

Congenital Chloride Diarrhoea in a Malay Child

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Summary

Congenital chloride diarrhoea is a rare disorder mainly reported in Finland. A Malay child with congenital chloride diarrhoea presenting at six months of age with watery stools from birth and failure to thrive is reported.

Key words: Congenital chloride diarrhoea, watery stools, Malay child.

Introduction

Congenital chloride diarrhoea (CCD) is a rare disorder. It was first reported by Gamble *et al* and Darrow in 1945. It is an autosomal recessive disorder. To our knowledge the condition has not been described in a Malay child. Here we report our experience with a single case.

Case Report

S was referred to us at the age of six months for failure to thrive. He was born at term to a 32-year-old gravida 5 para 4 Malay lady with a birth weight of 2.5 kg. She had polyhydramnios during the pregnancy but no other medical problems. He developed jaundice at day five of life which resolved with phototherapy. He was only breast fed for the first 20 days of life. He had watery stools three to four times a day since birth. There was no blood or mucous in the stool. Even with the changing of feed to a soya formula and weaning diet his symptoms persisted. Since the age of three months there had been no weight gain despite diet manipulation.

His systemic review and developmental milestones were normal. Parents were second cousins. The elder siblings, two boys and a girl died at the age of three to four months. They had presented with failure to thrive, poor appetite and abdominal distension. No further information on the causes of death could be obtained.

On examination, his weight, length and head circumference were below the third centile. He was not dysmorphic. He was 10 per cent dehydrated. The abdomen was distended but the bowel sounds were normal. Other systems were normal.

Laboratory investigations showed a haemoglobin level of 9.9 gm/dl, a total white cell count of 11.7×10^9 (neutrophils 33%, eosinophils 2%, lymphocytes 63%, monocytes 2%) and a platelet count of $660 \times 10^9/L$.

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The blood urea was 2.9 mmol/L, sodium 122 mmol/L, potassium 2.2 mmol/L, chloride 77 mmol/L and creatinine 47 mmol/L. An arterial blood gas examination showed that the pH was 7.647, pCO₂ 4.91 kPa, base excess 18.2 mmol/L, bicarbonate ion 39.1 mmol/L and pCO₂ 13.36 kPa. A 24-hour urine sample showed sodium, potassium and chloride levels of 0.396, 1.76 and 0 mmol/L respectively. Stool sodium, potassium and chloride were 69, 42 and 143 mmol/L respectively. Serum aldosterone was 64.8 pg/ml and plasma renin was 15.9 ng/ml (Both levels were elevated).

He was started on electrolytes replacement consisting of potassium chloride and sodium chloride 1.5 gram three times a day. His electrolytes abnormalities were corrected with treatment. However his stools remained watery. His length and weight were below the third centile but his developmental milestones were normal.

Discussion

Congenital chloride diarrhoea has been discussed authoritatively by several authors. Reports of cases have mainly been from European countries particularly Finland. The condition has rarely been reported in Asians. To our knowledge no case involving a Malay child has been described.

Maternal hydramnios is a constant feature in CCD¹. It was documented in all Finnish patients and 24 of the non Finnish patients previously described in the literatures. The patient's mother had no antenatal ultrasounds done during the pregnancies. However there was no history of polyhydramnios. His three elder siblings presented with history of abdominal distension, vomiting, failure to thrive. They subsequently died.

The diagnosis was based on the clinical evidence of hydramnios, watery stools from birth, failure to thrive and biochemical abnormalities i.e. hypokalemia, hyponatremia, metabolic alkalosis, high stool chloride of more than 90 mmol/L (exceeding the sum of sodium and potassium) and the absence of chloride in the urine.

In most reported cases of CCD, the diagnosis was made at the mean age of 4-30 months. The basic feature of CCD is the high concentration of chloride in the watery stools. The median concentration of stool chloride is 150 mmol/L. After three months of age it will exceed the sum of concentration of sodium and potassium. As a rule the urine is free of chloride.

All these abnormalities will be corrected with adequate therapy. The severity of the electrolytes disturbance depends on the amount of salt intake.

However in a proportion of patients in whom the diagnosis was made late or the therapy was inadequate, a renal lesion with juxtaglomerular hyperplasia, hyalinised glomeruli calcium deposits and vascular changes resembling those in hypertension developed².

Ileal and rectal biopsies and absorption tests (including faecal fat and carbohydrate) done in patients with CCD have all given normal results. This indicates that the abnormalities in CCD are defects in the absorption of chloride, sodium, potassium and water.

It was confirmed by perfusion studies of the small intestine of one patient that the transport of the water and electrolytes was normal in the duodenum and jejunum. The transport of chloride, potassium and sodium was abnormal in the ileum and colon. These findings were abnormal with the impairment of the ileal-chloride-bicarbonate ion exchange. The reduced sodium absorption could

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be due to the abnormal acidity of the intestinal contents secondary to the decreased bicarbonate ion secretion³.

Untreated patients may survive with persistent diarrhoea, renal complications, psychomotor delay and mental retardation. The aim of therapy is to maintain a normal fluid and electrolytes status. Since the intestinal defects cannot be corrected, the only rational therapy would be the full and continuous replacement of diarrhoeal losses of water, sodium, potassium and chloride. This is more physiological and without side effects as compared to other modalities of treatment.

The total dosage of sodium chloride and potassium chloride must be adjusted to the minimum maintaining the normal blood pH. The ratio of sodium and potassium should be adjusted so as to maintain the serum of both ions at about the middle range of normal. To our present knowledge all attempts to stop the diarrhoea is futile and also dangerous. Patients with CCD will make their social adjustments adequately.

In conclusion, CCD is a rare cause of chronic diarrhoea in children. However it should be considered in all children whose symptoms present early in life.

References

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