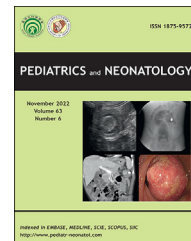


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Images

Congenital chloride diarrhea presenting as dilated fetal bowel loops

Pallabi Hui^a, Shruti Kanjilal^a, Tamoghna Biswas^a,
Atanu Kumar Dutta^{c,*}, Debojyoti Bhattacharjee^b,
Mrinal Kanti Das^a

^a Department of Pediatrics, IPGME&R and SSKM Hospital, Kolkata, India

^b Department of Biochemistry, IPGME&R and SSKM Hospital, Kolkata, India

^c Department of Biochemistry, AIIMS Kalyani, India

Received May 28, 2022; received in revised form Jun 15, 2022; accepted Jun 29, 2022

Available online 31 July 2022

A ten-month-old female, born out of second-degree consanguineous marriage, presented with complaints of 15–20 daily episodes of watery diarrhea since birth associated with poor weight gain and delayed motor developmental milestones. Antenatal ultrasound at 34-week 3 days gestation had revealed maternal polyhydramnios (Amniotic Fluid Index 223 mm, >90th centile), distended fetal abdomen (abdominal circumference 364 mm; >97th centile), generalized dilatation of fetal bowel loops (Fig. 1a) and increased estimated fetal weight (3044 g, >97th centile). The baby was delivered by Cesarean section at a gestational age of 36 weeks with birthweight of 2.4 kg. After birth, the child had voluminous watery stool, abdominal distension with visible peristalsis (Fig. 1b). Then onwards she was treated for watery diarrhea without much improvement. Her motor developmental milestones were delayed but other fields were normal. On admission the baby was lethargic with features of some dehydration and marked abdominal distension (Fig. 1c) with perianal excoriation. Laboratory investigations showed hypochloremia, hypokalemia, hyponatremia and metabolic alkalosis. The

stool pH was acidic with no reducing sugar. Spot urinalysis showed low chloride level of 11 mmol/L. After correction of serum electrolytes, chloride level in stool was estimated and found to be 133.9 mmol/L. The typical clinical picture of congenital secretory diarrhea with acidic stool, metabolic alkalosis and high fecal chloride levels, exceeding 90 mmol/L was suggestive of Congenital Chloride Diarrhea.^{1,2} Clinical Exome Sequencing revealed a novel Likely Pathogenic homozygous missense variant in *SLC26A3* gene [NM_000111.3: c.250G > A (p. Gly84Arg) (hg38)] thereby confirming the diagnosis. Treatment was started with correction of dehydration and dyselectrolytemia with appropriate fluid, sodium and potassium chloride supplementation as per recommendation in the published literature including avoidance of Ringers Lactate and standard Oral Rehydration Solution.³ At discharge, the child was active, playful, tolerating the oral salt substitution therapy and solid foods (Fig. 1d). She was on regular follow up with monitoring of serum electrolytes and urinary chloride. She achieved age-appropriate weight and developmental milestones with loose stool frequency of 5–6/day at 2 years of

* Corresponding author. Department of Biochemistry, Second Floor, Medical College Block, All India Institute of Medical Sciences, NH - 34 Connector Basantapur, Saguna, Kalyani, 741245. West Bengal, India.

E-mail address: atanu.dutta05@gmail.com (A.K. Dutta).



Figure 1 a) Antenatal ultrasound examination performed at 34 weeks of gestation revealing generalized dilated bowel loops on sagittal (SAG) and transverse (TS) views; b) Abdominal distension during infancy and c) childhood; d) resolved abdominal distension after treatment.

age. This report highlights the need to consider congenital chloride diarrhea as a diagnostic possibility with favorable prognosis in cases of generalized dilated fetal bowel loops and rectum combined with polyhydramnios with normal skeletal and urinary system morphology.

Declaration of competing interest

The authors have no conflicts of interest relevant to this article.

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