Final diagnosis

Congenital chloride diarrhoea.

Keywords: chloride diarrhoea; newborn; diarrhoea; intestinal atresia

- 1 Holmberg C, Perheentupa J, Launiala K, Hallman N. Congenital chloride diarrhoea; clinical analysis of 21 Finnish patients. *Arch Dis Child* 1977;**52**:255–67.
- 2 Lubani MM, Doudin KI, Sharda DC, *et al*. Congenital chloride diarrhoea in Kuwaiti children. *Eur J Pediatr* 1989;**148**:333–6.
- 3 Kagalwalla AF. Congenital chloride diarrhoea; a study in arab children. *J Clin Gastroenterol* 1994;**19**:36–40.
- 4 Hoglund P, Sistonen P, Norio R, *et al*. Fine mapping of congenital chloride diarrhoea gene by linkage disequilibrium. *Am J Hum Genet* 1995;**57**:95–102.
- 5 Lundkvist K, Anneen G, Esscher T, *et al*. Surgical implication of congenital chloride diarrhoea. *Z Kinderchir* 1983;**38**:217–9.
- 6 Kirkinen P, Jouppila P. Prenatal ultrasonic findings in congenital chloride diarrhoea. *Prenat Diagnose* 1984;**4**:457–61.

An unusual cause of leg ulceration

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A 46-year-old man was referred with a 2-year history of non-healing ulcers on both lower limbs. He had suffered two episodes of deep vein thrombosis, at the ages of 7 and 28 years. He was subsequently admitted to hospital for treatment of pulmonary embolism at the age of 30. A year earlier, he had suffered an uncomplicated inferior wall myocardial infarction. On examination, he had multiple superficial ulcers on both his legs in the gaiter areas (figure). The surrounding skin showed signs of advanced venous insufficiency in the form of venous eczema, lipodermatosclerosis and pigmentation. Duplex imaging of his venous system revealed extensive bilateral post-phlebotic changes. His only medication was warfarin, which he had been advised to take life-long. His 18-year-old son was also diagnosed to have a deep vein thrombosis and subsequent investigations revealed two of his three children to be suffering from the same condition.

Questions

- 1 What is the underlying condition?
- 2 How is this condition managed?
- 3 How should the ulcers be treated?

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Figure The patient's legs