mosaicisms are reported. The risk of recurrence due to germline mosaicism cannot be quantified, however indicates the need for prenatal diagnosis. Hence, it will be quite useful for relieving the anxiety of the couple with a previous affected child. The technique has been used earlier for prenatal diagnosis of the condition when sonographic features suggested the diagnosis of this particular condition(3).

Contributors: KMG was involved in clinical evaluation of patients, carried out molecular work and drafted the manuscript. SRP conceptualized the work and was involved in clinical evaluation of patients and correction of manuscript. FK was involved in DNA sequencing and contributed to drafting of manuscript. SA guided the molecular work and DNA sequencing. She also contributed to the correction of the manuscript. She will act as guarantor.

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Neonatal Bartter Syndrome

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Bartter syndrome is an inherited renal tubular disorder with hypokolemia, hypochloremic metabolic alkalosis, normal blood pressure with hyperreninemia and increased urinary loss of sodium, potassium and chloride. We report an infant with neonatal Bartter syndrome, who improved with potassium supplements.

Key words: *Bartter syndrome, Neonate.*

Bartter syndrome is a renal tubular disorder characterized by hypokalemia, hypochloremic metabolic alkalosis, normal blood pressure with hyperreninemia and increased urinary loss of sodium, potassium and chloride. Usual clinical manifestations are failure to thrive, polyuria and episodes of dehydration. A 70-day-old boy, born to non-consanguineous parents presented with failure to thrive, persistent hypokalemia,

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hypochloremic metabolic alkalosis with normal blood pressure, elevated serum renin levels and was diagnosed as Bartter syndrome.

Case Report

A 70-day-old boy, born to a non-consanguineous parents with a normal elder sibling and uneventful antenatal period, was delivered by cesarean section. There was history of maternal polyhydramnios. The baby weighed 2.5 kg and was on breast feeds, and apparently normal upto 20 days of life. He then developed frequent loose stools, non-bilious vomiting, abdominal distension and failure to thrive. As the investigations done elsewhere revealed persistent hypokalemia even after the control of diarrhea, the infant was referred for evaluation. There was no history of administration of aminoglycosides.

On examination, the child weighed 2.6 kg and was poorly nourished with length of 58 cm and head circumference of 38 cm (which was 10th percentile for the age). He was dehydrated with no facial dysmorphism. He was comfortable, normotensive and had no localizing signs on neurological examination. Investigations showed hypokalemic hypochloremic metabolic alkalosis (serum potassium 2.1 mEq/L; chloride 85 mEq/L; bicarbonate 33 mEq/L; pH 7.49; and PCO_2 40 mm), normal serum levels of creatinine, calcium and magnesium with urine revealing hyposthenuria (specific gravity 1.007) and increased urinary loss of chloride, potassium and calcium (chloride 45 mEq/L; potassium 40 mEq/L; calcium to creatinine ratio –0.38). Urine output was 4-5 mL/kg/hour. Ultrasonogram of the abdomen revealed early features of nephrocalcinosis in both the kidneys. Elevated plasma level of renin of 40 U/L in supine position (normal 4-8 U/L) was found and a diagnosis of Bartter syndrome was made. Child was maintained on supportive care, potassium supplements and dietary advice, with clinical and biochemical improvement. Blood values at 77 days of age were sodium 138 mEq/L; potassium 3.6 mEq/L; chloride 98 mEq/L; bicarbonate 28 mEq/L; pH 7.45; and PCO_2 42 mm. It is planned to initiate treatment with indomethacin at 3 months of age.

Discussion

Bartter syndrome is an inherited renal tubular disorder characterized by hypokalemia, hypochloremic metabolic alkalosis, hyperreninemia, hyper-prostaglandinism, normal blood pressure, with increased urinary loss of sodium, chloride, potassium, calcium and prostaglandins(1). The onset may be during the neonatal period, infancy or childhood. Antenatal features include polyhydramnios and premature delivery. Polyuria is seen later in life also. Other clinical features are failure to thrive, characteristic facies with thin, triangular face, prominent forehead, large eyes, protruding ears, drooping mouth, strabismus, sensorineural deafness, convulsions and increased susceptibility to infections.

The inheritance of Bartter syndrome is autosomal recessive with 3 subtypes. The neonatal type is characterized by fever, diarrhea, vomiting, osteopenia and elevated urinary excretion of prostaglandin E. In neonatal Bartter syndrome type 1, frame shift, non-sense and mis-sense mutations occur in the gene located on chromosome 15 q15-q21 encoding for bumetanide sensitive Na-K-2Cl cotransporter (NKCC2) of thick ascending loop of Henle(2). Mutations in ROMK gene located on chromosome 11q24-25 encoding adenosine triphosphate (ATP) sensitive K^+ channels, that recycles reabsorbed K back into tubular lumen, results in neonatal form of Bartter syndrome type 2(3). In the third type of

neonatal Bartter syndrome, there is associated sensorineural deafness and early chronic renal insufficiency due to mutations in gene BSND localised to chromosome 1p31, encoding for Barttin which is expressed in the ascending limb of Henle and inner ear(4,5).

The neonatal form differs from the classic Bartter syndrome by the age of onset, presence of nephrocalcinosis and very high urinary loss of sodium, calcium and chloride. Other differential diagnoses are Gitelman's syndrome (characterized by hypomagnesemia, hypocalciuria), pseudohyperaldosteronism (hypertension with no evidence of increased secretion of mineralocorticoids) and pseudo-Bartter syndrome due to administration of high doses of prostaglandin E1(6).

Prenatal diagnosis can be made by the high chloride content of the amniotic fluid and mutational analysis of genomic DNA extracted from cultured amniocytes obtained by amniocentesis(7). Therapeutic efforts should be directed to correct dehydration and electrolytic imbalance. Apart from potassium supplementation, administration of indomethacin after 6-12 weeks of life is useful. Indomethacin at a dose of 1-5 mg/kg/day is most frequently used and well tolerated(8). Other drugs used are acetylsalicylic acid (100 mg/kg/day), ibuprofen (30 mg/kg/day) or ketoprofen (20 mg/kg/day). Addition of potassium sparing diuretics may be initially effective in the control of hypokalemia but their effect is transient.

The long-term prognosis is guarded; lack of satisfactory control may lead to morbidity, growth failure and renal insufficiency.

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