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SCIENTIFIC LETTER

Nephrogenic Diabetes Insipidus in a 10-mo-old Infant: Complication of Nephropathic Cystinosis

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To the Editor: A 10-mo-old infant presented to outpatient department with chief complaints of failure to gain weight and length for 4 mo, on and off fever for 4 mo, polydipsia & polyuria for 4 mo, and nausea & vomiting for 10 d. There was a history of sibling death with same type of problem at 18 mo of age. There was one still birth at 7 mo of gestation. Development milestones were achieved at normal age. Child was alert, active and recognizing parents. Signs of some dehydration were present. There was moderate stunting and severe wasting. Her measured urine output was 20 ml/kg/h. Rest of the systemic examination and eyes were normal. The child subsequently had three admissions in follow up. Each episode of admission had hypernatremia and dehydration with acute kidney injury. This made us suspect nephrogenic diabetes insipidus (NDI) in the child.

Investigations were suggestive of proximal renal tubular acidosis (RTA). Paired serum and urine osmolality showed no rise in urine osmolality after vasopressin challenge – confirming NDI. Ultrasonography of the abdomen showed normal size kidneys and no feature of nephrocalcinosis. Radiographs of bilateral wrist showed rickets. Clinical exome showed recurrent pathogenic missense point variation [c.922G>A (p.Gly308Arg)] in exon 11 in *CTNS* gene. G308R affects the Transmembrane membrane 6 (TM6) of cystinosin protein [1]. This has not been reported from India till date. Largest Indian cohort of nineteen patients has found 8 variants in 12 patients from 11 families. Most common Indian variant: Exons 3; c.16_19delCTGA; p.Thr7PhefsTer7 (Frame shift mutation) has been reported in five unrelated patients [2]. Nephropathic diabetes insipidus was detected in subsequent follow up visits. NDI had been previously reported only few number of times as a complication of Nephropathic cystinosis (NC) [3]. This has been previously reported by Das et al. in an Indian patient [4]. This complication required treatment with hydrochlorothiazide and the child showed response to the treatment.

Declarations

Conflict of Interest None.

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