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Congenital Chloride Diarrhoea

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Abstract

A case of congenital chloride diarrhea in a patient with recurrent polyhydramnios was suspected antenatally, confirmed postnatally and managed successfully.

Introduction

Congenital chloride diarrhea is a rare autosomal recessive disorder caused by abnormality in the active transport of chloride from the distal ileum and colon. It is characterized by hypokalemia, hypochloremia, hyponatremia, metabolic alkalosis and dehydration in the newborn.

Case Report

A 26 year old, gravida 5, para 4, living 2, intrauterine fetal death 1, neonatal death 1, second degree consanguineous marriage, was referred with an ultrasound (USG) findings of polyhydramnios with dilated fetal bowel loops at 33 weeks of gestation in preterm labor. Her first pregnancy was an intrauterine fetal demise at 7 months of gestation with history of polyhydramnios. Her second pregnancy also had history of polyhydramnios with neonatal death at 2 months. Her third and fourth pregnancies had been uneventful normal deliveries with both fetuses alive and well. Her antenatal investigations including plasma sugar levels were within normal limits. She had received steroid therapy to cause fetal lung maturation. USG suggested fetus with cephalic presentation with a composite gestational age of 33 weeks, an estimated weight of 2.1 kg with amniotic fluid index of 28 cm and dilated bowel loops with peristalsis without fetal ascites, intraperitoneal calcifications and fetal structural anomalies. Due to cord presentation with fetal distress the woman was taken up for lower segment cesarean section. She delivered a female child of 2.18 kg with an APGAR at 1 & 5 minutes of 7/10 & 8/10 respectively. After birth neonate had a distended abdomen with visible peristalsis and watery diarrhoea (figure 1) and was immediately transferred to the neonatal intensive care unit. Hemogram was normal. Stool chloride level was increased (90 mmol/l) [normal range 6-17 mmol/l]. Polyuria and diarrhoea persisted and an USG of the abdomen on day 2 of life revealed dilated bowel loops with normal peristalsis. Serum electrolytes revealed hyponatremia, hypocalcemia, hypokalemia, and hypochloremia. Twenty four hour urinary levels of sodium, potassium, calcium and

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Rare serious side effect of low dose Ondansetron: ...

Labial Agglutination In A Postmenopausal Female

Pemphigoid Gestationis

Uncorrected Double Outlet Right Ventricle with Fal...

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► May (12)

► April (12)

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creatinine were corresponding to the serum levels. Serum renin levels were markedly increased (> 500 IU/ml). Hence a diagnosis of congenital chloride diarrhoea was established. The neonate gradually started accepting feeds well and electrolyte imbalance was corrected with 3% NaCl, calcium gluconate and KCl respectively along with maintenance fluids and broad spectrum antibiotics for 21 days. The neonate was discharged on day 26 of life.

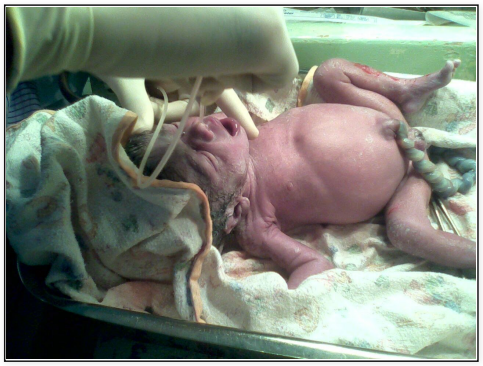

Figure1. Distended abdomen of the neonate at birth

Discussion

Congenital chloride diarrhea was first described in 1945 and is also known as Darrow Gamble disease.[1] Around 250 cases have been reported so far, mainly in Saudi Arabia, Finland and Poland. Congenital chloride diarrhea is a rare autosomal recessive disorder affecting both the sexes caused by mutation in the solute carrier family 26 member 3 genes mapped to chromosome 7. The pathogenic feature is defect in the chloride/bicarbonate channels affecting the distal ileum and colon resulting in fecal chloride loss and osmotic diarrhea which if untreated leads to dehydration and death.[2] Sodium and potassium in stool is not abnormally high but profuse diarrhea leads to absolute loss manifesting as hyponatremia and hypokalemia. Excessive loss of H+ through the kidneys leads to alkalosis. Hyperaldosteronism occurs as a compensatory mechanism to conserve sodium. 'Urine like diarrhea' leads to polyhydramnios which in turn causes preterm delivery.[3] Antenatal ultrasound features include dilated bowel loops with polyhydramnios without fetal ascites, intraperitoneal calcifications and fetal structural anomalies. Hence a differential diagnosis of jejunal and ileal obstruction (increased peristalsis), meconium peritonitis (ascites and intraperitoneal calcification) and cystic fibrosis (hyperechoic bowel loops) can be ruled out on USG.[4,6] Amniocentesis may show increased levels of alpha fetoprotein, bilirubin and chloride in the amniotic fluid but these are not diagnostic.[1,5] Most children become toilet trained at a normal age, social adjustment is not impaired and they can live a perfectly normal life.[7] The long term prognosis is generally favorable but complications like renal disease, inflammatory bowel disease, hyperuricemia, inguinal hernias, spermatoceles and male subfertility are possible.[2,8] Hence we conclude that careful evaluation of the differential diagnosis on ultrasound can help point towards such life threatening rare neonatal disorders. Uncommon causes of recurrent pregnancy losses can also be elucidated by offering the parents genetic analysis.

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Citation

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